

Public Perspectives on Genetic Discrimination

September 2004 - November 2004

**Secretary's Advisory Committee
on Genetics, Health, and Society**



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I. COMMENTS FROM MEMBERS OF THE GENERAL PUBLIC

Comments from the October 2004 SACGHS Meeting

Heidi Williams, Kentucky.....	2
Phaedra Malatek, Illinois.....	5
Rebecca L. Fisher, Virginia.....	9
Tonia Phillips, Virginia.....	14
Paula Funk, Arkansas.....	16
Carolina Hinestrosa, Washington, DC.....	20
Phil Hardt, Arizona.....	23

Written Comments

Anonymous.....	38
Celia A. Boyne-Schuh.....	39
Mary A. Brown.....	41
Lynda Carrion.....	42
Jay Daniels.....	43
Laila Gillam.....	44
Joeline Hollar (Hardt).....	45
Pam Kennedy.....	47
Terence McCarty.....	48
Sandra McElwee.....	49
Beth Pletcher.....	50
Sue Saladino.....	51
Stephanie Schmidt.....	53
Renee Shean.....	54
The Honorable Louise M. Slaughter.....	56
Jennifer Stephens.....	62
Kari Stubbs.....	63
Michelle Thompson.....	65
Ruth Yong.....	68

II. COMMENTS FROM HEALTHCARE PROVIDERS

Comments from the October 2004 SACGHS Meeting

Jeff Shaw, M.S.	73
Don Hadley, M.S., CGC.....	78
Mark Brantly, M.D.	82

Written Comments

Lorraine Dugoff.....	85
Sandra J. Dunbrasky, M.D., FAAP.....	86
Michael Farrell, M.D., MCW.....	87
Theresa Aly.....	88
Eric Fowler, M.S., CGC.....	89
Generosa Grana, M.D. and Dana Farengo Clark, M.S.	90
Susan V. Montgomery, R.N.	91
Robbin Palmer, Ph.D.	92
John Quillin, M.S., M.P.H.	93
Allen W. Root, M.D.	94
Rae S. Wruble, R.N., M.B.A.	99

III. COMMENTS FROM INSTITUTIONS AND PROFESSIONAL ORGANIZATIONS

Comments from the October 2004 SACGHS Meeting

Genetics and Public Policy Center	101
Rutgers School of Law and Licata & Tyrrell P.C.	105
America's Health Insurance Plans	109
Society for Human Resource Management	118

Written Comments

Alpha-1 Association and Alpha-1 Foundation	128
American Association for the Advancement of Science	129
American Association of Clinical Endocrinologists	131
American Board of Genetic Counseling	134
American Cancer Society	137
American Psychiatric Association.....	147
The Arc and United Cerebral Palsy.....	149
Council for Responsible Genetics	153
Genetic Alliance.....	157
Humana Inc.	199
LabCorp.....	202
Myriad Genetic Laboratories, Inc.	205
National Breast Cancer Coalition	210
National Partnership for Women & Families.....	212
National Society of Genetic Counselors	214
Supporting Those At Risk	217
Society of General Internal Medicine	218

IV. RESEARCH LITERATURE REFERENCED IN COMMENTS

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Secretary's Advisory Committee on Genetics, Health, and Society Public Perspectives on Genetic Discrimination

Preface

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) was established in 2002 to serve as a public forum to explore the broad range of health and societal issues raised by the development and use, as well as potential misuse, of genetic technologies and to make recommendations to the Department of Health and Human Services as needed. The potential for the misuse of genetic information in health insurance and employment is a high-priority issue for SACGHS, and the Committee made recommendations in 2003 and 2004 about the need for Federal legislation in this area.*

In the fall of 2004, the Committee sought to learn more about the scope and nature of genetic discrimination based on predictive or predispositional genetic information or carrier status by gathering information about cases of such genetic discrimination. The Committee solicited input from members of the general public and from health professionals with patients who have experienced or were concerned about genetic discrimination in health insurance or employment or who have taken steps to protect their genetic information. The Committee reached out to the public via the Federal Register and announcements on the SACGHS website and listserv, and by holding a meeting to hear directly from members of the general public, health care professionals, and other stakeholders about their experiences and perspectives on the issue.

This compendium of public perspectives includes all of the comments SACGHS received between September and November 2004 and during the Committee's October 2004 meeting. Parts I through III consist of oral and written comments from, respectively, the general public, health care professionals, and institutions and professional organizations. Part IV consists of scholarly articles referenced in the comments.

SACGHS appreciates all of the comments that were submitted and wishes to thank those who shared their personal and professional perspectives with the Committee on this important issue.

*Further information about SACGHS and its meetings and recommendations is available at <http://www4.od.nih.gov/oba/SACGHS.HTM>

Comments from the October 2004 SACGHS Meeting

Members of the Committee,

My name is Heidi Williams and my children, Jayme, 8, and Jesse, 10, were recently victims of genetic discrimination. In August of 2003, I saw a commercial on television advertising affordable health care insurance for individuals through Humana, Inc. I called the toll free number and talked with a young woman who quoted me a price for a policy that would cover both of my children. I was told that the monthly cost to insure my children would be approximately \$105.00, and I immediately told the young woman I would like to complete an application so that the coverage would begin as soon as possible. I was asked a series of questions about my children, including whether or not they had a preexisting condition. I relayed to the young woman, under the threat of a fine and incarceration for falsifying information, the fact that my children were carriers of the genetic disorder called Alpha-1 Antitrypsin Deficiency (AAT), a liver deficiency that can progressively effect the lungs, the liver, or both, but that my children, unlike their mother who is lung symptomatic, would never suffer from any aspect of the disorder. The young woman, who wasn't quite sure what to do with this information, asked me to hold on while she contacted her supervisor. As I spoke with her supervisor, I, again, explained how my children were only carriers of the AAT gene and that my children, themselves, would never suffer from any aspect of the disorder, as I am suffering, and that they are exceptionally healthy and active children. Again, I was told to hold on the line because, as this gentleman was uncomfortable with the information I had imparted, he needed to contact his supervisor.

As I spoke to the senior supervisor, I once again relayed the information about Alpha-1, and how my children were only carriers. To be born what is considered symptomatic, you must have two parents who are at least carriers of the AAT gene and be of a certain phenotype. I am what is considered the "symptomatic" phenotype of ZZ and my husband is considered to be of a "normal" phenotype; therefore, my children can only be carriers and, as research supports, will never be susceptible to the various problems symptomatic AAT's face, including lung and liver failure.

Once the senior supervisor and I finished speaking, I was given back to the young woman who initially interviewed me, and, after finalizing the application, was told by her that I would receive a reply to my children's application for health insurance within 24 hours. After five days of waiting, I knew, instinctively, that there had been a problem with my children's application. I received a letter two days later, exactly one week after the phone application, stating that my children were being rejected for health care insurance through Humana, Inc. due to their AAT status and for no other reason.

After much self-recrimination, I shared my woes with the Alpha-1 Lungs and Life Chat group, relating my frustrations and my fears from my children having been rejected for health insurance coverage. Nancye Buelow, a representative of the Genetic Alliance at that time, heard about my problems with Humana, Inc., and approached me about publicly coming forward with my story through the auspices of the Genetic Alliance. I agreed, and together with the Genetic Alliance, and the help of a prestigious Washington, DC law firm, and a wonderful and very knowledgeable AAT doctor, an appeal to the August 2003 letter, a letter which rejected my children for health insurance coverage on the basis of their genetic status, was drafted and sent to Humana, Inc.

Enclosed within the letter to Humana, Inc. was research information from both the National Institutes of Health and the Alpha-1 Foundation supporting my argument that both Jesse and Jayme, as carriers, would not become symptomatic of Alpha-1 Antitrypsin Deficiency, and that both would remain free of AAT's debilitating destructiveness throughout their lifetime.

In February of this year, I received my response to the written appeal, and was once again shocked to read that my children were being rejected for health insurance coverage only on the basis of their AAT carrier status, and nothing more. It was only after Humana, Inc. had been approached by a reporter for a well-known and well-respected newspaper, that they reversed their decision and offered my children full coverage under their company, prorated from August 2003, and paid in full until April 2004 by Humana, Inc., themselves. Fortunately for me, my children are now covered by a company that understands everyone is entitled to affordable health care coverage, and not Humana, Inc.

Needless to say, Humana, Inc.'s reversal of decision felt like a hollow victory. No one should have to force an insurance company to cover perfectly healthy children. In fact, I don't believe it should have mattered what their genetic status was to begin with. We are all viable members of a community with contributions to make and shouldn't have to be afraid that our genetic anomalies, in whatever form they arise, will be held against us. I should not have had to spend the better part of six months wondering if the decision to have my children's genetic status verified by their pediatrician was a huge mistake. I should not have to wonder if my children's genetic status is going to follow them into the workforce and render them unable to become employed in their chosen fields. And, I certainly should not have to feel guilty for unknowingly passing this genetic anomaly on to my children. Humana, Inc. made me feel guilty and ashamed for needing to know my children's genetic status. Furthermore, they made me feel guilty for needing a parent's peace of mind in regard to my children's future health, and, for that, I am angry.

Today, there is a current of fear reverberating throughout the genetic community. It is not just a fear of loss, but it is a fear of retribution. It is a fear that forces many within this particular community to accept what should be unacceptable; discrimination by genetic status. Many people are afraid to come forward and fight for their rights to employment and health insurance coverage because they are afraid of the retribution that may not only be taken against them, but could be taken against their families, as well. Therefore, it is because of the callous treatment of my children and the countless others before them that I want to make sure that this sort of "policy practice" never happens to anyone ever again. I want to make sure that I will never again exchange emails with someone who has been fazed out of a position due to her genetic status. I never again want to hear the story of someone who has been denied health insurance coverage, had their health insurance coverage cancelled, been passed over for promotion, demoted, fired, or simply not hired due to their genetic status. National legislation that would make it illegal for insurance companies and employers to use someone's genetic status against them has indeed been drafted. The Senate passed S1053, the Genetic Information Nondiscrimination Act, unanimously last October, yet one year later, this very important piece of legislation that would protect many Americans, is still stranded in the House of Representatives. As each day passes, and the genetic community waits for the House to bring this bill to a vote, scores of people across this Nation are being persecuted on the basis of their genetic status. It is

completely reprehensible that any type of discrimination still exists and has to be legislated against in this day and age. But, since discrimination still exists, it must be swiftly eradicated in any form that it is found, before its destructive force has had the chance to harm anyone else. Finally, my family and I were extremely lucky. We had the backing of several people and organizations to help us fight our battle in the war against genetic discrimination that very few people in the genetic community win. Only through legislation and education will genetic discrimination loosen its hold on a community of people who are suffering from its devastating effects.

Thank you.

Testimony of Phaedra Malatek
Secretary's Advisory Committee on Genetics, Health, and Society
October 18, 2004

Goodmorning Chairman Tuckson, and members of the committee. My name is Phaedra Malatek. I am a wife, mother, daughter, sister, and friend of people who care greatly about what we are doing here today. I currently live in Aurora, Illinois with my husband and two sons. My primary occupation is as adjunct faculty at two local community colleges. I am otherwise involved in my community and nationally working on issues related to women's cancers. But today I am here to talk to you about the hope that lies in genetic testing and more specifically the Genetic Information Nondiscrimination Act (S. 1053). Because as Ralph Waldo Emerson says, "We judge of man's wisdom by his hope," and today I am feeling pretty wise

For me, genetic testing and the protection offered by the S. 1053 can be compared in an analogy to weather tracking or storm prediction. Imagine if you will, that we had no knowledge or ability to predict the hurricanes that recently swept through the southeastern United States and Caribbean. How would the death toll change? How would the damage assessments change? And how would the insurance industry change? Now imagine never having any information on any storm ever. Well, I think that our understanding and consideration of genetic testing can be equated to the "What ifs...." What if people were given knowledge of potential storms in their lives? How would they be able to protect themselves? What would serve as the plywood for their windows? What evacuation route could be made available to them? And more importantly how many lives would be saved? That really is the question isn't it: How many lives can we save by what we do here today and through the enactment of S. 1053?

Continuing this storm analogy and the concept of discrimination, let's consider the situation where you know a storm is coming and are able to take precautions, such as boarding up the windows and putting the lawn furniture in the pool, but you can't get insurance for your property, simply because you know that a storm *may* come some day. Your neighbor on the other hand has no advance warning, but has a full measure of insurance. If the storm doesn't hit, that is all fine and good for you and your neighbor. Everyone wins.

However, if the storm does hit, the insurance company provides financial reimbursement to you neighbor's severely damaged if not destroyed property, but there is no coverage for your property no matter how large or small the damage. It seems to me that this is what we are talking about with genetic discrimination, a situation in which no one wins, and everyone loses. Those with advanced warning are actually harmed rather than helped by the knowledge they possess. And insurance companies and those without advanced knowledge suffer larger losses than had they been given the knowledge to protect themselves.

If fair warning is given to all parties, through genetic testing, the people who are able to protect themselves, and the insurance company who agree to be at risk for any loss suffered, all have a much larger measure of protection. Those who are informed about their risks, can be proactive and take either prophylactic measures or be monitored more closely, increasing their ability to entirely avoid developing a disease or having it detected in its earliest, most treatable and survivable stage. S.1053 offers even more protection when it eliminates the chances of discrimination compounding the effects of the potential storm caused by undetected diseases. Thus, the storms, or illnesses would cause that much less damage. And as we all know, this not only saves lives, but also dramatically decreases the cost to employers, insurers, and the individual.

A storm such as this hit my life through my father a number of years ago. In 1991, my father gathered together his children, siblings, cousins, and nieces and nephews to discuss a genetic illness he had been diagnosed with. The disease is called hemochromatosis, which is often referred to as iron overload. My father had been exhibiting symptoms including arthritis, heart arrhythmia, as well as a change in skin tone prior to diagnosis. At the time he was diagnosed, his liver and heart were fully involved. At the same time, his physicians conjectured that my grandfather and great-grandfather may also have carried and suffered the effects of this disease. Within a year of our family meeting he suffered a heart attack and within 10 years he died from complications of the disease. Since my father's diagnosis, two of my siblings developed complications of the disease. None of them, my father or siblings have had genetic tests for hemochromatosis.

Because of my family's history with hemochromatosis and the fact that it is the most common inherited disease in the United States, my husband and I are concerned for

the welfare of my children. I have two son's; Mitchel is 12 and Trevor, is 8, both of whom are here with me today. Throughout their lives we have received conflicting advice on how to approach their specific risk for developing hemochromatosis. We were told to have period blood tests to measure iron levels, we were told to do nothing, we were told to constantly monitor their diets and on and on. While any and all of it may have been good advice, none of it is as good as solidly knowing for certain that Mitchel and Trevor carry the genetic mutation for the disease that contributed to my father's death and is an issue in the lives of my siblings. With that knowledge we could take proven measure to lessen the impact that a predisposition to this disease might have on their lives.

Like storm predicting and tracking capabilities genetic testing seems to offer an opportunity to learn more about the constitution of diseases and potential serious damage. It can help us track the progression of diseases as well as determine treatment or even protective measures to avoid the storm that may develop as a result of the genetic mutation. For my children this could be life altering information – altering in that it will decrease the likelihood that they will be incapacitated by hemochromatosis. For diseases such as ovarian cancer it can mean the difference between prophylactic treatment that can allow women at high risk to lead a long and healthy life and the stark contrast of often futile and painful death prolonging treatment.

Throughout the recorded history of hurricanes, experiences have gone from the storms that seemed to come out of nowhere as recently as 100 years ago, to those that we were able to track minute by minute just 100 days ago. This dramatic change is not the result of a decrease in the power of the storm but rather an increase in technology and our understanding of hurricanes. Along the same lines technological advances in the arena of genetic testing can similarly provide enhanced predictability and a greater level of protection for those at high risk. And that risk or even knowledge of a potential risk can be protected through S. 1053.

While the technology for physical protection through genetic testing, seems to be in place or at least advancing at a relatively rapid clip, the social and economic protections are not. As it stands right now, if my children undergo genetic testing for hemochromatosis, they risk not being able to obtain health insurance when they are no longer on my husband's policy and possible discrimination when they seek employment.

So, we are given a choice, protect their health, or their livelihood – it's troubling to me that as American's we are at a point where we would have to make such a terrible choice. It is also troubles me that S. 1053 offers protection that would eliminate the need for my husband and I to make such difficult a choice but it has not been brought to a vote in the House of Representatives.

All of this is reminiscent of a series of choices that were being made 40 years ago. In the late 50's and early 60's my parent fought diligently for the rights of people who were genetically different than them. Not that these people were at higher risk of developing hemochromatosis or ovarian cancer, but that their skin was a different shade of beautiful. And my parents, along with many others won that fight. The civil right act amendments are there to protect people from discrimination based on genetic make-up that we can see, be it skin tone, gender, or physical disability. A person's genetic make-up that isn't visible should be equally protected under the same terms and can be through S. 1053.

It's remarkable to me to realize that the work my parents did for the civil rights act in the Sixties was not complete. Here I am 40 years later, working on the same issue, equal rights and protection under the law, no matter the genetic make up of the person. The fact that we can look inside the DNA of a person to know more about them, should not preclude them from the protection that was fought for so valiantly.

As I see it, genetic testing is the weather tracking device of health. Just as we rely on weather tracking technologies to predict and allow us to protect ourselves from hurricanes or other weather related storms, I urge you to allow us to do the same for genetic diseases. We must move forward in protecting people from the potential storms in their lives. You can do this by urging Secretary Thomson and my congressman, Speaker Hastert to bring this bill to a vote in the House of Representatives. I am sure you can agree with me when I say that protecting lives is equally or more important than protecting property. If we can, we should and S. 1053 will.

-END-

Secretary's Advisory Committee on Genetics, Health, and Society

Testimony

October 18, 2004

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Thank you for inviting me to participate in today's committee meeting; I appreciate all that this committee has done in the past to address genetic discrimination concerns, and I hope my remarks today help to inform the committee's actions going forward.

In a provocative October, 2003, editorial in the journal *Science*, Nobel laureate Sydney Brenner wonders what medicine will be like in the year 2053, one hundred years after the discovery of DNA. "Perhaps," he writes, "the prime value of our work to society will be the creation of a new public health paradigm in which...those who have a genetic background that makes them especially liable to one of the diseases of our civilization, will have to learn how to take extra care."

Dr. Brenner needn't wait 50 more years to see this prediction realized. Some of us – those who possess BRCA1 or 2 mutations known to predispose us to breast, ovarian, and colon cancer – are already taking extra care: we're premenopausal, we have children, and we're getting breast cancers at a geometrically-higher rate (some estimates reflect 5 times the risk) of the general population. Published late last year, the New York Breast Cancer Study – representing thousands of participants, 65 oncologists, surgeons, and genetic counselors from 12 hospitals and cancer centers throughout the New York area – established that 67% of women with BRCA1 or 2 are diagnosed with breast cancer by the time they are 50. I have a cousin who died of it when she was 28. I have another who is battling Stage IV ovarian cancer as we speak; she has a 4 year old. My mother, who had breast cancer at 35, was 4 years old when her mother died of ovarian cancer at 41. Her sister – my cousin's mother – was 32. I had an invasive, aggressive breast cancer when I was 31. My daughter, a 21-year-old, stands in this line, as well: she, too, tested positive for BRCA1. She, too, will "have to learn how to take extra care."

But the care she will “have to learn how to take” includes not only the low-fat diet and daily exercise regimen she has undertaken. It includes more than the breast self-exams she is required to perform monthly. It even goes beyond the MRIs of her breasts she will have to undergo every six months once she turns 25. The care she will “have to learn how to take” further demands that she – like me, like all the others in our family who have a known BRCA1 mutation – hide her genetic information, even – and perhaps especially – from those healthcare providers most likely to help her manage this lifelong predisposition to disease. Unfortunately, hiding integral health information is the only fail-safe way she can avoid discriminatory practices such as the loss or denial of health insurance or the loss or denial of employment. Our government has failed to pass comprehensive, federal legislation that patently forbids insurance or employment discrimination on the basis of genetic predisposition to disease.

The argument has been advanced that “seeking to ban DNA discrimination isn’t really necessary,”¹ because discrimination based upon genetic information simply doesn’t exist. Actually, genetic discrimination *does* exist. But the fact that it exists only sporadically and anecdotally is a function of the newness of the technology and the fact that useful predictive genetic information (like ours) is not yet ubiquitous – it is not a function of insurance companies’ and employers’ decision to take the moral high road and, out of the kindness of their hearts, remain disinterested in this information in the same way that they are legally obliged to remain disinterested in information such as race, gender, creed, or sexual preference.

I know from experience that insurance companies don’t work this way. When I was sick, I worked as a medical librarian for a small community hospital in south Florida. The hospital was self-insured and a third-party administrator managed our insurance plan. About a year after my last treatment I was sitting at my desk when the phone rang. The flustered young woman at the other end of the line told me I was the fourth person she had been transferred to, and someone along the line had advised her that I could give her the information she needed. “Perhaps I can,” I offered. “Well,” she began, “I am calling about Rebecca Fisher. Her bone marrow transplant and other health care costs exceeded the calendar year cap last year and I’m calling to find out if that’s going to happen again this year.” “I’m Rebecca Fisher,” I said. “And I *really* hope not!”

¹ Sharon Begley article – WSJ, March 2004.

This experience taught me that there are people who are paid to look at me and see – not my ability to contribute to a community; not my honesty, integrity, or faith; not my education, hard work, or social conscience; not my family members and the ways in which I have helped each of *them* succeed – but dollar signs, costs, increased liability, and the odds of my dying an expensive death. Let us face the fact that financial incentives to use genetic information are already present: the Washington Post reported last month that “employer-sponsored health insurance premiums rose 11.2 percent this year” and are expected to rise 13% next year.² With these increases in mind and no enjoinder against using genetic information to predict future losses, it is a failure of stewardship to expect insurance companies and employers to simply “do the right thing,” and – when they don’t – lavish precious man-hours, healthcare dollars, and litigation costs to undo the damage.

I fear for my children – especially for my daughter – who must live not only with an exponentially higher risk of developing a terminal disease but also with the burden of never knowing whether or when she will legally be asked to take a genetic test as a condition of employment, be lawfully fired from a job because of her genetic condition, or be legitimately denied health or life insurance on the basis of her genetic predisposition to disease.

It’s true: we live in a world that has no safety net for us: not even HIPAA. Many people simply do not understand that HIPAA is no panacea for all that ails health privacy: HIPAA addresses none of our employment concerns, and ERISA rules exempt employer-based health plans – like the one at the small hospital where I worked – from mandatory HIPAA compliance. If my BRCA1 positivity been known in 1994 and the HIPAA protections of today were in place then, the young woman on the other end of the phone could well – and legally – have recommended to her superiors that I not be extended further health insurance coverage. The “HIPAA Gap” is deep and wide: of the 137.1 million private sector American employees who have health insurance, a whopping 45% – 62.7 million Americans – fall into it. The genetic information of each

² Health Insurance Costs Keep Rising – September 10, 2004. Available at <http://www.washingtonpost.com/wp-dyn/articles/A8287-2004Sep9.html>

one of these individuals – together with the genetic information of every uninsured American (another 45 million people) – is fair game.

In my opinion and experience, genetic information is no different from any other essential distinguishing information about any human being, all of which is – by law – kept off the bargaining table that bears up our human rights-based society. But if this argument is truly different, if – because of its fiscal component, as the United States Chamber of Commerce might argue – we must locate this debate within the framework of an implicit utilitarianism, I would point to professional contributions I and other genetically vulnerable people have been able to make because we've been lucky enough to remain "employable." I'd point to the contributions my daughter hopes to make with her two degrees – in Public Policy and Economics – from Duke University. I would point to the way in which our families' completion of innumerable psychological questionnaires, the donation of tissue samples, and the giving of our blood have advanced medical science. I would argue that we are, in fact, making a difference for the health of *all* people, that we've lived up to our end of the 'social contract' and deserve the same fundamental legal protections that are extended to all Americans.

Last summer, attorney Lawrence Lorber (representing the U.S. Chamber of Commerce, the loudest voice speaking against federal genetic information protections) told a House Education and Workforce Committee that "...the threat of allegations of discrimination from both a liability and public relations perspective is enough to prevent...employers from ever contemplating acquiring any genetic information." I would like Mr. Lorber to tell that to my friend Susan, whose 38-year-old sister is being treated for breast cancer, whose mother had premenopausal breast cancer, and whose aunt died of it. We sat together at one of our sons' ice hockey games last winter as she shared her story. Without wanting to push, I gently asked her whether she had considered speaking with a genetic counselor. "Oh no!" she exclaimed, "I would never want to risk losing my insurance!" Fear and innuendo surround the Brave New World of genetic information; people are afraid. Their fear keeps them from being tested, even when that test might make the difference between whether they live or die. And at the risk of sounding paranoid, I would go on to suggest that none of us present today can afford the luxury of writing off this problem to just high-risk individuals and families. The stage is already set for a problem of catastrophic proportions: Guthrie spot programs – whereby every

newborn infant's blood is collected, screened, *and stored* – are found “in all of the states and territories of the U.S. [and provide] what is potentially the largest and most complete genetic bank and library available in the country.”³ The continued “non-use” of genetic information implied by insurance companies' and employers' lack of interest to date does not provide sufficient safeguards for any of us, high risk or no. We with strong family histories of disease, in which the baton of illness has been passed from generation to generation, are simply the first line of defense against a staggering spectrum of possible abuses. We want to be heard. We want to be protected. And we don't want to sit in the back of the bus anymore.

In closing, I'd like to return to Dr. Sydney Brenner's *Science* editorial. Asked by a student what ethical standards should be adopted by life scientists, he writes, “I could immediately think of two prescriptions. The first, common to all scientists, is to tell the truth. The second is to stand up for all humanity.”

Let us place genetic information in the domain of all that is sacred and inviolable about human life and work toward a world in which this information can be used only for our good and the good of all humankind.

³ <http://www.mostgene.org/gd/gdvol15b.htm#attract>

I am writing to tell you my story of Genetic Discrimination. I work for a small company of about 5 people including my bosses (the owners). We are a tight knit "family". They have been with me through my mother dying and my own genetic testing for BRCA mutations. I was very open with my experience just because we ARE a small company and there was no way to hide it. After finding out I was positive for the BRCA 1 mutation that means I have an 80% lifetime chance of getting ovarian cancer and a 45% chance of getting breast cancer I began preventative steps. I had a hysterectomy in October of 2003 and a Prophylactic Mastectomy in March of 2004 (I am still in the middle of reconstruction). A few months ago our health insurance bill came in the mail and it had gone up \$13,000 a year. My boss yelled it through the office. I knew exactly what she was getting at but what am I to do? Anyway, then she began asking me if I'd switch to my husband's insurance. They even offered me extra money per hour to switch. Since I am STILL in the process of reconstruction and it was like pulling teeth to get my insurance company to pay for my procedures I didn't feel it was in my best interest to switch right in the middle of all of this, AND I have great insurance and didn't think I should lose it. I feel that ANYONE in the company could be diagnosed with ANYTHING tomorrow and that it's not fair that just because I am taking these steps to PREVENT a horrible disease in the future that I be asked to drop insurance that is important to me. We finally came to an agreement that employees would have to start paying half - of course then I look like the "bad guy" to other employees because it's not like my bosses were discrete or anything about this. I just felt like I was being persecuted for the insurance bill going up as if I WANTED to be in my situation. The insurance company came right out and told my bosses that it was all because of me that the insurance went up and that if I were dropped it would not go up that much. I also take steps toward keeping healthy - I don't smoke, I'm not overweight, I workout and eat right (most of the time). While others in the company are smokers and overweight. It just seemed so unfair especially with all that I was dealing with at the moment. I didn't think it was right to add that stress to me. If someone in the company was diagnosed with cancer they wouldn't ask them to find other insurance because the insurance premium went up. So anyway, that's my story. I am worried about my future if I'm already feeling singled out. Thank you for your time and if you have any questions please feel free to email me.

Tonia Phillips
[REDACTED]

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Tonia Phillips
Office Manager
Heathcare Contract Resources
[REDACTED]

Testimony of Tonia Phillips

Now we'll hear from Tonia Phillips.

MS. PHILLIPS: Good morning, ladies and gentlemen of the committee. My name is Tonia Phillips, and I'm here to tell my story. It is short and sweet.

I work for a small company of about four people, including my two bosses, the owners. We are a tight-knit family. They have been with me through my mother dying of ovarian cancer in April of 2002 and my own genetic testing for BRCA mutations. I was very open with my experience just because we are a small company and there was no way to hide it.

After finding out I was positive for the BRCA1 mutation in March of 2003, which means I have an 80 percent lifetime chance of getting ovarian cancer and a 45 percent lifetime chance of getting breast cancer, I began preventive steps. I had a hysterectomy in October of 2003 and a prophylactic mastectomy in March of this year, and I'm still in the middle of reconstruction.

About four months ago, our group health insurance bill came in the mail, and it had gone up \$13,000 a year for four people. My boss got the bill and yelled it through the office. I knew that she was directing that towards me. I was immediately asked to switch to my husband's health insurance policy because my situation was the reason the insurance premium went up so much, and they said that if I was taken off the policy, it would not go up. I was even told they would raise my hourly rate if I switched.

I told her I was not comfortable with switching insurance companies at the time because I was still in the reconstruction process. It was like pulling teeth to get the insurance company to pay for these procedures, and switching would confuse and complicate everything. I didn't think it was in my best interest to switch while I still needed more surgery. My feeling is that anyone in the company could be diagnosed with anything tomorrow and that it's not fair that I be asked to drop insurance that is important to me. I was doing something that would prevent me from going through a horrible disease that would cost much more than these preventive surgeries I was having.

We finally came to an agreement that employees would have to start paying half of their premium, which was fine and fair with me, but I'm sure the other employees weren't too happy with me. It seems unfair to me that I am taking steps to keep myself healthy and to prevent cancer in the future, and I am being singled out and made to feel I am a liability. I also don't smoke, I work out, I eat right most of the time. If someone in the company were diagnosed with cancer or some other disease, they would not have been asked to switch insurance companies as I was asked. I hope that me coming here and telling my story will help with defining the problem and passing laws against genetic discrimination of any kind.

Thank you.

MS. MASNY: Thank you, Ms. Phillips. You definitely do help to define the problem.

Dear Committee Members,

Hello, my name is Paula Funk. I am from Little Rock Arkansas. I am thirty-three years old and married with twin three year old daughters. I have a strong family history of breast and ovarian cancer. The following is my family history. My dad is one of ten children and all five of his sisters have had breast cancer. I have eight cousins that have had breast cancer as well. Thirteen out of twenty-four women have had breast cancer. My Aunt Dorothy has survived breast cancer twice. Aunt Dorothy is currently fighting a very aggressive ovarian cancer. The breast cancer in my family develops at an early age. Two of the youngest women were only thirty.

This disease is something that the women of my family have to constantly think about. My first memory in life is taking my Aunt Dorothy to her treatments to fight breast cancer. Because of my family history, I knew that I had to aggressively fight the possibility of breast and ovarian cancer. Ten years ago I chose to pursue genetic testing for the first time. The fear of genetic discrimination was a real concern. Ten years ago it was necessary to have many women from one family give a blood sample to discover if there was a mutated breast cancer gene. I approached my family about the possibility of testing. Their doctors advised them not to take the test due to genetic discrimination. I chose not to be tested at that time based on those recommendations.

During the following decade, I watched more women in my family develop breast and ovarian cancer. Some of those women lost their battle to cancer. Because of the continuing devastation of this disease, I began to revisit the idea of genetic testing in May of 2003. The test no longer required blood samples from multiple women in the same family. I contacted Sarah Jackson, a genetic counselor at The University of Arkansas for Medical Science. Sarah advised me about both the medical and social implications of testing. She explained that genes are the instructions for how our body works and develops. We have two copies of every gene in our body. This happens because we get one copy from mom and one copy from dad. This is why we may look a little like your mom and a little like your dad. Just like genes influence how someone looks, they are also important in health. Some genes have mistakes which are called mutations. Every one of us has many mutations including those that we inherit. It is thought that we all inherit at least six disease causing mutations. Usually these are recessive and are only a problem if both parents have the same mutation. BRCA (BR) breast (CA) cancer mutation is different. It is a dominant gene. This mutation is enough to cause hereditary breast or ovarian cancer.

Sarah also advised me about the potential risks of being discriminated against. After talking with my genetic counselor, I asked my father to take the test first since the strong history of breast and ovarian cancer comes from his side of the family. He took the test, and it came back positive for BRCA1 gene mutation. My father's positive test for BRCA1 gave me a fifty percent chance of inheriting the same mutation. I then chose to take the test and also came back BRCA1 positive. Not only is this mutation a problem for me, it is a problem for my family. This gene change, or mutation, is one of my BRCA1 genes. The other copy is fine. My twin daughters, Audrey and Anna have a fifty percent risk of getting the working gene or the not working gene just as I did when my mother and father

conceived me. My life time risk for breast cancer is up to eighty- eight percent. I have a forty- four percent lifetime risk of ovarian cancer. Ovarian cancer is particularly alarming to me because by the time it is detected there could be a fifty percent mortality rate.

I am so grateful that I have an opportunity to save my life. I will have a prophylactic bilateral mastectomy this fall. I will go from having an eighty- eight percent chance that I will have breast cancer to a ninety- five percent chance that I will never have to face breast cancer. After I finish having children, I plan to have my ovaries removed. This procedure reduces my chances of ovarian cancer to five percent. That is amazing to me. I will no longer have to live with the constant fear of cancer and death.

I decided to risk genetic discrimination because I came to the place where the fear of death out weighed the fear of discrimination. I have had difficulties with discrimination through out my journey of discovering that I am BRCA1 positive. The first thing I mentioned earlier is that I had to put off testing for ten years because doctors advised against testing because of discrimination. The doctors told us that there are no laws to protect against discrimination. Genetic testing, now and in the future, could prevent us from getting insurance. Many women in my family have developed cancer during that time. Knowledge about my health is a gift. It grieves me to think of the lives and the pain and suffering that could have been avoided if my family had been tested ten years ago.

Based on the fear of discrimination, my father and I paid for our own tests. We wanted to prevent insurance companies from learning about my genetic status. Another problem that has occurred was last year while I was waiting to be tested. Even after I knew my dad was positive, I had to wait three torturous months to take the genetic test. My husband and I were starting a new computer programming business. I spent three months looking for an insurance company that would be less likely to cancel if I came back BRCA1 positive. This proved to be a difficult undertaking. I learned that individual insurance is not protected by the HIPPA laws. They can exclude coverage of many things based on pre-existing conditions. I knew that type of policy could not protect me from discrimination. I knew that BRCA1 could be considered a pre-existing condition. I had already been denied an individual family policy from Blue Cross Blue Shield because I had a cesarean section. I then began to look for small business group coverage. I was told time and time again that a company with only two employees does not qualify for group coverage. This was extremely frustrating. My husband and I had decided that if we could not find insurance that qualified our company for a group policy we would have to close the business. My husband would have to work for a big company with the protection of large group insurance. It seemed that my family had fallen in a gap where we had no protection from any of the existing laws to protect us from discrimination. After three months of looking for small group insurance, I talked with United Health Care and they had just started qualifying businesses with two employees as a small group. During the three months we were waiting for insurance, my genetic doctor and counselor advised me not to come in and have an official appointment because it could be recorded as a preexisting condition.

Even after I had a group insurance policy and knew I was BRCA1 positive, the possibility of discrimination continued to effect my medical management and the medical management of others in my family. There are many new screening tests that detect cancer much earlier than the traditional mammograms. The breast MRI is an example of the new advanced screenings. These tests are all very expensive. My doctors changed my diagnosis code to high risk instead of BRCA1 positive to justify having the MRI. If there was no threat of discrimination, doctors would code that I am BRCA1 positive which is much more powerful at justifying this expensive test. I have family members currently who are emotionally ready to take the test but choose not to because of insurance. I have one cousin whose husband is a preacher at a very small church. He has an individual insurance policy. She is undecided if she should take the test at this time because she can not be protected by the HIPPA law. She is another example of patients that are falling through the gaps of existing laws. I am currently contacting eighty- six family members to explain their genetically predisposed risks. As I talk with the different members of my family, one of their biggest fears and the area they have the most questions is about insurance and genetic discrimination in general.

One last area I will mention is the problem I am currently having with my insurance company. I have had my insurance for less than a year, and they have already raised my rates one hundred dollars a month. Other than the prevention screenings, I have had very few medical expenses. I have talked to a few medical people who have said this looks suspicious. I have also been waiting almost two months for approval for my mastectomy. Insurance has made little progress with the approval. I do not understand why a life saving surgery has to go to a board of approval when they know the risks of being BRCA1 positive. Five years ago my father had bypass surgery to prevent a heart attack. His surgery did not have to go to a board of review to be approved. Both surgeries prevent life threatening diseases.

I deeply appreciate the committee giving me a chance to tell not only my story, but my family's story. I pray that you will be able to push the Genetic Information Nondiscrimination Act of 2003 to be passed by the Congress. My medical management and the medical management of many in my family have been greatly effected by avoiding genetic discrimination. The avoidance of discrimination has become a major part of my life. Discrimination is something that worries me for the future as well. A few weeks ago my husband, Jonathan, and my girls, Audrey and Anna, and I walked in the Susan G. Colman's race for the cure. My girls asked many questions during that race. I kept my answers simple because they are young, but I could not help but think about what a complex issue this has been for me. As I mentioned earlier, my girls have a fifty percent chance of having the BRCA1 mutation. I hope that when they are old enough to decide if they want to be tested, laws will be in place where they do not have to even consider discrimination. Knowing my genetic status is permanent. It is not something I can take back. It is imperative that we pass laws that keep up with science to protect all patients from discrimination now and in the future.

Thank you for your time and attention in this very important matter,

Paula Funk

Sarata, Amanda (NIH/OD)

From: Paula Funk [REDACTED]
Sent: Thursday, August 19, 2004 1:29 AM
To: Sarata, Amanda (NIH/OD)
Cc: Jackson, Sarah A
Subject: Genetic discrimination testimony

Dear Amanda,

Hello, my name is Paula Funk. I live in Little Rock AR. I am 33 years old married with twin 3 year old daughters. I am BRCA Strand 1 positive. I am considering a prophylactic mastectomy this fall.

Here is some background about my family. My Dad is 1 of 10 children. All five of his sisters have had breast cancer stating as young as 30 years old. I have 8 cousins that have had breast cancer. 13 out of 24 women have had breast cancer. I have an aunt that has survived breast cancer 2 times and is now fighting ovarian cancer. I also have a 38 year old cousin battling breast cancer currently. There is now early screening and preventative surgeries that could save many lives in my family. I am in the process of sending out information packets to 86 different relatives to inform them of their risks and their children's risk of being BRCA Strand 1 positive.

Many of my relatives express concern about insurance and employment when they consider being genetically tested. As you can imagine this is a subject that I am passionate about. There are some laws in Arkansas that protect patients from discrimination but the laws are limited and only protect you in this state. The fear of insurance kept me from being tested for 10 years. Finally the fear of death out weighed the fear of insurance so I pressed on and discovered that I was positive. My father was tested first and paid for his own test and then paid for my test because of insurance concerns. My insurance now has access to my genetic status because of justifications for expensive screening tests such as the breast MRI. They have been easy to work with so far. I am grateful for the laws that Arkansas has.

I would love to give any information or testimony to help prevent genetic discrimination. I spoke at UAMS in June about my patients perspective as it relates to genetic testing. Please put me on your e-mail list and keep me updated on the bill's progress and let me know how I can help. I may have a surgery at some point this fall but I am undecided about that at this point.

I am grateful to you for your hard work on this important subject. The life of this bill could influence the health of many in my family.
Thanks again,

Paula Funk
[REDACTED]
Little Rock, AR 72211
[REDACTED]
[REDACTED]

Testimony of Carolina Hinestrosa

We'll now hear from Ms. Hinestrosa.

MS. HINESTROSA: Good morning. My name is Carolina Hinestrosa. I am a 10-year, two-time breast cancer survivor. I'm a mother of a 13-year-old daughter. I'm also the executive vice president for programs and planning of the National Breast Cancer Coalition.

My first diagnosis with breast cancer was at the age of 35. My second diagnosis was at the age of 40. My younger sister was also diagnosed twice, first at age 29, and then at 34. Over Christmas last year, two of my cousins and an aunt were diagnosed with breast cancer as well. Of course, we suspect there is a genetic mutation that predisposes members of my family to breast cancer.

I sought genetic counseling as part of a study. After carefully weighing the potential benefits and harms of genetic testing, I decided not to undergo testing for fear of potential consequences to my daughter. My fears are two-fold, first that the information may not be protected and might even be misused. I also worry that if I test positive, my daughter might be obligated to disclose the presence of a genetic mutation and that she might suffer future discrimination in health insurance and employment as a consequence.

I have four sisters and a brother. We all worry about our risk for breast cancer and the potential risk for our daughters, yet none of us feel safe enough to undergo genetic testing. My family experience illustrates why our nation needs strong nondiscrimination laws.

Since its founding in 1991, the National Breast Cancer Coalition, of which I am a member and an executive vice president, has changed the world of breast cancer in public policy, science, industry and advocacy by empowering those with breast cancer, our families and friends, and creating new partnerships, collaborations, research foundation opportunities, and avenues for quality access to health care.

The National Breast Cancer Coalition is now over 600 strong in terms of organizations who are members, and we represent several million patients, professionals, women, our families and friends. Coalition members include cancer support information and service groups, as well as women's health and provider organizations.

The mapping of the human genome has brought with it the promise of reducing human suffering by targeting interventions for those at risk for disease. The National Breast Cancer Coalition believes that strong legislative and regulatory strategies must be established to address the protection of individuals from the misuse of genetic information at the national, state and local levels of government. Genetic information is uniquely private information that should not be disclosed without authorization by the individual. Improper disclosure can lead to significant harm, including discrimination in the areas of employment, education, health care, and insurance.

The 1996 Health Insurance Portability and Accountability Act, HIPAA, took significant steps toward extending protection to individuals from genetic discrimination in the health insurance arena by creating privacy standards, but this law does not go far enough. It is time to extend protections against genetic discrimination to everyone. The development of new genetic tests necessitates legislative and regulatory strategies to address the issue of how to protect individuals from the misuse of their genetic information.

Fear of potential discrimination threatens both a woman's decision to use new genetic

technologies and to seek the best medical care. Women are also afraid to enroll in research and clinical trials that involve genetic studies, and this in turn threatens the viability of the scientific community to conduct the research necessary to understand the cause and find a cure for breast cancer. Many of the women testifying at present in this audience today have experienced exactly those concerns.

NBCC strongly supports the enactment of legislation that would protect millions of individuals against discrimination not only in health insurance but also in the workplace and that will provide strong enforcement mechanisms that include the private right of action. For this reason, NBCC supports H.R. 1910, the Genetic Nondiscrimination Health Insurance and Employment Act authored by Congresswomen Louise Slaughter. This legislation prohibits health plans from requesting, requiring, collecting or disclosing genetic information without prior specific written authorization of the individual; from using genetic information or an individual's request for genetic services to deny or limit any coverage, to establish eligibility, continuation, enrollment, or contribution requirements; and from establishing differential rates or premium payments based on genetic information or an individual's request for genetic services.

This legislation also prohibits employers from using genetic information to affect the hiring of an individual or to affect the terms, conditions, privileges, benefits, or termination of employment unless the employment organization can prove this information is job related and consistent with business necessity. Also, from requesting, requiring, collecting or disclosing genetic information prior to a conditional offer of employment; or under all other circumstances requesting or requiring collection or disclosure of genetic information unless the employment organization can prove this information is job related and consistent with business necessity.

It also prohibits from accessing genetic information contained in medical records released by individuals as a condition of employment in claims filed for reimbursement for health care costs and other services. Also, it prohibits releasing genetic information without specific prior written authorization of the individual.

Most importantly, H.R. 1910 contains strong enforcement language and provides individuals with a private right of action to go to court for legal and equitable relief if they are a victim of genetic discrimination whether they are subject to discrimination by the health plan or the employer.

NBCC does not support the Genetic Nondiscrimination in Health Insurance and Employment Act, S. 1053, passed by the Senate in October 2003, because it does not contain sufficient enforcement provisions. Unlike H.R. 1910, S. 1053 does not provide individuals with a private right of action should they become a victim of genetic discrimination in the individual insurance market. NBCC believes that a right with no enforcement is really not a right at all. It is for that reason that no matter how carefully a bill is worded, no matter how much effort is put into it, including protections that breast cancer patients need, if that bill does not have a strong enforcement mechanism, then NBCC will not support it.

As we clearly can see from the witnesses here today, genetic discrimination is a real and growing problem that needs an immediate solution, not one that should wait until we have further cases of women and men who have experienced this type of discrimination that is so detrimental to the ability to seek quality health care.

Thank you for the opportunity to share the views of the National Breast Cancer Coalition.

MS. MASNY: Thank you very much for your own personal experience, as well as for the views

SACGHS Meeting Transcript
October 18-19, 2004

of the National Breast Cancer Coalition.

Testimony of Phil Hardt

Lastly, we'll hear from Phil Hardt.

MR. HARDT: Good morning. It's a privilege to be here today, and I want to thank the committee for inviting me to share my thoughts and personal experiences with everyone on the critical subject of genetic discrimination.

I have two genetic diseases, hemophilia B, a bleeding disorder, which I inherited from my mother, and also Huntington's disease, a degenerative brain disorder, which I inherited from my father. My two biological daughters and granddaughters are all carriers of hemophilia B, and as a result I now have two handsome grandsons who must also infuse with clotting factor each time they get hurt. All three of my biological children were at risk for Huntington's disease, but I am happy to report that none of them carry the destructive gene and cannot pass it on to subsequent generations. One tested publicly, and two tested anonymously to conceal their outcomes.

I mention biological children because I also have five adopted children, four of whom have severe handicaps.

Nevertheless, our story is one of continuing genetic discrimination even though we have laws that are supposed to protect me, my children, and my grandchildren.

"It was the best of times, it was the worst of times," as Dickens said in "A Tale of Two Cities." Because of advancements with the Human Genome Project, we now stand on the brink of having more useful information that has the potential of helping literally millions of individuals prepare early for various diseases. However, the reality is the knowledge that you are carrying any particular genetic disorder, in my case hemophilia and HD, is just as devastating to you, your children and your grandchildren as the disease will be later. This is further exasperated in Huntington's disease because of the severity of its symptoms and the absolute necessity for those who face the 50/50 chance of inheriting it to prepare early and thoroughly in order to minimize its overall destruction.

Tens of thousands of individuals with Huntington's disease have lived and died and are already in the insurance company's profitability calculations. However, it wasn't noted on their death certificates because of genetic discrimination fears. It is ludicrous now to believe that because you can know early that you might inherit a genetic disorder that all of a sudden we're going to create higher medical costs. This is not the case. We are living examples of the Tiresias complex. If you remember, the blind seer Tiresias confronted Oedipus with the dilemma, "It is but sorrow to be wise when wisdom profits not."

Huntington's disease is an inherited progressively degenerative brain disorder that results in loss of both mental faculties and physical control. It causes brain cells to die prematurely. Loss of these brain cells causes very specific impairment and eventually death. Every child of an affected parent has a 50 percent chance of inheriting the gene and developing the disorder themselves. If HD is passed on by the father, another risk exists of anticipation occurring and each gene-positive child becoming symptomatic, even as early as a young infant or in their teenage years.

HD symptoms debilitate a person when they least expect it, usually in the prime of their lives, around 40 years of age, when they still have children at home and are actively pursuing careers. Living with HD is like living with Alzheimer's, Parkinson's, MS, and going insane all at the same time. Genetic testing has been available for Huntington's disease for longer than any other adult-

SACGHS Meeting Transcript
October 18-19, 2004

onset disorder, since 1993. The discovery of the genetic mutation causing Huntington's disease made possible the use of predictive testing to identify current unaffected carriers. In 2000 Cohen said, "Genetic testing is intended to give families with a family history of HD the opportunity to assess their own risk for developing the disease more specifically, monitor their health status closely and, if a predictive mutation is present, make informed choices about reproduction and lifestyle."

It is interesting to note here that before 1993, the almost quarter of a million individuals who are at risk for HD in the United States were polled, and overwhelmingly about 90 percent of them said that they would take advantage of the test to find out if they were carrying the destructive HD gene. However, since the definitive test became available, fewer than 10 percent have tested as a direct result of genetic discrimination.

I'd like to now tell a little bit about my family history. In 1971, I was diagnosed with hemophilia B. In 1989, I was hired by Allied Signal Automotive and told by the HR manager there not to tell my boss about my hemophilia or I would never be promoted or trained because he wanted to get the biggest return on investment for his bucks, and if he knew I might have a disability, I would never go anywhere in the company. Consequently, all future bleeding episodes had to be hidden from him.

In 1996, a claim I filed for credit insurance on a car I had purchased for my daughter was denied because I had recently seen a neurologist regarding problems that I was having. In 1997, I was diagnosed as having Huntington's disease. In the year 2000, my oldest daughter married and applied for mortgage life insurance. She was turned down by every major insurance company because of Huntington's disease. Copies of several rejection letters are included in your packets, and note that the insurance companies don't even have fear of putting their rejections in writing.

Each of her rejection letters state two pertinent facts that are important. Number one, they each state that they will not insure her until she has tested for Huntington's disease, and two, that she is found to be negative. Then the insurance agent on one of the letters where they insure her husband writes a note at the bottom that says when you find out your status for HD, then we can insure your children, showing that the discrimination is down to the third generation now.

In 2002, my grandson, Enoch Maximillion, is denied health insurance coverage because of hemophilia that he inherited from me, and a copy of this denial is also included in your handouts. They must now earn less than they are capable of to qualify for state welfare in order to get coverage.

In 2002, my daughter Michelle and son Phillip tested anonymously for HD to protect them in case either of them tested positive. I am over the Huntington's Disease Society of America, Arizona affiliate in the State of Arizona, and in 2001 a geneticist and I established anonymous genetic testing to protect individuals so that they can use a bogus name and social security number and address and all other information, and pay cash. But the problem is it's very expensive. It's around \$900 out of pocket to find out. But it is completely concealed. But it's a shame that we have to do this.

Last year I applied for long-term care insurance and was rejected on the basis of my HD after becoming divorced and realizing that I would probably need someone to take care of me later.

Now, here is a list of ways that open genetic discrimination adversely affects those with HD over and above the negative effects of the disease itself. Those who are at risk are reluctant to

SACGHS Meeting Transcript
October 18-19, 2004

participate in research, even anonymous research, because they fear being found out. For example, the PHAROS study for HD could have almost a quarter of a million at-risk individuals in it, but they have only been able to recruit about 1,000. Imagine the decrease in numbers. Other important research tests are no different. Because of our small numbers, unfortunately, we need every bit of data possible to make things significant.

Proper medical and mental health care are not sought on a timely basis that could have (inaudible) help reduce suffering and raise everyone's quality of life. Open communication is almost non-existent between parents and their at-risk children regarding how they can better prepare to minimize the destruction of HD if they do have it. HD must be kept shrouded in secrecy to protect everyone. For the same reason, at-risk children are not encouraged to seek good education, college education, careers with companies who offer group benefits, marriage and childbearing options, including adoption. Misdiagnosis and the same thing with medication occur because one doesn't know, or knows but can't be honest with their doctors and other health care providers for fear of being discovered. Healthy living habits aren't adopted either early on to postpone onset.

Now, using our negative experiences with being wise and our wisdom not only doesn't profit us but is even used against us. How many other future discoveries that have the potential to bless the lives of millions of others by predicting other diseases soon enough for individuals to take positive action against them will be thwarted because of flagrant genetic discrimination?

Thank you very much.

MS. MASNY: Thank you, Mr. Hardt.

And thank all of you for your very profound testimony.

From: Phil Hardt [REDACTED]
Sent: Tuesday, September 21, 2004 4:31 PM
To: Sarata, Amanda (NIH/OD)
Subject: RE: Additional information on HD - 2nd Response

Dear Amanda,

After reading your question about "Have [you] paid out-of-pocket for services to exclude genetic information from medical records." I wanted to tell you that I paid out-of-pocket for my daughter Michelle and son Phillip to be tested anonymously- about \$900, so their results would not be in ANY medical record or database, just in case they were positive. They still have copies of the results made out to bogus names if you'd like. I also have the Anonymous Testing Procedure I give everybody if you'd like a copy of that too. I spent about 10 hours on the phone convincing the geneticist in Tucson that we needed this and she has been great. Someone testing anonymously just sees her and has their blood drawn, they don't see anyone else. If they are positive then she has them call me so I can meet them, get them additional information and get them coming to our support group and I also make recommendations for them to see a counselor...Phil

Sarata, Amanda (NIH/OD)

From: Phil Hardt [mailto:philhard@xxxxxxxxx]
Sent: Tuesday, September 21, 2004 2:54 PM
To: Sarata, Amanda (NIH/OD)
Subject: Additional information on HD

Dear Amanda,
I wanted to also let you know that because of the flagrant genetic discrimination happening here in Arizona which discriminates against those with Huntington's Disease, their children and grandchildren that I have also established anonymous genetic testing which doesn't follow the recommended protocol for testing, which states that you must see a geneticist, neurologist, psychiatrist and psychologist prior to having your blood drawn. The person uses a bogus name, address, phone and social security number and pays cash to find out anonymously if they have HD so if they do, they can get all of the necessary insurances before they become symptomatic. We are being forced to do the same thing that those with AIDS were forced to do 20 years ago. Unbelievable isn't it? Have a great day and please call or e-mail if you have additional questions or if I can testify or do whatever I can.

Sincerely,

Phil Hardt, CVO
HDSA Arizona Affiliate
2001 HDSA Person of the Year
[REDACTED]
Phoenix, AZ 85011
[REDACTED]

From: Phil Hardt [mailto: [REDACTED]]
Sent: Monday, September 20, 2004 7:25 PM
To: Sarata, Amanda (NIH/OD)
Subject: RE: Genetic Discrimination in AZ against Huntington's Disease - 1 of 4 - Response

Dear Amanda,

My families situation is two-fold. I inherited hemophilia B from my mother and I also inherited Huntington's Disease from my father! A double whammy. Pretty unbelievable, isn't it? Both of my oldest daughters were unable to get health insurance for themselves and their children because they are carriers of hemophilia B. In addition, my oldest daughter, Joeline, was unable to get life insurance because of simply being "at-risk" for Huntington's Disease which is flagrant genetic discrimination but our Arizona laws are so gutless that they allow this to happen. I already sent you the PowerPoint slides showing her rejection letters. Unfortunately she only kept 3 out of about 20 rejections! If you think it would help to have them apply for medical insurance and make sure they keep their rejection letter this time I would be happy to ask them to do this. They are willing to help in any way they can too.

When I was hired by Allied Signal about 18 years ago the HR manager made me promise never to tell the plant manager that I had hemophilia B because he said he would get in lots of trouble for hiring me and he said if the plant manager knew then he would never promote me or invest ANY money in, fearing I would not end up being a long-term player. Fortunately for me, because of my hemophilia, I already knew that I had to get a job whihc had group benefits, etc. which turned out to be a really big blessing when I also found out I had Huntington's Disease. I am now out on medical disability with full medical coverage so I cannot be injured by speaking out.

Genetic Discrimination in AZ

AMERICAN FAMILY LIFE INSURANCE COMPANY
AMERICAN FAMILY LIFE INSURANCE COMPANY
Service Address: 109 E WASHINGTON AVE • MADISON WI 53703-0001 • PHONE: (608) 249-2111
Mailing Address: 6001 AMERICAN PKWY • MADISON WI 53742-0001

March 21, 2000

Joeline Hollar
[REDACTED]
Chandler, AZ 85225

Re: Joeline Hollar
[REDACTED]

We would like to take this opportunity to thank you for considering American Family Life Insurance Company for your life insurance needs.

After completing our review of the application, we have determined that due to family history of Huntington's Chorea for which you have not been tested for as of yet, we are unable to issue the application. Reconsideration would be available once the testing has been completed and you test negative to this gene.

Since your application cannot be approved and no insurance has become effective, this Company assumes no liability under the application. Any receipt you may have received is null and void.

Any premium payment submitted with the application is enclosed with this letter.

Thank you for your interest in American Family Life Insurance Company.

Nancy M Minter
Underwriter
Life Underwriting Services Department

NMM

Enc.

cc: JEFF ARRHDONDO
3110 S RURAL RD STE 104
TEMPE, AZ 85782-3852

cc: GEORGER NOVAK D-402

“After completing our review of the application, we have determined that due to family history of Huntington’s Chorea for which you have not been tested for as of yet, we are unable to issue the application. Reconsideration would be available once the testing has been completed and you test negative to this gene.”

Genetic Discrimination in AZ



Life Insurance: 1-800-252-6347
Fax: 1-800-874-3965

Annuity: 1-800-348-5789
Fax: 1-800-345-5344

15 May, 2000

Jolene N Hollar

Chandler AZ 85225

Dear Mrs. Hollar:

Thank you for applying to General Life Insurance Company. Unfortunately we are unable to issue you a policy due to admitted family history of huntington's without individual testing.

The "Notice of Insurance Information Practices" that you received when you signed your application for insurance explains your right to access the information in our files and the process for correcting or deleting information. I am enclosing a copy of that form for your information. Please note under the section "Limitations on Access", that we do reserve the right to provide details of a medical nature through your personal doctor, or other medical professional of your choosing.

If you have questions or need additional information, please contact your agent, Clifford Arnett.

Sincerely,

Timothy J. Conrad
Executive Director of Underwriting

TJC/Kf
Enclosure

“Thank you for applying to General Life Insurance Company. Unfortunately we are unable to issue you a policy due to admitted family history of huntington’s without individual testing.”

Genetic Discrimination in AZ

“Please advise if applicant has had genetic testing to determine if she has the gene for Huntington Chorea. If so, where can these records be located and what were the results? If not, we must decline coverage.”

Interstate Assurance Co.

Pending Business Report as of July 7, 2000

For CLIFFORD M ARNETT

Date: July 7, 2000

Agent Number: 7Y000110

CLIFFORD M ARNETT
CHANDLER AZ 85248

Policy Number	Proposed Insured	Product	Target	Face Amount	App. Received	Request Date	Received Date	Requirement Description
00477152	Hollar, Joeline	HOMR GUARD GOL	259	150,000	5/8/2000	5/8/2000	5/16/2000	Initial Premium/Balance Due Memo to Agent
Service Agent: CLIFFORD M ARNETT								
Policy status as of: 5/17/2000 Awaiting Underwriting Requirements								
Please advise term of mortgage, if term plan is to be decreasing or level and term of premium guarantee and if Moneyback is desired for the Home Guard Gold plan. Please also advise details to two other applications made in February and March and the disposition of those applications.								
5/9/2000 5/16/2000 Teleunderwriting Memo to Agent								

Please advise if applicant has had genetic testing to determine if she has the gene for Huntington Chorea. If so, where can these records be located and what were the results? If not, we must decline coverage.

*

Genetic Discrimination in AZ

Jeff Arredondo Agency
3110 S Rural Rd Ste 104
Tempe, AZ 85282
Phone: 602-829-1211
Fax: 602-829-4978



April 3, 2000

Timothy Hollar
[Redacted]
Chandler, AZ 85225

Dear Timothy

CONGRATULATIONS!!!! You have qualified for the life insurance program you recently applied for. I am happy to enclose your new policy which you should review and file.

I trust, that if you should ever have any questions regarding this program or any of your other fine American Family policies, you will feel free to contact me. Currently the premiums are paid to April 10,2000 at which time automatic monthly deductions of \$13.30 will begin from your checking account.

Thank you again, for this opportunity to be of service , and for placing your confidence in American Family Insurance and myself.

Sincerely,

Jeff Arredondo
Your American Family Agent

“P.S. Joeline – Let me know your test results, then we can reconsider the life policy. Also, we can do your children’s policies.”

P.S.
Joeline - LET ME KNOW ~~HOW~~ YOUR TEST RESULTS, THEN WE CAN RECONSIDER THE LIFE POLICY. ALSO, WE CAN DO YOUR CHILDREN'S POLICIES.

Genetic Discrimination in AZ

From: Jill Kreitman [mailto:jill@lincsolutions.net]

Sent: Tuesday, August 10, 2004 11:59 AM

To: [REDACTED]

Subject: Long Term Care Insurance

Mr. Hardt,

I received an internet request that you were looking into Long Term Care Insurance. I am here in the valley and licensed with 6 companies. Unfortunately, if you have been diagnosed with Huntington's Disease you would not be insurable for LTCI.

If you have any questions, please don't hesitate to call me.

Thanks and have a great day!

Jill Kreitman

LTC Solutions

Local 602-867-1226

Toll Free 1-866-737-2865



Genetic Discrimination in AZ

From: "Jill Kreitman" <jill@ltsolutions.net>
Date: 2004/08/10 Tue PM 03:38:10 EDT
To: [REDACTED]
Subject: Re: Long Term Care Insurance

Hi Phil,

I am sorry I could not help you. I am licensed with John Hancock, MetLife, GE, MedAmerica, Physician's Mutual, and Allianz. I also checked with Penn Treaty. Good luck with your search. If you find someone let me know so I > can keep information for future reference is someone asks.

Take Care,

Jill



Genetic Discrimination in AZ

From: "Ray Spatti" <spatti@cox.net>

Date: 2004/08/10 Tue PM 03:36:01 EDT

To: ~~spatti@cox.net~~

Subject: Long Term Care

Phil,

Since I received your e-mail this morning I ha[v]e checked with a couple of underwriters at different companies.

I am terribly sorry, but they have told me that Huntingdon's Disease Is a disqualifier for Long Term Care insurance.

I really regret to inform you of this because I suspect that it may be a pretty serious disappointment.

If there is anything else I can do for you please do let me know.

Sincerely,

Raymond Spatti, Ph.D.



Genetic Discrimination in AZ

From: Ray Spatti [mailto:spatti@cox.net]

Sent: Tuesday, August 10, 2004 1:06 PM

To: ~~XXXXXXXXXX~~

Subject: Re: Long Term Care

Phil,

The companies are: Allianz of North America Physicians Mutual
Penn Treaty Mutual of Omaha.

The[y] all said the same thing, that the disease makes one
ineligible for coverage.

I'm sorry.

Ray



Written Comments

September 14, 2004

Dear SACGHS Members:

In 1996, I was genetically tested as part of a research study for women who may be at high risk for breast and ovarian cancer. I had some breast cancer in my family: my mother died from breast cancer at age 52; her mother and her first cousin had also been diagnosed with breast cancer. While the history looked troubling, it was not expected to yield positive genetic results. However, it did yield positive BRCA1 test results for me. This means that I face estimated risks of developing breast and ovarian cancer of up to 87% and 44%, respectively, during my lifetime.

At that time, genetic testing was a very new procedure and little was known about the ramifications of such information. After careful counseling, I was told, as was my family, that it was best to handle the situation cautiously with any physician; making sure that anything discussed about gene status would not be put in a written chart for fear of genetic discrimination. Over and over again I heard, "don't have any of your gene status put in a medical chart." Ok that seemed doable enough--until I actually had to get medical testing and diagnostic screening on a much more frequent basis than any other normal female.

For fear of my insurance company finding out what was happening, I bought an entirely separate insurance policy to have just in case my primary insurance company found out my gene status and I would lose that coverage. I bought a separate policy and paid the premiums for the chance that I would not be able to use my good primary insurance and would have to lie about my history to use the second policy should develop cancer.

Since my diagnosis, I have followed a course of being seen by one of my doctors every three months; oncologist, breast surgeon, OB-GYN. I have a mammogram every 6 months, a breast MRI annually, and breast ultrasounds as needed along with transvaginal ultrasounds. Naturally, when one goes for a diagnostic test so often, the technician asks why are you there again? What is the purpose? Do you have cancer? You should not have a test so often unless your mother died of ovarian cancer (I told her she did, I lied). Clearly, not all medical staff is trained in the manner appropriate to deal with high risk women. So I've done a lot of lying to keep them from asking too many questions, or finally in exasperation I've said, "I'm BRCA1. Get it?"

For those of us who carry genes that predispose to cancer development, if we do reveal our status to a medical staff person, we must always follow it with, "you can't put that in the chart," so there is no record. But sometimes we are not so lucky. In 2002 I had a colonoscopy (one every 5 years), and while under sedation the doctor asked my why I was there. "High risk," I replied groggily. "What does that mean?" he said. "I'm BRCA1." Oh, he understood, and I made the mistake of not saying, "you can't put that in the chart." Well, the insurance company denied the procedure and he wrote a letter to them clearly stating my gene status as the reason for the procedure. I was mortified! How could he have written such a thing? I immediately paid the bill so as not to spark any inquiry from my insurance company, and most fortunately, the doctor's damaging letter got sent to the wrong billing department and is hopefully forever lost. I cannot describe the panic and nausea that resulted from that episode.

Clearly, the fear of genetic discrimination has been a pervasive force in my medical care ever since I learned that I carry a BRCA1 gene deletion. I have withheld information from some of my healthcare providers, asked others to withhold my gene status from my chart, paid for an extra insurance policy, and paid for expensive procedures out of pocket, all because of my fear of genetic discrimination by my health insurance company. I have been fortunate to be able to have increased screenings without telling my insurance company my status. Others are not so fortunate.

I co-founded a support group, SOOAR (Supporting Ourselves and Others At Risk), and a non profit organization, STAR (Supporting Those At Risk), that is dedicated to the needs of individuals at high risk for cancer. Through these organizations I have met many people with similar experiences. The concern over genetic discrimination is real in my life and in theirs. Therefore, I urge you to use your influence to support passage of The Genetic Nondiscrimination Act.

Celia A. Boyne-Schuh

[REDACTED]
Lawrence, KS 66047
[REDACTED]
[REDACTED]

September 17, 2004

Dear Most Honorable Members of SACGHS,

I am writing this letter in response to the call for testimony regarding genetic discrimination. I would like to share with you my story in hopes of helping you understand what it is like to live with a genetic predisposition to a life-threatening disease and the added burden of genetic discrimination.

My mother died of breast cancer. Her only sister (my aunt) died of breast cancer. Their mother (my grandmother) died of ovarian cancer. My paternal grandmother died of breast cancer and my only paternal aunt died of breast cancer. At the age of 36, my sister was diagnosed with breast cancer. I began to seek answers about what was happening to the women in my family. At the age of 39, through a research study, I was found to be a carrier of a *BRCA1* genetic mutation that gives me an 85% lifetime risk of developing breast cancer and a 60 % lifetime risk of developing ovarian cancer. Because I was tested as part of a study, my test results remained private and out of my medical records. I have three sisters who were also tested and found to be carriers. They did not have the option of being tested through a research study. Because of their fear of genetic discrimination, they paid for their genetic testing out of their own pockets, even though they had very good health insurance.

My life experiences indicate that if you are a woman in my family your death comes early due to breast or ovarian cancer. At the age of 39, I was not willing to wait for my genetic predisposition to rear its ugly head and strike. After learning of my *BRCA1* gene status, I spent the next 6 months undergoing prophylactic surgeries, first a bilateral mastectomy and then a hysterectomy with oophorectomy. My initial Doctor appointments all began the same way. I first had to ask them if they would be willing to withhold information from my medical file - why? - because I was fearful of what my insurance company might do if they found out I was a *BRCA1* gene carrier. Although I have never asked or expected my Doctors to lie, I did ask them to withhold my genetic test information from my medical file.

My sisters also chose the prophylactic surgery options that I did. There are four of us and we have all had mastectomies (one because of cancer, the other three prophylactically) and hysterectomies with oophorectomies. Fortunately, our insurance companies paid for our surgeries based on our significant family history. For that I am grateful. Nowhere in any of our records is our gene status indicated. Our fear of genetic discrimination prevents us from being completely honest.

I am very concerned for my children and nieces and nephews. They all have a 50-50 chance of also carrying this genetic abnormality. They have not yet been tested. When they are ready, I along with my siblings will pay for their testing out of our own pocketbooks. Although many insurance companies will

now pay for this genetic testing, I will not encourage my children or nieces and nephews to seek payment by insurance. There are too many unknowns. There are no guarantees.

A couple of years ago, I looked into participating in my employer's group health plan (I have always been under my husband's policy). Financially it made sense for me to switch policies and take advantage of my employer's plan. Once I began completing the necessary paperwork, it came to my attention that my gene status would be revealed if I answered the questions honestly. At that point I opted to stay on my husband's policy, even though we are penalized \$900.00 a year because I decline coverage through my employer. My fear of genetic discrimination was/is too powerful for me to go forward with changing policies. I'll pay the penalty.

As a result of my experience, I co-founded and co-facilitate a support group, SOOAR (Supporting Ourselves and Others At Risk), for women who are at high risk for breast and ovarian cancer. In addition, I am President of the Board of Directors of the STAR (Supporting Those At Risk) Foundation. The STAR Foundation's mission is to provide access to genetic counseling and testing, support, education and advocacy for individuals who are at high risk for cancer. I realize the privilege my family has enjoyed in being able to 'work around the system', pay for tests out of pocket, and be assertive and educated enough to know to ask my healthcare providers to keep my medical record free of potentially damaging information. I founded SOOAR and STAR to educate individuals about risks, to support them in their journeys of living with risk, and to help pay the expensive costs related to cancer risk assessment and evaluation. In these roles, I have heard numerous stories from our participants on action or non action taken as a result of their fear of genetic discrimination. I have met women who have had prophylactic surgeries without knowing if they carried the gene or not because they were afraid of the discrimination that might result if they were found to be gene carriers. I have met women who have paid large sums of money out of pocket for medical tests, even though they had insurance, because of their fear of discrimination from their insurance companies. I have met women who are afraid to share their health history with their employer because of their fear of genetic discrimination. I even met a woman who was told by her insurance company that they would pay for genetic testing only if she agreed to have prophylactic surgeries should her results be positive.

Dear Members of the SACGHS, I plead with you to use your influence and power from the committee to assure the passage of the Genetic Information Nondiscrimination Act in the House of Representatives. The fear of genetic discrimination had touched mine and many others' lives. In order to receive the best healthcare, we must all have the freedom to share openly and honestly our healthcare needs and concerns without the fear of discrimination. Please feel free to contact me if you would like additional information. Thank you for your time and attention.

Sincerely,
Celia A. Boyne-Schuh

Sarata, Amanda (NIH/OD)

From: Mary A. Brown [mailto:mary.brown@nihs.nih.gov]
Sent: Saturday, August 21, 2004 12:27 PM
To: Sarata, Amanda (NIH/OD)
Subject: genetic discrimination and TSC

Dear Amanda,

We experience discrimination in a different form than what I read in Vicki's statement. We had to file due process against Pandora-Gilboa Schools in Pandora, Ohio because the school refused to provide services for our daughter. They had mainstreamed the special needs children without appropriate supports or providing a continuum of services. During our hearing the school's attorney, John Britton, made the statement that our daughter's disability was due to a genetic condition therefore our request for services did not apply. This has bothered me since I heard such an uneducated statement from an attorney for Ohio School Boards.

*Thank you,
Mary A. Brown*

Sarata, Amanda (NIH/OD)

From: Lynda Carrion [REDACTED]
Sent: Friday, September 24, 2004 4:09 PM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic Discrimination

I can answer yes to the following:

Fear the potential impact of genetic discrimination on either health insurance or employment.

Have paid out-of-pocket for services to exclude genetic information from medical records.

I saw a neurologist who kept the word Huntington's out of my file and also urged me to pay out of pocket for the predictive test. I paid cash for the visits and the predictive test. Since my results were positive, I fear genetic discrimination may affect my long term disability insurance through my employer.

Let me know if you need any more info!
Lynda Carrion

Sarata, Amanda (NIH/OD)

From: [REDACTED]
Sent: Friday, August 20, 2004 9:45 AM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic discrimination

Attention Amanda Sarata

I have lost interviews due to my condition H E D (I barely perspire and have few teeth. I can't afford the necessary surgeries to correct my jaw deformity. and the peice of implants are roughly 1,000.00 each. So I have a bad set of teeth. I have been asked in interviews if I play Hockey. I can't always go on management sport functions if it is too hot. I have been accused of not being a team player due to this. People generally show disbelief when I mention my condtion. Sadly Even my father (when I was young) said I wouldn't be a real man when I grew up. This isn't easy to write about. If I remember any more incidents I will e-mail you.

Sincerely,

Jay Daniels

Sarata, Amanda (NIH/OD)

From: Laila Gillam [mailto:lgillam@nih.gov]
Sent: Thursday, August 26, 2004 12:21 AM
To: Sarata, Amanda (NIH/OD)
Subject: genetic discrimination

Hi,

My son has experienced genetic discrimination with regard to obtaining life insurance. He is 2 years old and in good health, but he probably has a genetic condition called Freeman-Sheldon Syndrome. We are not able to test him for it because the genes involved haven't yet been found.

I applied for life insurance for both of my children, but my son's application was denied. It was not on the basis of his health (which is good), but on the basis of his probable diagnosis. There is no evidence that someone with this condition has a shorter lifespan than average. He was denied simply because he has a genetic condition. In my opinion, the insurance company should have been required to at least give him a physical before determining his insurability.

I hope our experience is helpful to you.

Sincerely,
Laila Gillam

September 28, 2004

Rejections for Insurance

Shortly before I was married, my father was diagnosed with Huntington's Disease. Yes it was a shock and yes I was upset, but it wasn't until several years later that I "knew" what Huntington's Disease was. I was told it was genetic and I could be tested, but I was young and strong and in love and life wouldn't do that to me. After I watched my Dad disintegrate quickly over a short few years, it hit me. This could happen to me. But now "I" was "We".

I had been blessed with a beautiful daughter and if I had it, not only would I probably not be able to raise my daughter, but she could also get HD from me. I looked into getting tested and also into insurance, life insurance in particular, because my dad advised me to. He was suddenly stuck with the coverage he had before he was diagnosed.

Also my dad had purchased a car and had bought the insurance that if you die or become disabled the payment would be made for you. After he went on disability at work, he put in the collect for the car and they denied him because HD was genetic (implying he knew about it and bought the car and the insurance knowing that it would be paid off shortly and it was purchased too close to him being diagnosed. They did however reimburse the amount paid for the insurance.

I didn't want the same thing to happen to me. I didn't want the assurance of an insurance policy and then in the event that I was positive, the company coming back to my family and saying, sorry HD is genetic and it doesn't count, but we'll give you back the premiums that you've paid.

So, I was up-front when applying and stated on all of the applications that I was at risk of HD. The insurance agent that was already insuring our cars, house, and business submitted it to the several companies he did business with and came back to me saying that none of them would insure me until I was tested for HD and it came back negative.

I then went through a friend that was an agent and he had similar responses from some of the companies he worked with, but, one of them did come back with a proposal. They would cover me but with an increased premium, about 6 times the amount I would have normally paid.

I agreed for lack of anything better and cancelled the policy when my Test results came back negative.

Because of MY difficulty obtaining a policy I was also concerned about how hard it would be for my daughter to get on too. If I had problems when I was at risk after my dad had already tested positive, then if I had happened to be positive, my daughter would suddenly be at risk as well. Because of this I secured a policy for her Before my testing, making sure that she could raise it every so many years without a physical or any questions being asked.

I understand that I was an unusually high risk for the insurance

companies to consider. But I had always thought that was what insurance was for. The well pay for the not so well, the living pay for the dying, and some live longer than others. The risk is part of the business. Apparently I was misled in my thinking because only the confirmed well can get the insurance and the possibly dying can be denied coverage.

Sincerely,

Joeline Hollar (Hardt)

Sarata, Amanda (NIH/OD)

From: [REDACTED]
Sent: Friday, August 20, 2004 6:27 PM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic Discrimination

Dear Amanda Sarata:

I am a parent of a child that has Hypohydroic Ectodermal Dysplasia genetic disorder. I am writing this letter in hopes to support and document the genetic discrimination my son has received with the medical and dental insurance companies.

I understand that President Bush has called for Federal legislation to prohibit genetic discrimination in health insurance and employment, but there is current opposition and this is preventing further progress in the House. I understand it is considered that there is insufficient evidence that genetic discrimination is occurring and thus legislation is not warranted at this time.

This letter will support President Bush's call for legislation as I know from direct experience that there is health insurance genetic discrimination. My son is in high school, therefore I cannot speak for the employment discrimination but I will explain my experiences throughout Sean's years of growing up.

Sean was diagnosed with Hypohydroic Ectodermal Dysplasia in 1987, when he was 13 months old. When he turned approximately 2 years old, we started dental work with him. Our first experience with dental insurance rejection occurred when we applied for Sean's dentures. Each step of the way, through the next 15 years, Sean has tried to receive dental or medical insurance but has been rejected stating, "It doesn't matter that he has a genetic disorder and he has missing teeth. Our policy does not cover dentures, partials, crowns, or implants with preexisting missing teeth." Unfortunately, these missing or deformed teeth were due to his genetic condition and not due to poor and irresponsible oral hygiene. This treatment becomes a "medically necessary" concern and should be reviewed as such.

What is most important to consider for these young children and young adults, is that with deformed, pointed or missing teeth, a person's diet is inhibited causing medical problems and this person's self-esteem and well-being are affected? By supporting dental or medical coverage for a genetic disorder, you will eliminate medical and psychological treatment coverage's for this individual.

Also, the genetic discrimination goes beyond the dental arena. Many of these children and adults do not have sweat glands, therefore they need air conditioning in the schools and workplaces. Many schools do not have air conditioning and the medical insurance programs will not support the cost to accommodate this medically necessary treatment for their genetic disorder.

Hypohydroic Ectodermal Dsyplasia is just one of many genetic disorders. I hope my letter will explain the need for nondiscrimination health insurance coverage and employment. Genetic discrimination definitely occurs and it is very frustrating we cannot get health insurance coverage for a genetic disorder, particularly relating to dental treatment.

Pam Kennedy

[REDACTED]
[REDACTED]
St. Petersburg, FL 33701
[REDACTED]

Sarata, Amanda (NIH/OD)

From: McCarty, Terence L [REDACTED]
Sent: Thursday, September 23, 2004 4:46 PM
To: Sarata, Amanda (NIH/OD)
Subject: Re Genetic Discrimination

My wife was diagnosed with Huntington's Disease approximately ten years ago. She is in the middle stages of this incurable disease. Some of the problems we faced with insurance coverage was that if we treated just the symptoms and did not give a title of "Huntington's Disease" to it, the insurance company covers it under major medical. If however she needs to see a psychiatrist, in addition to our family doctor and her neurologist, it is billed differently under psychological care. The insurance company has a much lower "cap" on providing in-patient or out-patient psychiatric care. I have tried to explain that her condition is genetic and affects all of her being, not just psychological, but to no avail. So with the doctor's assistance, we try to submit claims that avoid any psychological reference.

Also, Huntington's Disease is one disease that automatically disqualifies the person with it for any long term care insurance. There is no company that will provide that insurance, even if one was to pay into the policy for decades before it is needed. My financial future looks bleak as she will eventually need admittance to costly nursing homes. Once I deplete our savings, the equity in our home and whatever other assets we might have, I might be able to qualify her for Medicare. But my fear is, what do I spend to support me and my living needs?

And if you're looking into other areas, if I needed to put her in a nursing home this week at age 47, I cannot find a single nursing home that will accept a Huntington's Disease patient at her age. Apparently there are laws that require the nursing home to provide programs, etc. to younger patients and apparently the average age in a nursing home is 70+ so no one wants to assume the responsibility for a younger and totally disabled patient. There is an organization online, www.aplaceformom.com which assists in locating nursing homes. After four months, I was informed that they couldn't find anyplace in Southern California that would accept my wife if I needed a nursing home right now. She will not live to be 60 or 70, that's for certain.

I have two children, both married, who have a 50% chance of inheriting this godawful disease from their mom. If it is too late to help my wife, then we need to protect the kids.

Thanks.

Mr. Terry McCarty
Lakewood, California

Sarata, Amanda (NIH/OD)

From: Rick and Sandra McElwee [redacted]

Sent: Monday, August 30, 2004 12:13 PM

To: Sarata, Amanda (NIH/OD)

Subject: Genetic Discrimination

What about prenatal testing designed to discriminate against those with genetic 'imperfections' in the spirit of eliminating them?

Sandra McElwee

Sarata, Amanda (NIH/OD)

From: Beth Pletcher [REDACTED]
Sent: Tuesday, August 31, 2004 1:12 PM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic Discrimination

Although I will not be able to attend the meeting, I thought I would share with you my own personal experience with genetic testing. Ironic I suppose since I am a practicing clinical geneticist. Because of a strong family history of premenopausal breast cancer on my Dad's side of the family, I asked my 24 year old cousin with breast cancer to undergo BRCA1 and 2 sequencing on my behalf about three years ago. Because her insurance only covered 80% of the cost, her sister and I paid the difference, since this was for our own medical information. When a point mutation in BRCA1 was identified, I chose to have mutation testing for a much lower fee - \$295.00 at the time. Because of some concerns on my part about the potential impact of these results, not on my health insurance, but rather life and/or disability coverage, I decided to pay out of pocket for the testing. When the test results showed that I did not carry the family mutation, I submitted the bill to my insurance carrier for reimbursement. They rejected my claim stating that this was "routine screening" not covered under my traditional health plan. Clearly they were wrong...I was most concerned since I had been working with this very same insurer to develop a breast/ovarian cancer screening policy in the year prior to my testing. Nevertheless, I filed a complaint and the testing was eventually covered. Although I had some individual concerns about this testing and my future insurability, I have more concerns about patient access to coverage for molecular based genetic testing. More than 3/4 of my patients cannot get DNA based genetic testing done because of overwhelming insurance obstacles and the sheer expense of such testing. Patient in HMOs or even PPOs usually are required to use a single large commercial laboratory; these laboratories do not do molecular testing for the vast majority of genetic conditions under consideration and will not send out to the specialty labs that do such testing. Examples would be Myriad Labs for cancer genetic testing, Athena for neuromuscular disorders, U of Chicago or Baylor or Univ of PA for specific disorders - CMT/lissencephaly/von Hippel-Lindau etc....Genetic discrimination is less of an issue today perhaps because of the difficulty in getting appropriate testing for patients who do not have insurance, are enrolled in HMOs or do not have the financial resource to pay for this testing. Patients can get MRIs when indicated, but not molecular genetic testing. This disproportionately impacts patients who are poor, uninsured or under-insured. Until there is a system in place to ensure that all patients can have access to appropriate genetic tests, the issue of genetic discrimination will only be the tip of the proverbial iceberg. Thanks. Beth A. Pletcher, MD, FAAP, FACMG

Sarata, Amanda (NIH/OD)

From: ██████████
Sent: Wednesday, September 01, 2004 11:32 AM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic Discrimination

Ms. Sarata,

I was informed that you are looking for public opinion on genetic discrimination. My son, was a victim of such discrimination, all his life.

He was born 2 months premature, to a birthmother who had no prenatal care. He was born with Down syndrome and a heart condition called TOF (Tetralogy of Fallot). My husband and I brought him into our home at 6 weeks old. Two weeks later, he had a medical crisis and had oxygen sats in the teens for over 7 hours. A "normal" person has sats of 99 or 100%. Alex survived, but doctors said that he had no brain function. They never looked at him, only at papers with test results on them. They said, there was no way, he could have any brain function left. The adoption agency took him out of our care, "for our own good", saying he would destroy our family and our marriage.

Alex was sent to an institution to wait to die. A feeding tube was inserted, because doctors said he would never eat normally. They said he was blind, deaf, had seizures and would be dead in six months. Fortunately for Alex, he could not read their predictions. My husband and I fought and 11 months later, brought our son home and adopted him. We found that he was neither blind nor deaf. He had never had a seizure and he began to eat and drink by mouth.

But, we would still come face to face with doctors who would say things like "He'd be better off dead" or "We didn't do him a favor by saving him". His cardiologist said that he would someday need a heart/lung transplant, but he would never get one because of having Down syndrome. But, Alex continued to thrive and by age four he was walking and talking and eating like a child with Down syndrome. Not perfectly, but much more than anyone ever predicted. At age four, he was in his last year of preschool, one with children like himself, and one with his "normal" peers.

In the spring of 2000, his cardiologist told us that he was doing so well, that he did not need to be seen for a year. (He had normally been seen every three months). That winter he was having recurring sinus problems and his pediatrician prescribed a sedated CT scan. We took him to the hospital, where a child with his history should have had a one on one nurse. There should have been resuscitation equipment in his room, according to hospital procedures. My son, who at age four weighed about 30 pounds, was given twice the adult dose of sedation, because his nurse didn't bother to double check it and have someone sign off on it. (Again hospital procedure). After the CT scan, he was put on a heart monitor, which I later found out the nurse had turned the alarm off, because the noise it made annoyed the nurse! My son was left alone and not checked on (in person) all afternoon. He was only monitored by the monitor at the nurses station. At 5 PM. Four hours after the CT scan, he flatlined. I had to alert someone, as no one was watching his monitor at the nurses station. When someone came, the proper equipment was not there to perform CPR. The crash cart across the hall was ignored, and one from two floors away was called. My son was revived, but the damage was done, and he was pronounced brain dead two days later on 12/31/00.


The hospital quickly decided to try to blame me for his death, because I was the one that found him. A lawyer spent 30 minutes with his file and discovered the truth. The hospital was sued and we settled. But, they still maintained that his life was worth nothing, and they had actually "done him a favor." His cardiologist, who had released him from treatment for a year, suddenly said, he had been dying and a transplant would have been his only hope. He never told us because he knew we would never get one.

Our family was devastated by the loss of our son, and we are even more worried, because our other three children, also have disabilities. We worry about genetic testing on fetus' because many parents would be pressured to end the child's life, before they are even born. Or if the parent decides to have the child, that insurance companies would refuse to cover that child. I could tell you hundreds of stories about battles we have

fought with the insurance company to get services for our children with disabilities.

I know this letter was long. I am sorry. I just wanted to make sure to get everything in. The future of my children, and all children like them, rests in the governments hands. I pray that you will choose to see these children as people first, instead of disabilities. They are people, like anyone else, and deserve to be treated with the same dignity and respect.

Thank you,

Sue Saladino


Sarata, Amanda (NIH/OD)

From: Wetexas4@aol.com
Sent: Friday, August 20, 2004 5:49 PM
To: Sarata, Amanda (NIH/OD)
Cc: Wetexas4@aol.com
Subject: Re: Genetic Discrimination Stories Needed for Hearing

Dear Ms. Sarata,

My son Christian has Ectodermal Dysplasia as well as asthma. Our health insurance was wonderfully inexpensive until he was diagnosed. We worked for a smaller company that was unable to offset the "cost" of his care and everyone's rates went up. My husband's employer was a good man who looked out for our best interests as well as those of his company, but in 3 years we've been with 3 different insurance companies. Our rates for everything keep climbing. The irony, to me, is that after much petitioning for coverage of my son's dentures under our medical policy and being denied repeatedly, he is the healthiest of our bunch! Baylor Medical school has taken up his oral care and his asthma is completely under control. He requires an allergy prescription and that's it. Unfortunately, we look terrible "on paper". My husband has changed jobs to a larger company, but I am afraid this will plague us forever for something completely out of our control to fix. There should never be discrimination against anyone for a genetic condition. This is wrong. Thank you for your efforts on our behalf.

Sincerely,
Stephanie Schmidt

Sarata, Amanda (NIH/OD)

From: Renee Shean [REDACTED]
Sent: Wednesday, August 25, 2004 8:59 PM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic Discrimination

Story of Discrimination

I am now 43 and was born with a rare genetic disorder called Freeman Sheldon Syndrome. It can affect many areas of the body, but generally the hands, feet, and facial features are the most common and most noticeable. By outward appearance, I had a somewhat mild case, which affected my hands and face. My differences never caused me any physical pain or kept me from performing everyday activities and otherwise living a "normal" life. However, others reactions and perceptions of me often made life very challenging. The following story is an example of one of those challenges where I was discriminated against many years ago when I applied to a medical assisting school.

I was 19 and never really thought I was smart enough to go to a 4yr college, so I thought this 6mo medical assisting course sounded like a great way to learn a skill and get a job. When I went to the school the Representative was very excited as she showed me around the school and convinced me to sign up for a student loan. There was no mention of concern or doubt in my ability to handle the course, that is until the first day of class.

I was called out of class within the first hour of the first day, to go and speak to the Director of the school. He was a very large intimidating older gentleman and I was a quite petite, and very naive 19yr old. He proceeded to tell me that he thought it would be best if I dropped out of the school because he was sure that I would have too much trouble learning and performing all the necessary skills in order to graduate, and he just didn't want to see me get hurt, so he was just trying to help me. Of course I was in shock, I just thought, "doesn't want me to get hurt, it's a little late for that now". Surprisingly enough, this was probably the first time I had ever really had someone tell me they didn't think I could do something. I had always been raised that I could accomplish anything I set my mind to, I may not do it exactly the same way everyone else does, but the end result was the same. So I tried to explain that to him, while holding back tears, and he told me I should just go home and think it over very carefully. So I went home, and in tears, I told my mom what had happened. At this point I was devastated and not sure if I even wanted to go to the school any more, but my mom was determined that they were not going to get away with this. She called her lawyer, who then called the school, and before the day was over the Director was calling my mom and apologizing all over the place for the "misunderstanding". Since I was so upset by the whole thing they agreed to let me wait and start the following month.

However, all was not forgotten, on their part anyway. Once I started, for the second time, I was again called in on my first day and told that they wanted me to sign a waiver saying that the school would not have to guarantee job placement upon graduation, something they did for everyone else. They said that with my, "problems" as they called it, they couldn't be sure that they would be able to find me a job. O.K., at this point I was no longer upset, now I was mad. Obviously I could have called my lawyer again and fought them, but I at that point I just wanted to go to school, and since, up until then, I had always been able to get whatever I wanted, I figured I could find my own job without them.

So I finally started class, and graduated with the highest score, but it was not without a lot of extra work, literally. What I mean is, apparently the Director had told at least one of the instructor's to make sure that I performed every check-off (or skill) at least twice before I was allowed to move on, (I guess, hoping they could make me quit). At first I thought I was just imagining it, but then it became obvious. So much so, that the other students were even coming up to me and asking why I was having to do everything so many times when it was obvious that it was ok the first time, I just acted like I didn't know. I was determined that they were not going to get to me, so I stuck it out, down to the last day.

The day before graduation I still had one check-off left that she had made me re-do at least 4 times, and then I still had to take the final exam. So at the end of the day, she still refused to check me off on the last skill, and told me that I would have to come back in the morning try and check off and then take the exam, and if I passed then I could graduate.

So I was there bright and early and I guess at that point she just couldn't justify failing me one more time, so she let me check off, and then handed me my final exam, which was over the whole six month course. Of course there was no way I had been able to study for this, but all I could do was try. So after the exam I waited to see if I had passed. Just before the ceremony she came in to say it was almost time, but said nothing about my score. Someone else spoke up and asked, "What about Renee?" "Oh," she whispered, "she got a 98." Of course everyone else congratulated me, but she just turned and walked away.

Ultimately I never really got a job as a medical assistant, not because I couldn't find one, but because I realized I was a lot smarter than I thought and decided I did want to go to college after all. I did however, do my one-month externship in a general surgeon's office, where I was able to perform all my tasks just fine, and even assisted in a minor surgery to remove a cyst.

P.S. I did go on to college where I received an A.A. in Business Administration, a B.S. in Marketing, and eventually a Teaching Credential in Elementary Education.

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CONGRESS OF THE UNITED STATES
LOUISE M. SLAUGHTER
28TH DISTRICT, NEW YORK

September 3, 2004

COMMITTEE ON RULES
SUBCOMMITTEE ON
LEGISLATIVE AND BUDGET PROCESS
SELECT COMMITTEE ON
HOMELAND SECURITY
CONGRESSIONAL CAUCUS FOR
WOMEN'S ISSUES
DEMOCRATIC CO-CHAIR
CONGRESSIONAL ARTS CAUCUS
DEMOCRATIC CO-CHAIR
COMMITTEE ON ORGANIZATION,
STUDY, AND REVIEW
WHIP PARLIAMENTARY GROUP
COMMISSION ON SECURITY AND
COOPERATION IN EUROPE

Secretary's Advisory Committee on Genetics, Health, and Society
Office of Biotechnology Activities
National Institutes of Health
6705 Rockledge Drive, Suite 750, MSC 7985
Bethesda, Maryland 20892-7985

Dear Members of the Secretary's Advisory Committee on Genetics, Health, and Society,

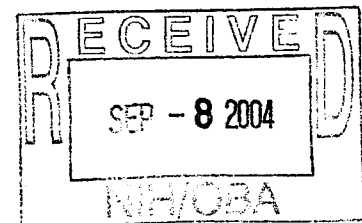
I appreciate this opportunity to share my concerns as part of the October 18 hearing on genetic discrimination. I hope very much that the evidence and testimony gathered during this hearing will lead to speedy and decisive passage of strong genetic nondiscrimination legislation in Congress.

Ten years have now passed since the world was electrified by the discovery of the first genetic mutation linked to breast cancer in 1994. In that short decade, scores more genetic links to disease have been identified, dozens of genetic tests have become commercially available, and genetic technology has become firmly embedded in the practice of medicine.

As technology has raced ahead, ethical, legal, and social challenges have presented themselves. We are now faced with critical questions about how we, as a nation, will allow genetic information to be handled and used.

Almost nine years ago, I introduced the first legislation in Congress to ban genetic discrimination in health insurance. I considered the bill to be a simple, straightforward, noncontroversial proposal that would pass easily. I could hardly have imagined that six years would pass before the House held the first hearing on the issue, and far more than that without any meaningful action at all. At this point, it looks all but certain that the 108th Congress will also adjourn without acting on genetic discrimination, postponing this issue for another year when time is already short.

Genetics – A Primer



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No human being has a perfect set of genes. In fact, every one of us is estimated to be genetically predisposed to between 5 and 50 serious disorders. Every person is therefore a potential victim of genetic discrimination.

Simply carrying a given genetic mutation almost never guarantees that one will fall ill, however. A genetic flaw simply confers a level of risk upon the carrier. Today, with our knowledge of genetics still in its infancy, scientists have only a rudimentary understanding of how much additional risk a genetic mutation may carry. We have virtually no understanding of how environmental factors – such as diet, smoking, and exposure to chemicals or radiation – interact with genetics to cause disease.

Given that scientists cannot accurately predict when or whether a carrier will develop a genetic disorder, it seems ludicrous to allow this information to be used by health insurers or employers to discriminate. An insurance bureaucrat or human resources professional would be as accurate with a dartboard as with a genetic test result in predicting who will get sick.

Genetic Discrimination – Cases and Fears

Some have called the legislation in Congress “a solution in search of a problem” because they state that genetic discrimination is rare, if it happens at all. Unfortunately, genetic discrimination is occurring:

- In 2000, the Burlington Northern Santa Fe Railroad performed genetic tests on employees without their knowledge or consent. The workers involved had applied for workers compensation, and the tests were being done in hopes of undermining their claims.
- A Kentucky family was denied health insurance for their children because they were known to be carriers for a genetic disorder – even though they did not have the two copies of the mutation required to become ill.
- A North Carolina woman was fired after a genetic test revealed her risk for a lung disorder and she began the treatments that would keep her healthy.
- In the early 1990s, Lawrence Livermore Laboratories in Berkeley was found to have been performing genetic tests on employees without their knowledge or consent for years.
- In the 1970s, many African Americans were denied jobs and insurance based on their carrier status for sickle cell anemia – again, despite the fact that a carrier lacks the two copies of a mutation necessary to get sick.

It is imperative that Congress stop this practice before it becomes widespread. Moreover, the fear of genetic discrimination is playing a major role in many patients' decisions about whether to take a genetic test or participate in genetic research. A survey of 159 genetic counselors found that 108, or 68%, would not seek insurance reimbursement for a genetic test for breast or colon cancer due to the fear of discrimination. Sixty percent would not share the information with their colleagues due to the fear of discrimination in the workplace. Several other studies have shown that the

fear of discrimination plays a significant role in decisions about whether to take a genetic test, whether to do it under one's own name, paying out of pocket versus seeking insurance reimbursement, and with whom the information would be shared, including health care providers, coworkers, and family members.

House Legislation, H.R. 1910

I am proud to be the author of H.R. 1910, the Genetic Information Nondiscrimination in Health Insurance and Employment Act. This legislation currently has the support of 242 bipartisan Members of Congress and has been endorsed by over 300 organizations that care about health issues. Despite this broad support and an aggressive grassroots campaign, the House has taken no action on H.R. 1910. In July, the Education and the Workforce Subcommittee on Employer-Employee relations held a single perfunctory hearing on the issue of genetic discrimination. Afterwards, the chairman was quoted in the media as saying no further action would take place due to legislative gridlock in the House of Representatives. This represents nothing more than a simple lack of political will on the part of committee and subcommittee chairmen.

In April, an article in *Congress Daily AM* described the lack of action on this legislation as "a textbook case of obstruction by inertia." The article also identified the U.S. Chamber of Commerce as the primary interest group lobbying Congress not to take up this bill. Unfortunately, this organization continues its misguided opposition to this bill, seeking to deny to tens of millions of Americans a shield against genetic discrimination.

Senate Action

Throughout the first half of the 108th Congress, a group of committed Senators came together to negotiate a compromise genetic nondiscrimination bill. Under the leadership of Democratic Leader Tom Daschle, Majority Leader Bill Frist, Health Committee Chairman Judd Gregg, Health Committee Ranking Member Edward Kennedy, Senators Olympia Snowe and Tom Harkin, among others, the Senate produced a mutually agreed-upon version of the legislation. In October 2003, the Senate passed S. 1053, the Genetic Information Nondiscrimination Act, by a unanimous 95-0 vote. The White House issued a Statement of Administration Policy expressing its support for this legislation.

Here in the House, S. 1053 has not even been referred to committee. Instead, it has been "held at the desk" for the past year at the direction of the House leadership. This action ensured that it would be impossible to take up this bill from a parliamentary perspective even if a committee wishes to do so.

Myths About Genetic Discrimination Legislation

Those opposing S. 1053 and H.R. 1910 have made a number of arguments in opposition. I have reviewed these concerns at some length and would like to share my conclusions.

1. There is no evidence that employers or insurers are, in fact, engaging in discrimination based on genetic makeup.

Several cases have emerged where employers did indeed engage in genetic discrimination or attempted to do so.

Congress should not wait to act until hundreds or thousands of people have experienced genetic discrimination. Today, the opportunities for genetic discrimination are limited precisely because *people are not taking genetic tests for fear that this information will be used against them*. By doing so, however, they are denying themselves valuable information that they could use to make important health care decisions.

2. Genetic information can be useful in making some employment decisions. For example, a health condition likely to cause seizures could properly be considered a threat to others if the employee were a bus driver or an airline pilot.

Scientists and geneticists have been unable to identify any existing genetic test that would guarantee that a person would develop a condition that would pose a significant danger to others. A genetic mutation only confers a higher risk of developing a disorder; it is not a guarantee. Moreover, few such conditions develop in adulthood suddenly or without warning. Should such a genetic test exist in the future, however, the legislation passed by the Senate would permit employers to test workers and make decisions in accordance with any guidelines produced by OSHA.

Expecting a human resources professional to interpret a genetic test accurately is about as realistic as asking them to predict the weather for October 2009. The vast majority of genetic tests have no bearing whatsoever on an individual's ability to perform the duties of his or her job today. Employers should not be permitted to deny job opportunities to entire categories of workers on the theory that a person might get sick someday.

3. It's too difficult for employers to comply with 50 different state laws. If Congress enacts legislation barring employment discrimination based on genetic information then it should include a safe harbor providing that employers in compliance with the federal standards cannot be liable under state or local laws banning such discrimination.

A federal law can provide valuable uniformity, but it does not have to trample states' rights in the process. At present, over 30 states have passed laws dealing with some aspect of genetic discrimination, but they are a patchwork of different definitions, standards, and remedies. A federal "floor" would provide a coherent national statement of policy while allowing states to pass additional protections for their residents

if they so chose. This is the same model followed by civil rights laws, the Health Insurance Portability and Accountability Act (HIPAA), and numerous others.

Congress has a long history of avoiding state pre-emption whenever possible in deference to states' rights. If a given state wishes to be more explicit or extensive in banning genetic discrimination, it should have the right to do so.

4. It makes sense to allow a genetic nondiscrimination law to expire. Any federal legislation should include a sunset date at which time Congress can decide how effectively the law has worked and whether it should be reauthorized.

Congress routinely uses its committee oversight and hearing processes to examine whether existing laws need to be updated or changed. A sunset could only create a dangerous situation where the law would lapse and genetic discrimination would become legal after a period of being banned.

No major law protecting Americans' rights has ever contained a sunset – including the Americans with Disabilities Act, the Civil Rights Act of 1964, or the Health Insurance Portability and Accountability Act. There is no reason why genetic discrimination should be banned only temporarily.

5. A genetic nondiscrimination law can be effective if it only protects the genetic information of immediate relatives. Genetic discrimination should only be illegal if the employer has direct knowledge of the history of an immediate family member related by blood, not more distant relatives.

If an employer engages in genetic discrimination, it should not matter how close or distant the blood relationship is. The legislation before Congress does not penalize employers from coming into possession of family medical history or other genetic information inadvertently. It does, however, prohibit the employer from using that information to discriminate. If all genetic information is not protected, Congress could create a perverse loophole that would allow employers to discriminate based on the genetic mutations of distant relatives, but not close ones.

The Senate-passed bill would not outlaw a casual workplace conversation where someone mentions that an uncle or cousin died of cancer. But it would bar employers from using that information in decisions about hiring, firing, promotions, and other job-related benefits.

6. Only actual genetic tests should be protected. A bill should focus on employment discrimination based on genetic tests, not family history.

There is no reason to allow employers to discriminate based on an individual's family medical history. A healthy worker should not be denied jobs or opportunities based on a relative's health status. The fact that a person's parent, cousin, or great-uncle died of cancer or Alzheimer's should be irrelevant to an employer.

As stated above, the bills before Congress would not outlaw a casual workplace conversation about a relative's illness. But employers would be prohibited from using that information in determining the terms and conditions of employment.

Conclusion

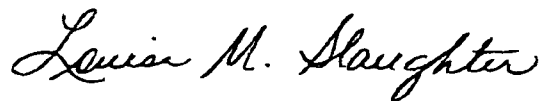
Congressional action on genetic discrimination is long overdue. The House of Representatives is doing a tragic disservice to the people its Members are sworn to serve by allowing this practice to continue. I appreciate your effort to bring attention to this issue by holding this hearing. I hope very much that your efforts will spur Congress to act quickly.

The American people desperately want these protections in federal law. The Genetics & Public Policy Center at Johns Hopkins University recently released a set of surveys on the public's views about the privacy of genetic information. In 2002, 85% of those surveyed did not want employers to have access to their genetic information. By 2004, that number had risen to 92%. In 2002, 68% of those surveyed said their genetic information should be kept private from health insurers; by 2004, it had increased to 80%. Clearly, overwhelming majorities wish to keep this information out of the hands of insurers and employers, who may use it to undermine rather than advance an individual's best interests.

The arguments against this legislation are no more than delaying tactics. Action is already long overdue. The Senate has passed S. 1053 unanimously; the House of Representatives should follow suit as quickly as possible.

I look forward to working with the members of this committee to protect all Americans against genetic discrimination in health insurance and employment. Again, thank you for this opportunity to add my voice to the millions of others calling for action on genetic discrimination.

Sincerely,



Louise M. Slaughter
Member of Congress

Sarata, Amanda (NIH/OD)

From: Jennifer Stephens [redacted]
Sent: Thursday, August 19, 2004 4:47 PM
To: Sarata, Amanda (NIH/OD)
Cc: info@nfed.org
Subject: Genetic discrimination legislation

NFED (National Foundation for Ectodermal Dysplasias) suggested that I contact you regarding the effect of genetic discrimination on our family, due to the pending legislation on genetic discrimination. We strongly support the legislation that President Bush has proposed that would eliminate or limit the effects of genetic discrimination.

My 3 year old son, Cooper Stephens, has Ectodermal Dysplasia (ED), which is a genetic disorder affecting the outer layer of the skin. My twin 18 month old sons, Ben and Sam, are carriers of the disorder. The effects of ED include severely deformed and/or missing dentition, hair, skin and nail problems, jaw malformations and a reduced ability to sweat. (Information on the disorder may be found at www.nfed.org) Though my sons are young and Cooper is just beginning his treatment, we have already experienced genetic discrimination and fear that it will only worsen.

Ectodermal Dysplasia is a horrifically expensive condition to treat...in Cooper's case, estimates of the out of pocket cost of his treatment run as high as \$75,000 before his 18th birthday, with more to come in adulthood. We have already been told that insurers in our state (and, according to NFED, in nearly all 50 states!) are able to refuse us coverage due to the fact that treatment for the disorder focuses primarily on dental issues. Insurance companies are essentially "separating the mouth from the body" and refuse to cover treatment under medical policies which insure coverage for other genetic anomalies.

Basically, insurance laws in all 50 states do not require insurers to recognize that this is a genetic condition with whole-body effects, with the end result being that these children may seek coverage only under dental policies. Dental policies generally do not cover the extensive surgeries, prosthetics and orthodontics required to treat these children...they generally cover only basic preventative care for "sound, naturally occurring teeth." So, the end result is that children with ED are discriminated against as a group and must pay for treatment out of pocket or suffer the jaw disintegration, nutritional issues, speech issues and social ostracism that result from going without treatment.

Families who fight for insurance coverage must do so over and over again each time they switch insurers, often unsuccessfully. Each time a family takes on a new job with new insurance, the possibility also exists that ED will be considered a "pre-existing condition" and treatment refused. Lobbying for change at the state level is costly and time consuming. Something must change!

My hope is that legislation preventing insurers from discriminating against genetic disorders will force them not only to cover ED treatment under medical policies, where it rightly belongs, but also to prevent it from being categorized as a pre-existing condition. This would prevent my child, as well as generations of others, from having to choose between paying for his treatment, or paying for his college tuition!

Thanks for your help with this.

Jennifer Stephens
[redacted]
Richardson, TX 75082
[redacted]

Sarata, Amanda (NIH/OD)

From: Kari Stubbs [REDACTED]
Sent: Tuesday, September 14, 2004 10:42 AM
To: Sarata, Amanda (NIH/OD)
Cc: Kari Stubbs
Subject: genetic discrimination

I am sending you this email as part of your information gathering process for the hearing on September 18th. It is an issue that is near and dear to me. Please consider me for "in person" testimony as well as follow up information-gathering.

I am 35 years old. My mother discovered that she had breast cancer at the young age of 44. Although detected when it was only a small lump, the cancer had already spread to her lymph nodes in her arm. She underwent a mastectomy, and followed that with aggressive chemo and radiation. The year was 1987, and I was a senior in High School.

Fortunately, my mother survived. Her doctors watched her carefully, and she was able to watch her children complete college and marry. On her 9-year well check, the cancer had returned. Sadly, this time it appeared in the bone adjacent to the breast where it had started initially. Again, we were fortunate to have qualified doctors guiding us through the process, and an unrelenting positive attitude on the part of my parents. My mother participated in the Stem Cell harvest and transplant process at the University of Omaha. Again, she survived. She welcomed her first grandbaby, my son, into the world five months later. (She had been lifting canned vegetables to build up her strength, so that she would be strong enough to hold him.) :) More grandchildren were to follow.

A few years later, her spine began to dissolve. Chemo was started again, as well as some of the more aggressive new drug therapies. Eventually, the cancer won. I lost my mom in April of 2001, fifteen years after the onset of cancer.

I tell you this background because it is important for you to know. I am a thirty-five year old female; a single parent with two young children. I have taught third grade for eleven years, and just took a leave of absence to pursue a PhD with an emphasis in educational technology. This past year, I went through the process of getting genetic testing. Knowledge is power, and although a difficult decision, both my father and I agree that it would be better to know if I am a cancer gene carrier. My father is a person whom I respect above all others. He was a loving, supportive force behind my mother's cancer struggles. He fought insurance companies for the right to have her treated in Omaha. His insurance company repeatedly declared that she needed to use the local hospitals whose reputations were not as strong. He made sure that she had the best of care. He is continuing that legacy by watching out for me, his only daughter.

I absolutely could not have afforded to get the genetic testing done without my father's assistance. It is an expensive test, and one that is not covered by insurance plans. We were also both concerned about my age, and the potential

impacts that the testing might have per insurance companies. I did not want the fact that I had the test done, let alone the results, to impact my "insurability" in the future. We worked very hard with Amy Tranin in Overland Park, Kansas to pay cash for the testing and code my name and identity as it related to the results. I plan to "change the world" in the field of education, and will be changing jobs and insurance carriers repeatedly throughout the duration of my life. If I get cancer, I don't want to be robbed of the chance to get the best care because my cancer is determined to be a "pre-existing condition." It is a very real concern of mine.

I am sad to think that insurance companies have created an environment in which they often times prevent you from getting the diagnostic care that you need. Cancer is expensive: chemotherapy, doctor visits, lab work, drug therapy, loss of work. The list goes on and on. It seems to clear to me that by having genetic testing you would increase your ability to detect the cancer at an early phase, thus decreasing subsequent costs on the part of the insurance companies and individual patients. Again, knowledge is power. Science has made alarming progress in the field of cancer detection and treatment. Let's work to create an atmosphere where the insurance companies work with that scientific progress, not against it.

Kari Stubbs

9/22/04

To whom it may concern:

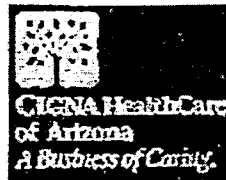
My family has had some discouraging experiences with genetic insurance discrimination. When you are self-employed, you don't have a whole lot of insurance options, but to purchase an individual plan. Our 4-year-old son was rejected from every insurance company that I called because of his inherited Hemophilia B. As a mother of an active preschool aged child, it is very distressing to know that because your son is unique and different, you will face extraordinarily high medical costs for care, unlike someone who have the option to be covered under a group insurance plan. The financial risk took a toll on us as we tried to take extra jobs, go on COBRA (which broke our bank and almost financially ruined us because it cost the same as our monthly rent payment), and even lowered our income so that he would qualify for state funded insurance.

Then as we were faced with yet another unfair genetic situation, the same risks resurfaced. As I learned my father had Huntington's Disease, a degenerative brain disorder (usually diagnosed in adulthood when symptoms occur), I learned that not only was I a carrier for Hemophilia (which was not my fault or choice), but that I had a 50% chance of inheriting Huntington's Disease as well (not my fault or choice). Because of my previous experience with "pre-existing condition exclusions", we found a way to go around the system and be tested anonymously. I could not risk the chance of losing my health coverage and becoming a financial burden to my husband and family, but I needed to know the results. I saw a genetic counselor that informed me about my options, and she agreed that anonymous testing would be possible. I then went to be tested with a fake name, social security number and address, and the cash to pay for the test. After the results were in, the "alias" name and results were returned to the counselor and she delivered our results.

It is a shame that we have to be sneaky and try to avoid the system that is supposed to be available to help and insure imperfect human beings and their families. No person on this earth will ever be free of some sort of physical ailment, therefore it is blatant discrimination on the part of insurance companies to deny coverage for an inherited disease or disorder that you had no control over inheriting. Obviously there is a risk to the insurance company financially to cover such individuals, but group insurance plans do it and thrive. There has to be another option.

Thank you for your time,
Michelle Thompson

February 12, 2002



ENOCH THOMPSON
[REDACTED]

TEMPE, AZ 85282

Dear Mr. THOMPSON:

We have reviewed the medically underwritten application you submitted for CIGNA Private Practice Plan of Arizona's Individual and Family Plan and regret to advise that membership has not been approved for:

Person:

ENOCH

Due to a history of:

HEMOPHILIA B FACTOR

If you would like this decision reconsidered, submit in writing your request and any additional information. For more details refer to the enclosed SUMMARY OF RIGHTS FOR ADVERSE UNDERWRITING DECISIONS. If more than one person was applying for coverage please notify us by calling the number below if you would like the application to be processed without the above person(s).

As CIGNA HealthCare previously advised you in the application materials, you may be eligible for coverage under our HIPAA Individual Portability Plan in the event you have lost employment-related group coverage in the last 63 days.

Thank you for your interest in CIGNA HealthCare of Arizona's Individual and Family Plan. We regret we are unable to assist you with your health care needs at this time.

Sincerely,

Medical Underwriting Department
CIGNA HealthCare of Arizona
11001 N. Black Canyon Hwy., Suite 300
Phoenix, AZ 85029
602.861.8200



**BlueCross
BlueShield
of Arizona**

An Independent Licensee of the Blue Cross and Blue Shield Association

MEDICAL RISK ASSESSMENT

Post Office Box 13466 Phoenix, AZ 85002-3466
2444 West Las Palmaritas Drive Phoenix, AZ 85021-4883

Thursday, February 28, 2002

ID#: 600-79-4311
DOB: 04/30/1998

ENOCH M THOMPSON
c/o ENOCH S THOMPSON

~~XXXXXXXXXXXXXXXXXXXX~~

TEMPE AZ 85282

Thank you for your application for individual health care coverage with Blue Cross Blue Shield of Arizona. As individual coverage products are medically underwritten, previous health history is reviewed to determine eligibility.

Our review status is as follows:

Status	Action
Regrettably, must decline <i>HEMOPHILLA (MEDICAL UNDERWRITING GUIDELINES REQUIRE DECLINATION OF COVERAGE.)</i>	

BCBSAZ will not issue any policy until Medical Risk Assessment has completed processing of all applicants unless you elect the Early Enrollment Option. See reverse for details. If this letter indicates BCBSAZ needs additional medical records for you, you are not eligible for the early coverage option as your application is still under review and a final eligibility decision has not yet been made.

Please note: Changes of any kind in any applicant's health history, including recent changes or new symptoms (pain, bleeding, etc.) that may be undiagnosed or untreated, from the date you first applied for coverage until the effective date of your coverage, must be reported to BCBSAZ. Failure to report such changes may result in rescission of the insurance contract rendering it null and void leaving you financially responsible for all medical expenses. Such changes must be reported for each applicant, even those previously approved by BCBSAZ who elect to wait for the completion of processing prior to beginning their coverage.

Requests for reconsideration concerning eligibility for coverage must be made in writing and contain adequate medical documentation to support your position. Physician's letters/narrative statements are insufficient. Adequate documentation should include hospital records, physician progress records, diagnostic studies, etc., as appropriate. Once reviewed, you will receive a written response from us.

See reverse side for additional information regarding Individual Portability coverage, definitions and early enrollment option for eligible individuals.

If you have any questions, please call 602-864-4040 or 1-800-232-2345, extension 4040.

Sincerely,

Medical Risk Assessment

Sarata, Amanda (NIH/OD)

From: samyong [yongs002@hawaii.rr.com]
Sent: Saturday, October 16, 2004 10:19 AM
To: Sarata, Amanda (NIH/OD)
Subject: genetic discrimination

I just saw your notice right now--Friday Oct. 15.It's 3:00pm in Hawaii. I'm not sure where you are, but due to your time cut-off I'm sending this over. Hope it is something you want.
(P.S. I worked on this for so long it's now 4:00 am Oct. 16)

I have been having trouble getting my husband and teenaged daughter tested for Huntington's Disease (HD); I call that a type of genetic discrimination. It's like reverse discrimination. Due to "protocols" that are supposedly "to protect " patients, I am having a very difficult time getting diagnostic tests for my husband and daughter.

Some labs and HD centers think you should go through genetic counseling to help you deal with the fact that you have a terminal disease--so they won't test you unless you go through whatever protocols they want you to go through. But, daily, people are tested and diagnosed with terminal illnesses, such as cancer, without going through genetic or psychological counseling. People get cancer biopsies and learn they have cancer. That might not be a gene specific disease, but it's still terminal. To me if someone is displaying the signs of a serious disease, then they should be tested as quickly as possible, and begun on treatment as quickly as possible (as long as the patient wants treatment).

There are international guidelines for **predictive HD testing**, published in 1994, which I feel are somewhat outdated. The guidelines state that since there is no cure or prevention for HD, there is basically no medical need for predictive testing, and thus one needs to be really sure he wants to know he has Huntington's Disease. Therefore one should go through genetic counseling before being tested.

These guideline also indicate that learning that you have HD is such a shock that you need follow-up counseling. Do people with other terminal illnesses go through all of this counseling? It might help some cancer or Alzheimer's patients, but it should not be mandatory.

Since the guidelines were published in 1994, new medicines have been developed, and treatment options advanced, so it makes sense when one is beginning to have symptoms, to, as quickly as possible, find out if these are indeed HD symptoms, because there are possible treatments that can potentially slow the progress of HD.

It should also be noted that these international protocol guidelines are "guidelines," not hard and fast rules, and most importantly even though they are guidelines, these guidelines state that

6.2 Refusal to undergo these and other additional examinations will not justify the withholding of the test from applicants.

Source: NEUROLOGY 1994;44;1533-1536

Foreword: Recommendations concerning the use of a predictive test for the detection of Huntington's disease (HD) were drawn up by a committee consisting of representatives of the International Huntington Association (IHA) and the World Federation of Neurology (WFN) Research Group on Huntington's Chorea.

Again these guidelines are for "predictive testing." Yet, labs are repeatedly requiring genetic counseling and protocols based on these guidelines--not only for predicative testing, but **also for diagnostic testing.**

Regarding HD testing of minors, "An international consensus exists that asymptomatic individuals should not have testing during childhood." "Gene Reviews: www.genetests.org).

I have no problem with that, but labs have "protocols" that make it difficult to have symptomatic teens tested for HD, even though there is a juvenile form of HD that strikes before age 20 and has a life expectancy of less than 10 years from onset.

Some geneticists have acknowledged that in certain cases, where a child is symptomatic, then gene testing is appropriate: "*The testing of minors is advantageous when early intervention is the most effective prevention or treatment; careful attention must be given to designing appropriate informed consent permissions for minors.*" Source: The Tiresias Complex: Huntington's disease as a paradigm of testing for late-onset disorders. NANCY S. WEXLER Department of Neurology and Psychiatry, College of Physicians and Surgeons, Columbia University, New York, NY 10032, USA.

Before explaining the details of what happened to my family, I will give a little more information about the Huntington's Disease test. Below is information about how Huntington's Disease is diagnosed, which I copied from a National Institute of Neurological Diseases and Stroke (NINDS) website. It is also printed in booklet form:

How is HD Diagnosed?.....*The discovery of the HD gene in 1993 resulted in a direct genetic test to make or confirm a diagnosis of HD in an individual who is exhibiting HD-like symptoms. Using a blood sample, the genetic test analyzes DNA for the HD mutation by counting the number of repeats in the HD gene region. Individuals who do not have HD usually have 28 or fewer CAG repeats. Individuals with HD usually have 40 or more repeats. A small percentage of individuals, however, have a number of repeats that fall within a borderline region*

.My Family's Experience

To make a long story short, for a number of years, I have noticed various symptoms in my husband and his father, and more recently in my teenaged daughter. As my husband's symptoms worsened, I recently realized that my husband probably had Huntington's Disease. Looking on the Internet at a website that supposedly listed HD resources, I found the phone number of Queen's Genetics. It was listed as a place in Hawaii that did HD genetic testing.

I called this place and asked if they tested for HD. I was connected to a genetics counselor. She said she didn't do the testing, but a doctor came from Stanford once a month at the Hawaii Community Genetics Center, which I later learned was a new, somewhat hybrid center of Queen's and Kapiolani Medical Center Genetics Departments.

Upon realizing that my family's appointment was 2 1/2 months away. I called back to inquire about the long wait and was told that only 1 doctor did Huntington's and he came only every 3 months.. So we waited 2 1/2 months and went for genetic counseling with 2 genetic counselors and an MD in training--an intern, resident, or something. All 3 of these agreed that there definitely was some kind of genetic link with the problems my daughter and husband had.

They reported back to the "real" doctor, Dr. Hoyme, who turned out to be a pediatric geneticist. After having about a 10 min. briefing/summary of our 1 1/2 interview, the "real" Dr. reported back that he would not do the test because he didn't think my husband had Huntington's Disease. To which I responded, "Couldn't you just do it anyway?"

The real Dr. went on, "We need to get this man treated, and I'm not going to scare this young girl about a horrible disease. Huntington's disease is a disease for "young men and he (my husband) is an old man." (My husband is 65 and he's been displaying symptoms for 15 years or more, but we only now figured out that it is probably HD due to the sudden rapid worsening of his symptoms. Getting my husband **diagnosed** and **treated** was my whole motive for going to that place. **We are trying to get him treated**).

The genetic counselor interrupted the "real" doctor saying maybe we wanted to get a 2nd opinion, and she gave me the names of 2 neurologists. Obviously she felt HD was a real possibility, but the doctor was overriding her.

This made me think that there must be other places in Hawaii that did HD testing so when I went home, I called the lab that works with Queen's. They said they don't actually do the tests, but they draw the blood and send it off. I asked for the names of doctors who had ordered the test in the past and was told that my family doctor could order the test. So we went to our family doctor, who, observing that my husband had all the symptoms of HD, ordered the test. But when the lab worker came to draw the blood, he said a person had to go through genetic counseling, and be referred by a genetics counselor before lab would do an HD test.

I called back to the lab, and was told if the test was for diagnostic reasons, genetic counseling was not necessary; but if it was predictive reasons, genetic counseling was required.

Meanwhile, my doctor had talked to the lab, and they told her we had to go through genetic counseling--with the same people we had already been through counseling with. The same doctor would be back in Hawaii the following month, and we could see him again.

I called back to the lab; this time they told me that for diagnostic purposes, they would draw the blood, and send it to their genetics dept. (the same one we had already dealt with), which made me think that diagnostic testing really wasn't done by this lab.

There is also another lab here so I called them, asking if they did the HD test, and if there were any requirements for the test. They said they draw the blood for HD testing, but send it off to the Mainland for the actual testing, and that there were no requirements for testing.

Our family doctor wrote the lab order; we went to the lab, and my husband had his blood drawn. The local lab mailed it off to Genzyme in California.

A few days later, our doctor got a FAX from Genzyme, stating their "protocol," part of which is copied below, taken from their website, which had exactly the same wording as in the Fax. The web site gave their MA address contact information.

Contact Information

Genzyme Genetics
3400 Computer Drive
Westborough, MA
01581
T 800 326 7002
F 508 389 5577
Client Services
T 800 848 4436

Diagnostic Testing Protocols

Adult Symptomatic Testing *Confirmatory testing will be provided for patients with a family history of HD who have been evaluated by a neurologist and determined to have symptoms of HD. For patients with clear neurological symptoms but no known family history of HD, testing is available, and may be extremely helpful in the differential diagnosis*

(my emphasis)

Although it says that testing for those with no known family history is extremely helpful, and that they will test when clear neurological symptoms are present--it **does not say anything about being evaluated by a neurologist for the category my husband is in**--but an attached cover sheet to my doctor stated that they would hold my husband's blood for 2 months while he met these requirements--i.e. see a neurologist, which is a stated requirement of only those with a known family history of HD.

Also their protocols for minors is printed below:

Testing of Minors *In accordance with the published guidelines regarding testing for HD, Genzyme Genetics will not accept specimens from minors. For a compelling medical reason identified by a genetics center specializing in HD, exceptions may be considered.*
(my emphasis)

There are no genetics test centers in Hawaii--does that mean that no one in Hawaii ever develops HD? I don't think so. People in Hawaii are entitled to have their suspected diseases diagnosed and treated. But Genzyme states that if a minor does not have symptoms recognized by an HD center, they cannot be tested at their lab, which, to the best of my knowledge, is the only lab that tests for HD for people in Hawaii. Yet 10 % of the people who have HD, have juvenile HD. Juvenile HD affects juveniles; they need testing and treatment now, while they are juveniles--not when they are adults or when they are dead!

If I finally succeed in getting my husband tested and HD is verified, I then would like my daughter tested to find out how many repeats she has because usually the higher number of repeats, the quicker the disease progresses. My daughter needs to get appropriate treatment and plan her life. Is she to plan for college and a long life, or, on the other hand, is she to try and make the most of her last few years of life?

All I want to do is first get my husband tested, and, if his test is positive, then get my daughter tested so that they can be properly diagnosed and begun on treatment. I have been trying to get this done since July. We do have an appointment for my husband with a neurologist, whose specialty is epilepsy. (There are no HD specialists on Oahu.) But, waiting a month and a half to see this neurologist is just an added layer and additional delay in diagnosing my family. Many websites talk about how HD can be definitely and easily diagnosed through genetic blood tests so this would seem the route to go. Why waste time and money on MRI's, CT's, unneeded visits to specialists, etc., none of which can definitely diagnose HD disease, when a genetic blood test can?

And while I am somewhat sure my husband has HD, it could be Parkinson's or one of a number of other neurological diseases. But with the HD genetic blood test, HD can be easily confirmed or ruled out. Some Parkinson's disease medications help HD patients, but some other Parkinson's drugs seem to worsen HD. So we want to wait for the correct diagnosis before beginning treatments.

And, while we spend months trying to get this done, my husband's and daughter's conditions are deteriorating. My family, and I believe that this is wrong because, while there are no cures for HD, there are treatments. And my family and others should be able to get their loved ones diagnosed and treated as quickly as possible to try and slow the ravages of HD before excessive damage occurs in our loved ones' brains.

I feel like we have been discriminated against because the test we want, while very easily confirming or denying HD, is a genetic test. If it were just a lab test, for cancer or something, it would have been done by now. There would not be worries about "treatment teams, psychologists, counselors," etc

Hopefully I have provided an accurate picture of how I perceive that our family has been discriminated against by the "keepers of genetic tests."

Ruth Yong

The above is my story, but I've copied below, the story of a lady, which I found on an old internet forum about HD. The lady's name and email-mail have now

been deleted by the website. but her story shows the same anger and desperation that my family feels. In addition to the story below. I have also read about another family that had similar problems getting tested for HD. I don't think it's an uncommon thing to have difficulty getting the HD test, even though one's doctor has tentatively diagnosed him with HD.

This article submitted by on 12/15/97.

My husband's mother was recently diagnosed with Huntington's Disease in her mid 70s. The diagnosis was confirmed with a genetic test. This was a surprise since her own parents lived to be 68 and 89 with no symptoms. My husband would like to be tested. We are mature, stable people with supportive friends and family and we've been happily married for twenty-five years. He really wants to know. We went together to our family doctor who is wonderful. He asked us if we would prefer to go a HD clinic. My husband did not - he really likes and trusts our doctor. So, our family doctor provided counseling and information - he is really well-informed about HD. He ordered a genetic test and sent a sample my husband's blood off to a lab. Six weeks go by. The lab director decides not to do the test BUT HE DOESN'T EVEN CALL OUR FAMILY DOCTOR. My husband calls the lab and the lab director says he's not going to do the test. He wants my husband to go through an HD clinic. He says he was primarily responsible for putting together a set of protocols and my husband will have to follow them. He says he has to be convinced that my husband won't commit suicide; he says he doesn't know my husband. THAT'S EXACTLY RIGHT and NEITHER DOES THE HD CLINIC. BUT our family doctor does! We figure this can't be right. We call the closest HD Clinic. They are incredibly patronizing. WE call the HDSA and we're told we have to go to a HD Clinic and follow these protocols and if my husband passes the psychological testing THEN AND ONLY THEN can he have a test. We feel like we're dealing with the HD MAFIA. We have seen this disease rob my mother-in-law of control over her life. My husband says it may happen to him but he's not willing to have this process start with medical and lab personnel who are strangers to him taking control of his life and decision-making process under the guise of 'protecting him.' Does any one know of a lab our family doctor can deal with? Did everybody really have to go through a HD CLINIC to get tested? Please help!

Comments from the October 2004 SACGHS Meeting

SACGHS Meeting
Bethesda, Maryland
October 18th, 2004

Jeff Shaw, MS
Penrose Cancer Center
Colorado Springs, CO
Email: Jeffreyshaw@centura.org

Introduction

My name is Jeff Shaw and I am the director and genetic counselor for the Hereditary Cancer Service (HCS) at Penrose Cancer Center in Colorado Springs. I would like to thank you for giving me the opportunity to present information from our program to this committee.

I have provided genetic counseling for individuals and families in every area of medical genetics from prenatal to adult onset conditions. For the last seven years I have worked exclusively with patients concerned about hereditary cancer predispositions. The purpose of the HCS is to;

1. provide the best estimate of cancer risk so that screening can be appropriately modified so that if a cancer occurs it is caught as early as possible when survival is highest and
2. provide the appropriate implementation of medical and lifestyle interventions to drastically reduce the risk of cancer, especially those with an inherited predisposition.

Our program is clinically based and bridges the gap between research-based programs and the implementation of genetic testing into general medical practice. Although it is a clinically based program, at the onset we set up a database to collect important patient information. The data presented today covers seven years of clinical service to over 900 individuals.

Fear of Genetic Discrimination

At the initial genetic counseling appointment, a 3 to 4 generation pedigree is collected to ascertain the family history of cancer. When available, medical records are requested to confirm the diagnoses. The next step is to educate the patient regarding the differences between sporadic, familial (multifactorial) and inherited cancer predispositions. Sixty-one percent of patients seen in our program have a strong enough family history to pursue genetic testing.

If an individual is offered the possibility of genetic testing, we proceed with the informed consent process that includes discussing risks, benefits and limitations of the testing. Discussion of genetic discrimination issues almost always takes up the bulk of the time devoted to informed consent. In addition to expressing concern for their risk for genetic discrimination most patients also express concern of the impact the testing could have on their children and siblings.

In our program's experience, 20% of those individuals eligible for testing decline having the test. Of those that declined the test, 22% did so because of a fear of discrimination. Of these patients that decline the testing due to a fear of discrimination, 90% had a 40% or greater chance of testing positive for an inherited predisposition. It is these patients that would have the greatest potential benefit from the test results.

Patient Experiences

These numbers take on a more personal tone when considering an actual patient's situation. I recently saw a woman that had a strong family history of cancer. She had just been diagnosed with a stage I breast cancer at the age of 46. Her mother died of ovarian cancer at the age of 52, two maternal aunts had breast cancer in their early 40's, and the maternal grandmother died from breast cancer at age 41. We determined there was at least a 43% chance she carried an inherited mutation that was causing such a high occurrence of early onset breast and ovarian cancer in her family. If she pursued testing and was determined to have an inherited predisposition, she would have up to a 60% risk for developing a second primary breast cancer and up to a 44% risk for primary ovarian cancer. Prophylactic surgical intervention could reduce her risk by over 90% for the development of another primary breast and/or ovarian cancer.

If she tested positive, each of her four daughters would have a 50% chance to inherit the faulty gene that would give them these high risks for cancer.

She has declined testing, as she fears how this information could affect her children's ability to obtain health insurance in the future. Without the genetic testing, it is unclear how to proceed with prevention options, especially surgical ones, to reduce her risk for new cancers. Without documentation that she tested positive for an inherited mutation, her insurance company will not pay for the surgical interventions. She remains in a state of anxiety using imperfect breast and ovarian screening methods hoping that another cancer does not occur.

Due to her current employment situation she might have to change insurance companies. She is afraid that if she were to change insurance companies she could be denied insurance. With her current diagnosis, she cannot afford to be without health insurance.

Another patient had a strong family history of FAP. A dominantly inherited colon cancer predisposition characterized by the early onset of hundreds to thousands of colonic polyps. Polyp formation can begin as early as age 10 and colon cancer occurs in these individuals decades younger than sporadic colon cancer. He has worked his whole life at a relatively small company with a small group health insurance plan. He had been warned by his doctor not to have genetic testing for FAP as he could lose his job or his health insurance if they found out. He also neglected to have colon screening performed in order to keep the family history a secret.

At the age of 42 he had significant rectal bleeding and finally went in for an evaluation. He was found to have over 400 polyps in his colon. It was so extensive he needed to have his entire colon removed, a drastic but lifesaving procedure for individuals with FAP. Luckily, he did not have an invasive colon cancer. Other family members were not as lucky, most of those affected with FAP dying from colon cancer in their late 20's. At the age of 46, he came to me for genetic counseling.

He has two children ages 22 and 24. He had not informed them of the condition as he did not want the family history in their medical records for fear of future discrimination. Unfortunately this meant they were not having appropriate screening. We had several genetic counseling sessions with him and he finally decided to let them know about the inherited colon cancer predisposition in his family. He was tested and the genetic mutation causing FAP in his family was identified. His children decided to have testing and one child tested positive and one child tested negative for FAP. The 22 year old that tested positive is now having appropriate

screening and so far has only had a small number of polyps. The fear of genetic discrimination could have caused the same early deaths in his immediate family as it did in his aunts, uncles, and cousins.

I just saw a 24 year old patient whose mother tested positive for a mutation in one of the breast/ovarian tumor suppressor genes. When she informed at risk family members, over half stated they would not have the testing due to a fear of genetic discrimination. Although anecdotal, my experience tells me that this is common in families with an inherited predisposition.

What About After Testing?

We conduct one-year follow-up surveys of all patients seen by our program. We have a 72% response rate to these surveys. Of those patients that have had genetic testing and tested positive for an inherited cancer predisposition, 70% report having continued anxiety that they could experience genetic discrimination in the future. Fear of future genetic discrimination remains a real concern for our patients that have tested positive.

What about the people that don't even make it to genetic counseling?

In addition to fear of discrimination from genetic testing, there is also a fear of discrimination simply from participating in a genetic counseling session. In 2001, Geer et al. studied factors that would influence an individual's decision to proceed with genetic counseling (1). Of those declining genetic counseling, the biggest reason was a fear of genetic discrimination accounting for 40% of those surveyed. Over the years of our program's existence we have had a significant number of physician referred patients not show up for their genetic counseling appointments. After seeing the article by Geer et al. we conducted an informal six-month survey of those patients not pursuing a referral for genetic counseling.

In this time frame, we had 60 patients that had not shown up for their scheduled appointment. Fifteen percent of these patients could not be contacted. Of those we could contact, 49% stated they had changed their minds and did not want to have the genetic counseling due to a fear of genetic discrimination regarding their health insurance. The remainder stated they wanted to delay the appointment due to current cancer treatment or needed to reschedule due to forgetting their appointment or logistical problems making it to their appointment.

This informal survey supports the data seen in the Greer study. It shows us that a fear of genetic discrimination is a barrier for individuals that could benefit from genetic counseling and possibly genetic testing for hereditary cancer predispositions.

Conclusion

When I graduated with my genetic counseling degree in 1994, there were but a handful of genetic tests available for inherited conditions. In 2004, just ten years later, there are over 1,000 genetic tests available on a clinical or research basis (2). The number of genetic tests that will become available for single gene and complex genetic disorders is expected to increase exponentially over the next decade. I fear that without strong Federal protection, the appropriate use of these tests will continue to be underutilized.

None of us are genetically perfect. Learning what genetic imperfections we have inherited and how they affect our risk for disease is a difficult, sometimes frightening and life-changing experience. The decision to have presymptomatic genetic testing is multifaceted. It encompasses issues regarding one's sense of self, family relationships, anxiety, depression, and complex decisions regarding medical intervention. The citizens of our country need to be assured that when they are deciding whether or not to pursue genetic testing, a fear of genetic discrimination is not a factor.

Thank you for your kind attention.

References:

1. Geer KP, Ropka ME, Cohn WF, Jones SM, Miesfeldt S. Factors influencing patients' decisions to decline cancer genetic counseling services. J Genet Couns. 2001 Feb;10(1):25-40.
2. GeneTests: <http://www.genetests.org> - (funded by the NIH)

Sarata, Amanda (NIH/OD)

From: Shaw, Jeff [JeffreyShaw@Centura.Org]
Sent: Wednesday, August 18, 2004 1:12 PM
To: Sarata, Amanda (NIH/OD)
Subject: Genetic Discrimination

I find it very disturbing that certain members of the house are not supporting the antidiscrimination legislation they have stuck in committee.

I work with individuals that have very strong family histories of cancer. By having genetic counseling and testing, we are much better able to assess their risk for the cancers that run in their family. Unfortunately I have had many patients that will NOT have the genetic testing performed due to a strong fear of genetic discrimination. Without a Federal law to protect them in whatever state they may live in, they will not pursue the testing that could save their life.

For example, individuals that test positive for a faulty BRCA tumor suppressor gene have a risk for breast cancer between 44-87%, and a very substantial increased risk for ovarian cancer. Those that test positive can have increased screening to increase the chance that if they develop cancer, they will catch it at an early stage when it is most treatable. Other patients have prophylactic surgery to reduce their risk of getting a breast or ovarian cancer by over 90%.

Those that test negative do not need increased screening or surgical procedures. In other words, genetic testing allows for the appropriate management of patients, and the appropriate utilization of health care dollars. However this is not possible when patients deny testing based because there are no legal protections against genetic discrimination. These patients are NOT followed appropriately and lose out on information that could save their life, in addition to saving millions in health care expenditures.

There should be no experimentation or "let's see what happens" when it comes to the issue of genetic discrimination. Basing medical insurance coverage on and individuals genetic information and potential for disease is morally wrong, racist, and we should put protections in place to make sure it never happens. Everyone in the country has faulty genes that increase their risk for disease, so everyone would be at risk for discrimination as genetic testing expands to included very common conditions, including diabetes and heart disease.

I have contacted several of these patients to see if they would write a letter expressing their concerns, but they do not want to be publicly identified.

I write in strong support of the current legislation, and to express the support of the thousands of patients I have cared for. This issue will NOT go away. If they choose to shoot down the current legislation, their will simply be another to take its place.

Jeff Shaw, M.S.
Oncology Genetics Counselor
Penrose Cancer Center
110 East Monroe St.
P.O. BOX 7021
Colorado Springs, CO 80933

Phone: 719-776-5274
FAX: 719-776-2516
Email: Jeffreyshaw@centura.org
Website: www.penrosecancercenter.org

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Social and Behavioral Research Branch
National Human Genome Research Institute
National Institutes of Health

10 Center Drive - MSC 1852
Building 10, Room 10C103
Bethesda, Maryland 20892-1852
Phone: (301) 496-3980
FAX: (301)480-7825
e-mail: dhadley@mail.nih.gov

Secretary's Advisory Committee on Genetics, Health and Society
Provider Testimony presented October 18, 2004

My name is Don Hadley. I am an Associate Investigator within the Social and Behavioral Research Branch and a Genetic Counselor within the Office of the Clinical Director within the National Human Genome Research Institute at the National Institutes of Health. I'd like to thank you for the opportunity to present our work to this committee.

My professional experience and work has focused primarily on providing education and counseling to families who are threatened by genetic and inherited diseases. I have had the privilege of working with these families for the last 23 years. In October 1993, I was invited to join the then newly established National Center for Human Genome Research. Our goal was to develop research that focused on identifying the factors that influence interest in genetic counseling & testing and the associated psychological, social and behavioral outcomes. Our research agenda was inspired by the identification of a rapidly growing number of genes that predispose or increase susceptibility to disease.

My research has specifically focused on families who are newly diagnosed with a hereditary cancer syndrome known as Hereditary Non-Polyposis Colorectal Cancer or HNPCC and, in whom a deleterious mutation has been identified. The identification of families with a HNPCC mutation allowed the opportunity to learn from them before, during and after the provision of genetic counseling and the offer of genetic testing. We felt that identifying the factors influencing decisions and their outcomes was necessary to plan for a future when genetic testing will be more routinely used to predict risks for rare as well as common diseases in the general population.

Within our study, once a family is identified to carry an HNPCC mutation, we sequentially offer participation to first-degree adult relatives who are at 50% risk of inheriting the mutation. This offer includes the provision of comprehensive information about HNPCC, the cancer risks associated with it, recommended cancer screenings, a discussion about the genes responsible for HNPCC, the pattern of inheritance, the potential benefits, limitation and risks associated with genetic testing and finally the offer to undergo genetic testing. For those electing to pursue genetic testing, the NIH Clinical Center pays for genetic studies removing the issues of costs and insurance coverage from the decision-making process. However, prior to the education and counseling sessions, we ask participants to complete a questionnaire that collects demographic information, their knowledge of genetics and genetic testing related to cancer, their perceived cancer risks, and standardized scales assessing mood, coping style, spirituality, control over

Don Hadley, M.S., CGC
Associate Investigator & Genetic Counselor

health related issues and family relations. Additionally, we also ask them to specifically identify what factors motivate them to consider genetic testing such as the desire to clarify their childrens' risk for cancer or to guide their cancer screening behavior. Likewise, we also ask them to identify factors that concern them about undergoing genetic testing, such as emotional concerns about handing the results for themselves or family members, their level of confidence in prevention strategies and their concerns about test results affecting their insurability. These questions are asked individually so that we might obtain a level of significance for each issue. In addition, we ask the participants to identify the most important reason for them to consider testing and likewise, the greatest concern they have in considering testing. All of the questions are validated measures developed and used by the Cancer Genetics Studies Consortium of the NHGRI Ethical, Legal and Social Implications Program.

As we began to talk individually with each participant describing the intent of the study and the process involved, one key issue was consistently identified; that issue was posed in the form of a question "how might participation in this study affect my insurance or that of my family?" The question came unsolicited in the beginning of the informed consent process. This question and the associated worry seemed to persist even after we had provided each participant with information ahead of time that 1) reassured them of the confidential nature of our study, 2) the fact that the study has a Certificate of Confidentiality, 3) that all participants are assigned study ID codes that removes their personal identifiers from the data and test results, 4) that the costs of testing are paid for by the study so insurers are not involved and 5) that records are protected by the NIH Clinical Center and only released with written permission. Never the less, it was clear that there was an overwhelming concern and, in some cases, a palpable anxiety about the potential impact of genetic testing on health insurance. These concerns dominate our informed consent processes and recur session after session with an intensity that opened our eyes to the level of concern the public feels about genetic discrimination.

I specifically recall one young woman and her mother, both of whom had experienced uterine and colon cancers at young ages within a family riddled with HNPCC cancers. Even though this young woman had experienced cancer twice and felt there was little residual discriminatory risk to her, she was immobilized by her concerns about the potential that a genetic test result might brand the family as uninsurable. She opted to wait on testing but would periodically call our team to discuss the safeguards our study provided regarding test results and information obtained. She was admittedly tortured by the concerns about insurance risks which she felt was keeping her from protecting her family. Finally after months of considering the implications of testing she returned to pursue testing knowing that her results may well prevent others from experiencing what so many in the family had already endured from cancers diagnosed too late. Sequencing efforts did identify a deleterious mutation providing a tool for those within the family to clarify their cancer risks, to focus their cancer screening and to consider preventative steps such as prophylactic surgery. We anticipated that within the months that followed, we would be hearing from at least a few of her family members. But there were no calls, no e-mails, no letters, which surprised us. Fortunately, our study includes follow-up calls allowing us contact with study participants at selected time intervals. Through that follow-up, we learned that she had shared the results with her sisters, and some of her other relatives. Amongst her sisters, they expressed that their concerns regarding discrimination were too great to safely allow them participate in a genetic counseling study with the option of genetic testing. They were worried about being in small companies with limited insurance options and the associated risks that genetic testing might impose on their children for future employment and insurability.

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Associate Investigator & Genetic Counselor

We stepped back from this experience and wondered, how many others won't participate in our research or pursue clinical genetics services because of the concern regarding genetic discrimination. How pervasive is the threat of genetic discrimination on this very personal issue regarding our genetic make-up?

In 2003 we published a paper within the Archives of Internal Medicine which I have brought along for your consideration. In this paper we reported on the attitudes, intentions and uptake of genetic testing of individuals within these families. Of particular relevance to this group were our findings regarding the level of concern that exists within these families about participating in a genetic counseling and testing research study.

The questions that we use to identify their concerns regarding genetic testing are included within the packet before you. These questions are part of a comprehensive questionnaire that assesses many psychological, social and behavioral variables.

To assess participants' perceptions of genetic testing we used a series of 14 Likert-style items, adapted from previous research to assess perceptions of the benefits, limitations and risks of genetic testing. Participants read a series of benefits and limitations/risks of HNPCC testing and were asked to rate the level of importance as *not at all important, somewhat important, or very important*. In addition, the participants were asked to choose the single most important benefit and limitation/risk of genetic testing from the series.

In looking specifically at what factors influence decisions against genetic testing, we identified and published that 39% of participants reported that their most pressing concern was their worry about the potential for the genetic test result to affect their or their family's insurability. I recently looked at our data to see if the level of concerns has held true from the earlier analysis as ~80 additional participants have completed the study since the initial report. I found that our current baseline data suggests that the number is slightly higher with 43% identifying that their greatest concern regarding genetic testing pertains to the potential of genetic discrimination by insurance companies. Furthermore, on follow-up with these people at 6 and 12 months, a greater proportion of them identify insurance discrimination as the single most worrisome factor at each follow-up time point. Specifically, at 6 and 12 months, 48% and 54%, respectively, identify concerns about genetic discrimination by their insurer as their principal concern. Obviously, the concern is not going away with time and adjustment to the outcome of testing. This seems surprising since research from other studies focused on pre-symptomatic and susceptibility testing demonstrate that other variables, such as anxiety, distress, and mood seem to return to pretest baselines by one year out. What's different about concerns regarding insurance? If people have not experienced what they perceive as discrimination, why are there increasing concerns? Participants often ask, "Has anyone within the study ever reported discrimination on the part of their insurance company or employer?" Our answer is always the same – not that has ever been reported to us. But apparently just reassuring them that discrimination in general, and within our study is rare, doesn't help. There is a pervasive mistrust that seemingly worsens with time.

In summary, from a qualitative perspective, the informed consent process is dominated by concerns about genetic discrimination by insurers. Quantitatively, the most common concern expressed at baseline, 6 and 12 months following genetic counseling and testing is concerns about insurance discrimination, with increasing numbers of participants identifying genetic discrimination as the most worrisome concern at each follow-up interval.

Don Hadley, M.S., CGC
Associate Investigator & Genetic Counselor

The prevalence of genetic discrimination by insurance companies does not appear to be the key issue. The real issue is that the public perceives that the potential for genetic discrimination by insurance companies is an overwhelming risk and in my experience this fear provides a barrier to genetics research and clinical genetics care. This barrier limits our potential for research in basic sciences and social and behavioral research. The greatest tragedy, however, is the missed opportunity to prevent cancer or diagnose it early in persons at high risk who are unwilling to risk the potential of discrimination. Providing federal legislation prohibiting genetic discrimination will 1) reassure the public that genetic discrimination is not a risk, 2) provide increased opportunities for research to address other more significant issues and, 3) most importantly reduce morbidity and mortality associated with cancers diagnosed at later stages.

Thank you for the opportunity to present our work. I welcome your questions.

Don Hadley, M.S., CGC
Associate Investigator & Genetic Counselor

Testimony of Mark Brantly, M.D.

The last of our health provider presenters will be Dr. Mark Brantly.

DR. BRANTLY: I'd like to thank the committee for inviting me to come and speak. My name is Mark Brantly. I'm a pulmonary physician and a physician scientist at the University of Florida. I've been involved in alpha-1 antitrypsin deficiency testing since approximately 1983 and have tested about 20,000 individuals, identified about 2,000 alpha-1 individuals over the last 20 years. In recent years I've been testing approximately 5,000 to 6,000 patients per year for alpha-1 antitrypsin deficiency.

I follow approximately 150 alpha-1 antitrypsin deficient individuals in my clinic at the University of Florida and have first-hand experience regarding the impact of this diagnosis on them personally and also their families.

Let me begin by giving a brief expose of alpha-1 antitrypsin deficiency. It's a very easy disease to diagnose. It requires simply an alpha-1 antitrypsin level and a PI type or a genotype. It's one of the more common genetic diseases, with a frequency of 1 in 2,500 to 1 in 4,000 individuals. The phenotype is primarily chronic obstructive pulmonary disease and liver disease. It's oftentimes associated with a rapid decline in lung function punctuated by lung infections. However, it's one of the classic genes in which there's an environment and gene interaction. That is, individuals who have alpha-1 antitrypsin deficiency lose lung function much faster when they smoke cigarettes. Indeed, they die 20 years prior to non-smoking individuals.

Importantly, in my clinic population I have individuals that are 80 years old with profound alpha-1 antitrypsin deficiency who are living active lives. Therefore, prevention of behaviors and interactions is a critical aspect of this disease. It is not all about having expensive therapies. People can live their entire lives with not having disease or disability if they're identified early and we're able to protect them. That, I think, forms the basis of early diagnosis and preventive care being critical if we are to make a significant impact in this disorder.

In the State of Florida only, there are 900,000 individuals with COPD, and 9,000 die per year. Almost 1,000 of these individuals have at-risk alpha-1 antitrypsin deficiency alleles. In the State of Florida we've had a program in which we have done targeted detection. We first began by establishing a consensus among the community with the help of the Alpha-1 Foundation that testing exceeded the risk of testing. We established a high-throughput laboratory, and we provided professional and lay educational materials to deal with some of the educational issues that are associated with alpha-1 antitrypsin deficiency diagnosis. We developed an easy testing system where patients can prick their finger and send it to our central laboratory, yet we still have significant barriers to testing these individuals despite major recommendations from the major thoracic societies recommending a Category A recommendation for testing.

These barriers include genetic discrimination, and particularly fear of genetic discrimination, ignorance regarding the disease among the physician population. We've also established tertiary care referral systems to make sure that when physicians do identify these patients, that they have someplace to go with these patients.

So we have yet still an important job, and that is to be able to -- instead, right now, we have 5,000 individuals that are identified with alpha-1 antitrypsin deficiency, and there are approximately an estimated 95,000 that haven't been identified. If these patients were identified early on, they perhaps could be protected from developing disability.

One of the approaches that we've used is doing a coding testing trial through the Medical College of South Carolina and Charlie Strange. This is funded entirely by the Alpha-1 Foundation, and it's been a longitudinal study looking at the reasons why people do not wish to be tested through their physician. We've tested now more than 3,300 individuals in this testing program and have done some initial longitudinal follow-up. I've provided you with one manuscript that gives you some of the results, but I'd like to focus in on a couple of things most recently that we have done.

The first one is the risk and benefits of genetic testing. Thirty-three percent of individuals said that the reason why they chose the coded testing trial was because of fear for losing their health insurance or higher health insurance costs. The other thing is in the post-test, who would you give your results to? Well, not surprisingly, they would give the results to their children and their spouse, and not surprisingly they wouldn't give it to their ex-spouse.

(Laughter.)

DR. BRANTLY: In addition, they would not provide this information to their health insurance companies or their life insurance companies. Indeed, only about 16 percent would disclose that. Sadly, though, I have to say that only 80 percent of these individuals who were profoundly deficient would even tell their personal physician, and that's problematic as far as I'm concerned.

Finally, one of the things that this study I think brings up in close contrast is that when patients were diagnosed with alpha-1 antitrypsin deficiency, obviously one of the major therapies is to do smoking cessation. While there was a trend towards individuals who had alpha-1 antitrypsin deficiency quitting smoking, this was not significant. In actuality, it was higher for alpha-1 antitrypsin deficient individuals, still there was a large portion, greater than 80 percent, who did not quit smoking.

In my clinic and in many of the physicians' clinics who take care of alpha-1 antitrypsin deficient individuals, I have a 95 percent quit rate for cigarette smoking. The national average is 10 percent. Why is that? That's because I hound these patients to death. I schedule them for appointments to see me every month, I have my nurses hassle them, because I know of all the things that I do for these individuals, getting them to quit smoking is clearly one of the most important things that I can do.

When we have to resort to coded testing and we leave out the physician and the health care provider in helping these individuals cope with and make these changes, we short-change them in a big way. We short-change them because they're afraid, because they can't trust our system to protect them and to give them the correct information. There's only one difference between my patients and me. We all as complex genetic organisms have five to fifteen "lethal mutations" that may be associated with our demise or our disability. The difference between me and my patients is I don't know about mine. My patients know about theirs and they have the ability to do risk prevention.

Thank you very much.

MS. MASNY: Thank you for all your testimony. It continues to clarify that genetic discrimination, and especially the fear of genetic discrimination, is very real.

Written Comments

Sarata, Amanda (NIH/OD)

From: Lorraine.Dugoff@UCHSC.edu

Sent: Wednesday, September 15, 2004 1:27 PM

To: Sarata, Amanda (NIH/OD)

Subject: SACGHS

Amanda,

I am a clinical geneticist. I wanted to let you know of instances where patients with children at risk for having neurofibromatosis have not felt comfortable openly discussing their medical concerns regarding their child with their physician because they are afraid that if comments re a potential diagnosis of neurofibromatosis appear on their child's medical record that their child will have difficulty obtaining medical insurance later in life. This can ultimately compromise the medical care provided. This is a serious concern for many people/families--with respect to many diagnoses.

Sincerely,

Lorraine Dugoff

Sarata, Amanda (NIH/OD)

From: Sandra Dunbrasky [sjdunbra@tvpweb.com]

Sent: Wednesday, August 25, 2004 8:37 PM

To: Sarata, Amanda (NIH/OD)

Subject: genetic discrimination

I am a Pediatrician in Ontario Oregon and have had great experience with perceived genetic discrimination while dealing with patients and their families. I would conservatively estimate approximately 6-8 times per year that I have difficulty getting parents to agree to genetic testing on their children who are at risk due to their fear of not being able to insure them or losing their insurance if the tests are positive. I have not actually had insurance companies turn away a claim based on genetic diagnosis. Other questions, please call or email. Thanks Sandra J Dunbrasky MD FAAP. Ontario Oregon

Sarata, Amanda (NIH/OD)

From: Farrell, Michael [mfarrell@mail.mcw.edu]
Sent: Tuesday, August 24, 2004 10:37 AM
To: Sarata, Amanda (NIH/OD)
Subject: Hearing on Genetic Discrimination comment

Greetings,

You might be interested in hearing a brief comment from an ongoing project of mine. I spend most of my time researching quality (processes and outcomes) of communication after newborn genetic screening. A few years ago, after being somewhat frustrated with the quality of the literature on psychosocial complications, I began a back-burner project to find everything I could written on genetic testing (adults or kids) and classify mentions of psychosocial complications by type and quality.

The key finding thus far has been that there is a LOT more written about this than there is actual data, even of the historical and anecdotal type. Most mentions of the problems (discriminatory or otherwise) after genetic screening are of the "lingering questions" variety, i.e. at the tail end of another paper, somebody mentions that discrimination, anxiety, etc could be a problem. Interestingly, there are also a lot of essays on the subject, but many of the essays tend to cite the same papers and anecdotes themselves ... if they cite at all. In many other cases, many papers discussing one disease cite papers about OTHER diseases to support the possibility of discrimination.

My interim impression of all of this is that discrimination and psychosocial complications have happened, but that the reason why they keep coming up is due to their dramatic nature. We really like talking about these complications, but for some reason we don't like to study them.

This disconnect has been a major impetus for my main research, leaving the review as more of a hobby. So, I don't have any ready-for-primetime data yet from this review, I just moved institutions and am trying to find some grad students who would be interested in helping. If that doesn't work, I'll probably pursue some small funding to get qualified people to do reviews with me and finish the project.

Good luck with the hearing process ... I hope that you are able to collect and report on your findings so I can cite the document in my study!

Mike

Michael Farrell MD-----
Asst. Professor, MCW Internal Medicine and Pediatrics
Center for Patient Care & Outcomes Research (PCOR)
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genetic discrimination

Sarata, Amanda (NIH/OD)

From: Maria.Fernando@UCHSC.edu
Sent: Thursday, August 26, 2004 11:21 AM
To: Sarata, Amanda (NIH/OD)
Subject: genetic discrimination

When I was a medical student doing a rotation in genetics at Eastern Virginia Medical School, I encountered a case of genetic discrimination that concerned me enough that I embarked on a research project with it in mind. A child was born and screened for galactosemia, a serious metabolic disorder, and was diagnosed with Duarte galactosemia, which is not so well characterized but is generally thought to cause no medical problems whatsoever. The health insurance company for this family (I can't remember the name, maybe Tricare) decided to refuse insurance for the baby on the grounds of the diagnosis of Duarte galactosemia. We wanted to send the health insurance company a letter saying that Duarte galactosemia was completely harmless and they had no reason to deny insurance to the baby. However, after a literature search, we found that there was some debate about whether to avoid certain foods over the first year for these babies and there was enough ambiguity about the case that we couldn't write with absolute certainty that the baby would not need any special care. In the end, the insurance company backed down and gave health insurance to the baby but we spent a great deal of time trying to persuade them to do so.

Good luck,
Terry Aly
theresa.aly@uchsc.edu

Sarata, Amanda (NIH/OD)

From: Eric Fowler [Eric.Fowler@BMHCC.org]
Sent: Wednesday, September 22, 2004 5:12 PM
To: Sarata, Amanda (NIH/OD)
Subject: A case of genetic discrimination in the workplace

Ms. Sarata,

I am the genetic counselor that Agnes Masny mentioned to you. My patient was recently fired and was told she was terminated because she had genetic testing (for BRCA1/2 mutations - and the test results aren't even in yet) and that would increase insurance rates for the company employees. I am including the e-mail I sent to the NSGC list serv below.

Any additional information you might need, please do not hesitate to contact me.

Eric Fowler

Hello, Everyone.

I had a disturbing phone call last night from a patient...

I have been working with this patient, and she has a strong family history of ovarian, breast, and fallopian tube cancer. She is unaffected, and there are no living affected people in her family who were willing to be tested.

Shortly before Labor Day, her benefits were checked, Comprehensive BRCAAnalysis was approved at 100%, and her blood was drawn. Her results are still pending.

She was upfront with her boss - going into detail about why she needed to miss work (for our appointments). The day she had her blood drawn, her boss asked her how long it would take for her test results to come back. She stated a few weeks to a month, as I told her.

Within a day or two, her boss called her into his office, and said he was firing her because her testing would make everyone's insurance rates in the office (company size about 60 employees) increase. This was the only reason given for her termination. She requested a separation notice, which should have listed reasons for termination, but the form was blank.

This all happened on or about 9/3, and she only just called me yesterday. She said she has been hospitalized and had other issues which prevented her from contacting me sooner. She also said that her boss did her a favor, because she did not like her job.

I told her that under the ADA, it was illegal for her to be dismissed on this basis (we don't even know what her test results will be, which should be inconsequential in this situation anyway!). In TN, we do not have a state law that specifically addresses genetic discrimination in the workplace. I encouraged her to seek legal counsel and that I would help her in any way that I can. I am not certain that she will take these steps, the 'principle' of the issue may be lost on this patient - she is happy not to be in a negative work environment and said as much to me over the phone.

Does anyone have any suggestions as to any additional steps I should be taking?

Your input would be greatly appreciated.

Eric

Opinions expressed above are not necessarily those of BMH
Eric Fowler, MS, CGC Certified Genetic Counselor
Director of Genetics, Baptist Centers for Cancer Care, Memphis, TN Phone (901) 226-4036,

October 26, 2004

Three Cooper Plaza
Suite 220
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cooperhealth.org

The Honorable Tommy G. Thompson
Secretary of Health and Human Services
U.S. Department of Health and Human Services
200 Independence Avenue, SW
Washington DC 20201

Dear Secretary Thompson:

Generosa Grana, MD
Medical Director

Dana Farengo Clark, MS, MS, CGC
Certified Genetics

Dianne L. Hyman, RN
Genetics Nurse

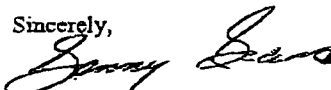
Vicki Lynn Kay
Secretary II, Genetics

We are writing on behalf of SACGHS in support of your efforts to prohibit discrimination in health and life insurance on the basis of genetic information. This legislation is extremely important to those of us who treat patients. Working in a Hematology/Medical Oncology practice, we see patients who have had a diagnosis of cancer or who have a strong family history of cancer. When speaking with these, we often hear concerns expressed that a positive test for a cancer predisposition gene will result in cancellation of their health insurance benefits or a dramatic premium increase. Patients also fear that an insurance company could deny coverage to themselves or their relatives for a diagnosis of cancer in the future if they test positive. Many of our patients present to us with the fear of having a "pre-existing condition" and although we make every attempt to dispel these concerns, there are times that this interferes with the process of testing.

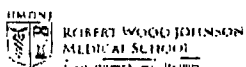
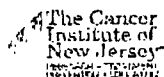
Our Camden, New Jersey clinic is a major urban center and we see a large number of minority patients. Through a research protocol, we are able to offer genetic testing, free of charge, to minority women at risk for hereditary breast and ovarian cancer. Since this testing is through a research laboratory and not a clinical laboratory the results are not a part of a woman's medical record and that the results remain confidential as part of the study. Unfortunately, issues of mistrust of genetic testing and its repercussions upon the future health of themselves or family members run rampant in the minority community. Even though free and confidential testing is available, we still encounter many barriers within this community. Even with recent HIPAA protections, minority women express distrust in the ability of healthcare providers and the government to protect their medical information and they often fail to see how genetic information will benefit themselves and their communities. Genetic information is a pivotal part of how we care for patients and their families, especially when making management plans for cancer screening and prevention.

We strongly believe that this legislative protection is imperative to ensure that our patients will suffer no ill consequences from having genetic testing. Furthermore, we implore you to provide your total support to this effort and know that the medical profession is behind you. On behalf of all of our patients, we would like to thank you for your continued work in this crucial area.

Sincerely,


Generosa Grana, MD
Hematology/Medical Oncology


Dana Farengo Clark, MS, MS
Certified Genetic Counselor



The Core Teaching Campus of the
University of Medicine and Dentistry of
New Jersey, Robert Wood Johnson
Medical School at Camden

Sarata, Amanda (NIH/OD)

From: Montgomery, Susan [S_Montgomery@fccc.edu]
Sent: Friday, September 24, 2004 12:21 PM
To: Sarata, Amanda (NIH/OD)
Subject: genetic discrimination

I am a cancer genetics nurse who recently counseled a young woman with a strong family history of breast cancer. Her mother, who is affected, had decided to proceed with testing. The physician in the local hospital's lab strongly advised the mother not to proceed with testing because of potential insurance discrimination. He informed her that she would have great difficulty obtaining or keeping insurance if she tested positive. The mother and daughter are both now very concerned about this issue and are second guessing their decision about testing. Both are seeing us next week for intensive genetic counseling.

Hope this information is helpful.

Sincerely,

Susan V. Montgomery, RN, BSN
Fox Chase Cancer Center
215-728-2405

Sarata, Amanda (NIH/OD)

From: Robbin Palmer [rpalmer@nnps.reno.nv.us]
Sent: Sunday, October 17, 2004 11:22 PM
To: Sarata, Amanda (NIH/OD)
Subject: comments for SACGHS meeting

My comments focus on two topics on the SACGHS agenda.

First, regarding Perspectives on Genetic Discrimination. Some patients at high risk for having a BRCA1 or BRCA2 mutation, and for whom genetic testing is indicated, do not seek testing because of fear of genetic discrimination-- particularly discrimination from their current or future health insurer. I am never involved in anonymous testing. I feel that it is irresponsible for a health care provider to knowingly arrange for anonymous testing, and arranging anonymous testing may lead to future legal liability for the provider. In my opinion, a clinical laboratory should not promote anonymous testing, or testing upon patient request, without involvement of a physician. Because I am not involved in anonymous testing, some patients at high risk for a mutation, seek testing through a physician willing to do anonymous testing. I suspect that positive genetic test results (mutation identified) are not included in the patient's medical records. If a mutation is identified in an individual, I don't know how other at-risk family member's undergo site-specific mutation analysis. If results are anonymous, it seems that the patient, physician, and laboratory assume legal liability when family members are tested for the familial mutation.

Second, regarding the Draft Report on Coverage and Reimbursement of Genetic Tests and Services. I believe I am the only certified genetic counselor in this state that offers cancer risk genetic counseling. Several of my clients pay my fee out-of-pocket because their insurance does not reimburse for genetic counseling or genetic testing. I am not a Medicaid provider in this state, and usually people on Medicaid can not afford my fee, although I discount my out-of-pocket fee substantially from the low fee I bill insurance. This prevents a sizable proportion of the population from accessing my services. Medicare does not reimburse for genetic counseling either. Based on the experience of other genetic counselors who see Medicare patients, some individuals meet the criteria for Medicare to reimburse for genetic testing, although that individual's actual risk of a mutation is low, and he/she would not ordinarily be offered genetic testing. A genetic counselor actually assesses an individual's risk of a mutation, rather than simply determining if an individual meets the Medicare criteria. Since genetic testing is often quite expensive, the money spent by Medicare for reimbursement of testing individuals at low risk for a mutation could be used for other more pressing health care needs.

Many providers are not trained in genetics, and have limited knowledge and understanding of the complexities of genetics. Genetic counselors too frequently provide information to a patient that is contrary to the information that was given by an M.D. I find that the patient doesn't know whether the M.D. (who has no training in genetics) or the genetic counselor (genetics professional) is providing accurate information.

Also, the time providing genetic counseling is often billed to insurance (or Medicaid) under a physician's name. The physician may not see the patient at the time genetic counseling is provided. Because the physician has limited knowledge and understanding of genetics, he/she may leave the management of genetic issues to the genetic counselor. This practice is wide-spread, although it is, in my opinion, insurance fraud. Genetic counselors usually can't bill insurers directly for the provision of genetic counseling services. Sometimes a genetic counselor is not involved, and the patient only gets information about genetic issues from a physician.

Thank you for your consideration of my opinions.

Robbin Palmer, Ph.D.
Certified Genetic Counselor

Sarata, Amanda (NIH/OD)

From: jmquilli@hsc.vcu.edu
Sent: Thursday, September 09, 2004 10:59 AM
To: Sarata, Amanda (NIH/OD)
Subject: re: genet discrimination hearing

Dear Ms. Sarata,

Overall, I am writing to lend support to the Genetic Information Nondiscrimination Act, although I recognize there are valid arguments for and against the legislation as I understand it in its current form.

It is clear to me, as a cancer genetic counselor, that fear of genetic discrimination is a prevalent and influential concern among actual or would-be genetic counseling patients. I am not aware of genetic discrimination actually experienced by any of our patients. However, concern has been cited by many and has led to cancellation of appointments, decline of genetic testing, or payment of genetic testing / counseling out-of-pocket. This latter option is clearly a choice not available to everyone in an economically diverse environment, and this is a concern.

At least two arguments against the legislation as I understand it are:

*Potentially, genetic test results could be used to classify a person as low-risk (e.g., an informatively negative test result in someone with a strong family history of cancer); legislation that prohibits any use of genetic information would limit both the risks and benefits of genetic test results.

*Financial penalties for non-compliance with the legislation may be insufficient.

I also wonder if legislation will truly influence behavior or work to relieve fears. When citing protection from discrimination under our own state laws, I have had patients comment about the impermanence of laws (compared to the relative permanence of DNA and family histories).

If work on this legislation is consuming efforts that would otherwise work toward, for example, comprehensive coverage of genetic services for all, I would be in favor of shifting efforts to the latter.

Again, I hope it is understood that I do not support limiting access to healthcare services and tests based on genetic make-up. On the other hand, I am not fully convinced that the currently-drafted legislation is a panacea for access to these services.

Please feel free to contact me with any questions at (804) 628-1925.

Sincerely,

John Quillin, MS, MPH
Genetic Counselor



August 23, 2004

Ms. Amanda Sarata
Secretary's Advisory Committee
on Genetics, Health, and Society
6705 Rockledge Drive, Suite 750
Bethesda, MD 20892

Re: Hearing on Genetic Discrimination
October 18, 2004

Dear Ms. Sarata:

I have been informed that a hearing to learn about discrimination in health care or employment based on genetic information has been scheduled for October 18, 2004. I write to bring to your attention the case of an 11 year old boy with congenital primary hypothyroidism detected in the neonatal metabolic screening (NMS) program of the state of Florida who has been denied medical insurance.

As indicated by the attached correspondence, this lad who has no known genetic mutation and who has thrived physically and intellectually while receiving thyroxine, has been denied medical insurance for reasons unknown to this writer.

If this action is widespread, it poses a significant problem not only for this child but for other children detected in the state-sponsored NMS programs throughout this country. One of the prime objectives of these programs is to detect newborns with serious but treatable disorders before the underlying disease becomes manifest. That this objective has been realized in the case of congenital hypothyroidism is well documented by the many reports that document the greatly improved outcomes of these children relative to the pre-newborn screening era. If these clearly well children cannot obtain medical insurance because of their congenital disorder, then the programs are not fulfilling one of their primary obligations to these children, that is, to enable to

lead a normal and independent life. This includes the ability to obtain insurance of all types, including medical.

I hope that the Secretary will consider these problems and take appropriate measures to ensure that the children identified in NMS programs are able to obtain such insurance. Thank you for your attention to these comments. Please do not hesitate to contact me if you would like further details or additional comments.

Sincerely,

A handwritten signature in cursive script that reads "Allen W. Root".

Allen W. Root, M.D.
Professor of Pediatrics,
Biochemistry and Molecular Biology




Chairman
Newborn Metabolic Screening Advisory Council
Florida Children's Medical Service


UNIVERSITY OF
SOUTH FLORIDA
ST. PETERSBURG



MAILED
3/9/04

March 2, 2004

Ms. Lynn Anderson
Individual Medical Underwriting Department
Humana Insurance Company
Humana Inc.
Individual Product Segment
2 Riverwood Place
N19 W24133 Riverwood Drive
Suite 250
Waukesha, WI 53188

Dear Ms. Anderson:

I was appalled to receive your letter informing me that  was denied medical insurance because he is a boy with congenital hypothyroidism. The entire point of the state-sponsored newborn screening programs for congenital hypothyroidism is to identify and treat the newborn with this disorder before there are clinical manifestations of this illness in order to restore the infant to the euthyroid state. This has been one of the most beneficial public health projects of the past century.  is a prominent example of the success of this program. With thyroxine he has been growing and developing normally and is comparable to his peers in every respect (including his health) except for the need to take thyroxine. I know of no illness for which the patient with successfully treated congenital hypothyroidism is at greater risk than his/her peers. Thus, I write to request that you reconsider your decision concerning the denial of medical coverage for this child.

DIVISION OF PEDIATRIC ENDOCRINOLOGY, DIABETES AND METABOLISM • COLLEGE OF MEDICINE
University of South Florida • 801 Sixth Street South, Box 6900 • St. Petersburg, FL 33701
(727) 767-4237 • FAX (727) 767-4219

Thank you for your prompt attention to this matter.

Sincerely,



Allen W. Root, M.D.
Professor of Pediatrics
Professor of Biochemistry & Molecular Biology
Chairman
State of Florida
Neonatal Screening Advisory Council

cc: Endocrine chart
USFMC #713711
Jose Colon, M.D.
Neonatal Screening Program, Tallahassee
Neonatal Screening Folder - Attn: R.N.
Mr. and Mrs. Hector Torres
5823 Laguna Woods Court
Tampa, FL 33625-4140

February 26, 2004

DR. ALLEN ROOT
USF MEDICAL CLINIC
TAMPA FL 33613



[REDACTED]

PATIENT HEALTH CONDITION NOTIFICATION
Important Information from Humana

Dear Dr. Allen Root,

We recently considered an Application for Insurance on the above-named person. During review of this application, we received information regarding a history of congenital hypothyroidism, and yearly testing with the Endocrinologist.

Based on the risk of future medical claims associated with the conditions presented, and in accordance with Humana's underwriting guidelines, the indicated medical history is outside the acceptable parameters for an offer of coverage.

To prevent misinterpretation of this medical information, and to maintain the confidential physician-patient relationship, we believed it best to release this information to you rather than directly to the above named patient. Discussions concerning a patient's history are best handled on a doctor/patient basis. Please keep in mind that we evaluated the history from a risk selection viewpoint, not a clinical one.

In the event there are any errors in the above-listed history, please send any corrections, in writing, to our office. We will review these to determine whether it's possible to reconsider our decision in this matter.

Thank you for taking the time to discuss this matter with your patient.

Sincerely,

Lynn Anderson
Individual Medical Underwriting Department
Humana Insurance Company

reply to email re CAG SIG Communiqué -FRM Judie Much

Sarata, Amanda (NIH/OD)

From: Rae Wruble [raew@baptisthealth.net]
Sent: Wednesday, August 18, 2004 4:36 PM
To: Sarata, Amanda (NIH/OD)
Subject: reply to email re CAG SIG Communiqué -FRM Judie Much

To: SACGHS

c/o Amanda Sarata

This email is in response to a memo from Carol Demarco.

I am a health care provider who coordinates and runs the Genetic Risk Education Service for Baptist Hospital in Miami, Florida. I do all of the patient education. The majority of patients are concerned about genetic discrimination. They are concerned that future generations in their families will be harmed in some way by their being tested. They want to learn whether genetic discrimination is myth or reality. Of greatest concern to patients is that their test result will not be part of their medical record; none want a paper trail. They do not want the physician to note in their chart that genetic susceptibility testing was done, unless the result is "negative" for a deleterious mutation. Anonymous testing is done on every possible occasion. Physicians keep "confidential" sections in patients' charts. Our legal department has researched for cases of genetic discrimination regarding BRACAnalysis testing and have found none that went to trial.

Similar protection for persons with "individual" health insurance policies, like that for "group" health insurance policies would lessen the "fear" of genetic susceptibility testing. The test result is enough to handle without the additional concern of genetic discrimination. Patients want to feel that by taking the test and finding out the result that they are helping their families, not stigmatizing them.

Rae S. Wruble, R.N., MBA

Coordinator

Genetic Risk Education Service

Baptist - South Miami Regional Cancer Program

Phone: 786-596-2446

Fax: 786-596-2973

E-Mail: raew@baptisthealth.net

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Comments from the October 2004 SACGHS Meeting

American's Attitudes about Genetic Discrimination
Presented to the Secretary's Committee on Genetics, Health and Society
October 19, 2004

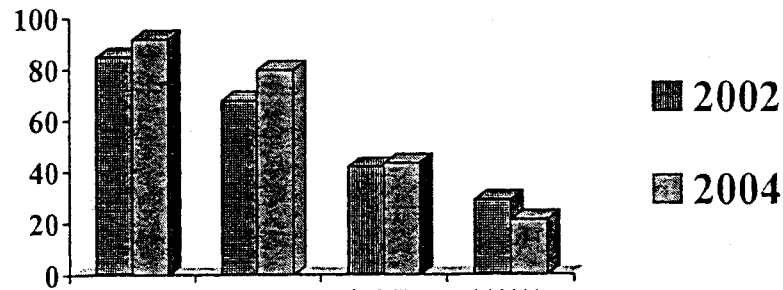
Kathy Hudson
Genetics and Public Policy Center
Berman Bioethics Institute
Johns Hopkins University

Thank you for inviting me to discuss public attitudes about genetic discrimination. My name is Kathy Hudson and I am the Director of the Genetics and Public Policy Center and Associate Professor in the Berman Bioethics Institute and in the Institute of Genetic Medicine at Johns Hopkins University. Established with a grant from The Pew Charitable Trusts, the mission of the Genetics and Public Policy Center is to be an independent and objective source of credible information on genetic technologies and genetic policies for the public, media and policymakers and to create the environment and tools needed by key decision makers to carefully consider and respond to the challenges and opportunities that arise from scientific advances in human genetics.

Genetic testing is undergoing tremendous growth. Scientists are identifying disease-causing mutations in humans at a rapid pace and developing tests to detect them. The number of tests has increased 10-fold over the last decade and there are now over 1000 tests available that give people information – information about that person, their health risk, and, potentially, their family's health risk. Genetic testing information can be used to guide decisions about risk management and treatment options. But the full benefit of advances in genetic testing will not be realized if people do not avail themselves of tests out of fear that the information will be used against them, instead of for them.

Over the past two years the Genetics and Public Policy Center has conducted extensive quantitative and qualitative research to assess the public's hopes and fears about genetic testing in the reproductive context. Today I will share with you what we have heard from Americans about their concerns about genetic discrimination.

We have surveyed over 6000 Americans about a broad range of genetic testing issues. In 2002 we conducted a random digit dial survey of 1,211 members of the general public and in April of 2004 we conducted an Internet survey of 4,834 individuals. One of the questions asked "In your opinion, if a genetic test shows that someone has a gene that increases the risk of disease, does that person's employer, insurance company, husband, wife or partner, or immediate family have the right to know?" As shown below, in 2002, 85% of the people surveyed stated that a person's employer had no right to know, and 68% thought the insurance company had no right to know. Those numbers went up in 2004 to 92% for an employer and 80% for an insurer. It is of interest to note that the percentage responding that insurers and employers should not have access to genetic test results was even higher among respondents who were aware of genetic testing before the survey and among those with higher education levels.



The Center also conducted 21 focus groups in 5 cities in the U.S. involving 181 members of the general public in April of 2003. Focus group participants were presented with scenarios involving reproductive genetic testing and asked what they thought the positive and negative effects might be to individuals, families, or society. Despite the fact that we did not ask specifically about genetic discrimination or mention it in the scenarios, many participants *spontaneously* voiced concerns about potential discrimination by insurance companies. Participants clearly worried that genetic testing would lead to a loss of insurance or the inability to obtain insurance coverage. A male focus group participant in California put it this way, “[I]f they would test and find out, say, a woman is more susceptible to breast cancer, would they deny her insurance later in life?”

Focus group participants also went on to speculate that the availability of reproductive genetic testing would result in denial of coverage for an affected child if the parents have chosen not to test or to terminate. The suggestion that prospective parents would face loss or denial of insurance led some to voice their worry about coercion to test and terminate. A woman in a focus group of evangelical Christians in Colorado said, “*I think that's a real fear, that health insurance companies might say, if you don't have this test, or you don't have this procedure, we are not going to pay to take care of that ill child that you have.*”

One of the criticisms of both quantitative and qualitative opinion research, especially in the area of biomedical and science policy, is that individuals are asked to comment on issues involving complex technologies which they may have had little opportunity to consider in depth. The public participation practice called Deliberative Democracy holds that people have more informed, reflective opinions if they first have a chance to learn

more about the subject, hear contrasting viewpoints from “the experts”, and engage in discussion with their fellow citizens about the issues.

So this summer the Center conducted an extensive public engagement activity based on the Deliberative Democracy model called *The Genetic Town Hall: Making Every Voice Count*. Using two different methodologies, the engagements took place in six U.S. cities using a town hall format, and with 15 discussion groups online using state-of-the-art Internet meeting capability. Over 500 citizens in Sacramento, CA; Seattle, WA; Kalamazoo, MI; Fort Worth, TX; New York City, NY; and Nashville, TN took part in the in-person town halls and over 133 citizens took part in the on-line version. Participants in both the on-line and in person Town Halls were asked to consider three major issues in reproductive genetic testing - acceptable uses, the safety and accuracy of these tests, and the impact on individuals, families, and society. Background information about the technology and views of a diverse group of medical experts, policymakers, bioethics scholars, and the clergy were provided through an educational video series developed by the Center entitled *Chosen Children: Issues in Reproductive Genetic Testing* to ensure that the content delivered in each Town Hall was the same. Participants were queried at various times before, during, and after the sessions to document changes in attitudes as a result of participating in the Town Hall and engaging in discussion with their fellow citizens about issues of concern to them.

Two of the questions posed to the participants were “What do you think are the factors to consider when setting limits for the use of Reproductive Genetic Testing?” and “What are some of the possible benefits and harms to individuals, families, or society, of parents’ ability to choose and select characteristics of their children?” Participants were given time to discuss these questions with their tablemates after which each table “called out” the issue most of concern to those at that table. If a concern had already been mentioned by another table, they moved to the next on their list. All of the concerns were projected on a screen so everyone in the room could see and discuss them. Then, using electronic keypads, the participants ranked them in order of priority. A similar format was used in the on line Town Halls in which the question was discussed, the set of concerns collected, and the participants voted on which were of highest priority.

The issue of genetic discrimination based on genetic test results came up as an issue of concern *in every single town hall*. In fact, in Sacramento and New York, genetic discrimination ranked as the *number one issue of concern* when considering the potential harm of reproductive genetic testing to individuals, families, or society. In Seattle, Fort Worth, and Nashville it ranked second.

In Sacramento every single table listed as a concern how genetic tests and their results would affect the basic issues of jobs and paying for health care.

“Will you have trouble getting a job because you have this gene that may cause cancer, whether or not you have cancer?” Sacramento Town Hall

Like the focus group participants, Town Hall participants pointed out that if insurers paid for tests, they would want to know results, and these could affect what an insurer would be willing to cover. In New York some feared that insurance coverage would implicitly guide reproductive choices through economics, i.e., they would refuse coverage for children with disabilities where the disability could have been detected through testing and the birth avoided. As one Nashville participant said,

“Discrimination *will* arise from genetic testing.”

In conclusion, our research shows that an overwhelming majority of Americans do not want insurers or employers to have access to genetic test results and there is widespread concern about genetic discrimination. This high level of public concern suggests that when legislative protections are enacted to prohibit genetic discrimination, it will be necessary to have public information campaign to insure that citizens are aware of their rights and to make sure that fear of discrimination does not adversely affect genetic testing decision-making.

Testimony of Jane Massey Licata, Ph.D., J.D.
Partner, Licata & Tyrrell P.C. and
Adjunct Professor of Law
Rutgers School of Law- Camden
before the Secretary's Advisory Committee on Genetics,
Health and Society
October 18, 2004

Federal law does not provide adequate protection to deter genetic discrimination. The only federal law which directly addresses genetic discrimination is the Health Insurance Portability and Accountability Act (HIPPA) which was enacted in 1996. While HIPPA does prohibit certain group plans from using any health status-related factor, including genetic information, as a basis for denying or limiting eligibility for coverage or for charging more for coverage and specifically states that genetic information in the absence of a current diagnosis does not constitute a preexisting condition, it does not prevent insurers from collecting genetic information or limit disclosure of genetic information about individuals to insurers. HIPPA also does not prevent insurers from requiring applicants to undergo genetic testing. The individual insurance market and some group plans are not covered by HIPPA.

The most likely current source of protection against genetic discrimination in the workplace would be the American with Disabilities Act (ADA), which is enforced by the Equal Employment Opportunity Commission (EEOC), and similar disability-based nondiscrimination laws (i.e. the Rehabilitation Act of 1973). However, these laws do not explicitly address genetic information. While the ADA does protect individuals with symptomatic genetic disabilities, it also allows an employer to obtain extensive medical information about a person that is under a conditional offer of employment, including obtaining and storing genetic samples, requiring genetic screening as a condition of employment or to purchase genetic information about applicants from a genetic information data bank and once employed, to request medical information that is job related and consistent with business necessity. The ADA does not deal with unaffected carriers of a disease who may never get the disease, individuals with late onset genetic disorders who may be identified through genetic testing as being at risk for developing a disease, or others identified through family history as being at high risk for developing the disease. ADA also does not protect workers from requirements or requests to provide genetic information to their employers. While the EEOC has attempted to provide ADA protection to individuals who do not have symptomatic genetic disabilities but who may be subject to

discrimination based on genetic information, its guidance is limited in scope and legal effect. It is policy guidance that does not have a legally binding effect. It should also be understood that cases based on the argument that an employer has discriminated against a worker by regarding them as disabled (which would be the argument in the case of an asymptomatic genetic disease or a carrier) have not been well received by the courts, so reliance on the ADA to protect victims of genetic discrimination is misplaced.

While Title VI of the Civil Rights Act could provide a basis for an argument that genetic discrimination based on racially or ethnically linked genetic disorders constitutes unlawful race or ethnicity discrimination, there are very few genetic diseases for which it would be relevant.

Most states have enacted legislation on genetic discrimination in health insurance (41) and in the workplace (31). However, state law is highly variable in scope and nature of protection. As a practical matter, it would be highly desirable to have a consistent legal threshold and then allow state law to address additional areas of concern. A review of enacted state legislation concerning genetic information and the workplace reveals a wide variety of approaches, however only 3 state statutes would actually create a greater liability for employers than the Senate compromise bill which was passed in October 2003. In those cases, provisions for a private right of action, punitive damages and/or prison terms are provided for.

The Genetic Nondiscrimination Act of 2003 was passed unanimously by the Senate in October 1993. The Act amends the Employment Retirement Income Security Act of 1974 (ERISA), the Public Health Service Act (PHSA), the Internal Revenue Code (IRC) and Medicare coverage provisions to prohibit discrimination based on genetic information. It applies to all (any size) ERISA plans whether insured or self-insured including those offered to federal, state and local government employees, by health insurance carriers and HMOs and church-based plans and Medicare supplemental polices. The House has yet to consider similar legislation although proposals are pending before 3 different committees. An important aspect of the bill passed by the Senate (S.1053) is that it focuses on protecting the insured/worker and their families while a claim of nondiscrimination is being evaluated. The greatest fear is of losing insurance or a job, so by allowing the insured/worker to maintain the status quo, this concern is alleviated. There is no private right of action, but rather a clear prohibition of discrimination based on genetic information and a regulatory process to assure compliance and address complaints. The goal of the legislation is to provide rules for conducting business where genetic

information may be involved and reasonable penalties for failure to comply. The philosophy of the legislation is to encourage responsible behavior and compliance by the insurer/business.. For example, the per day penalties for noncompliance are quite modest (\$100/day) and the maximum liability is capped (\$500,000). Therefore, like other effective federal regulatory schemes, the goal is to encourage compliance (i.e., protection of highly confidential and potentially damaging information if it was placed in the wrong hands or used for a purpose not related to business necessity or medical treatment or wellness programs). As experience with HIPPA has now shown, it is possible to protect genetic information in an effective and cost effective manner in the normal course of business. It required education, commitment and clear regulatory requirements, but it has been achieved. Extending similar coverage into the entire group and individual health insurance market and the workplace is a realistic and achievable objective.

The time is now for federal genetic information nondiscrimination legislation. The compromise bill passed by the Senate in 2003 addresses some of the most urgent needs in protecting an individual's privacy and in assuring access to genetic testing and services. Until recently, access to this type of testing was limited to those who could afford to pay for it privately. By paying for it themselves, they could also have greater assurance of confidentiality concerning the testing and the results. While wider acceptance of the need and validity of genetic testing has made insurers more comfortable with reimbursement for this type of service, there is a huge risk to the insured or employee that very sensitive information, which could easily be subject to misinterpretation, may be widely distributed as a part of the insurance information system. Genetic information provides information not just about the insured/employee, but also their parents, siblings and children which impacts not only on individual but also on family privacy. The potential for misunderstanding is so great that it is essential that we establish a national policy for the protection of an individual's privacy interest in their genetic information as soon as possible.

Jane Massey Licata has been a biotechnology patent and FDA attorney for almost two decades and teaches patent and FDA law at Rutgers School of Law. Trained as a scientist (Ph.D. 1978), her interest in genetic privacy is both academic and practical. She has written and taught about ethical issues in research, served on and advised institutional review boards on genetic and biotechnology research issues, evaluated genetic technology for venture capital firms and

pharmaceutical companies, filed over 2000 patent applications concerning biotechnology inventions, most involving the use of genetic information to develop new diagnostics and therapeutics, and negotiated hundreds of research agreements concerning genetic research. Her expertise and experience for the nonprofit research as well as the business sector has led to an understanding of the ethical and economic need for federal legislation to address genetic nondiscrimination and privacy.

**Secretary's Advisory Committee on Genetics,
Health, and Society**

**Fifth Meeting
October 18-19, 2004**

**Statement of Joanne Armstrong, MD, MPH
Senior Medical Director, Women's Health
Aetna**

On Behalf of

America's Health Insurance Plans

Thank you for inviting me to testify regarding this important issue. I am Joanne Armstrong, Senior Medical Director, Women's Health, for Aetna, one of the nation's leading providers of health care and related group benefits. I am testifying today on behalf of America's Health Insurance Plans (AHIP) and its nearly 1,300 member companies.

America's Health Insurance Plans is the national trade association representing the private sector in health care. AHIP's member companies provide health, long-term care, dental, vision, disability, and supplemental coverage to more than 200 million Americans. Aetna serves approximately 14.4 million health care members, 11.9 million dental members, and 12.0 million group insurance customers through a nationwide network of more than 527,000 health care services providers, including approximately 321,000 primary care and specialist physicians and 3,300 hospitals.

Genetic medicine is not new. The number of human chromosomes was discovered in 1956 and the chromosomal abnormalities associated with conditions such as Down Syndrome and certain types of cancer such as Chronic Myelogenous Leukemia (CML) were described by 1960. Non-DNA based tests for screening and diagnosis of genetic conditions such as abnormal hemoglobin production that results in sickle cell disease and newborn screening tests for metabolism errors have been available for decades.

The *rate* of new genetic discoveries that help us understand the basis of health and disease has advanced at a dizzying pace in recent years. The speed of new breakthroughs in genetic knowledge, however, is challenging the ability of our health care systems to effectively integrate these discoveries into clinical practice and optimize their benefit to prevent, and possibly cure, disease.

Because of the complexity of genetic information, the optimal use of genetic technologies requires informed providers and patients and the coordination of services across a complex array of delivery systems. Unfortunately, much work is needed in all of these areas. The front line providers of genetic services, primary care physicians, acknowledge

that they are not adequately prepared for the age of genomic medicine. For example, 72 percent of non-genetics physicians rate their knowledge of genetics as fair to poor.¹ Patients are also not adequately prepared to navigate these waters. Fully 82 percent of consumers cannot correctly answer most genetic medicine knowledge questions in national surveys.² As with the adoption of other medical services and technologies, health plans are and will continue to be instrumental to the coordination of this care. This process in genetics has already started. For example:

- Genetic counseling in reproductive health has been long promoted by health plans to encourage members to get the most appropriate information to inform reproductive decision-making. When genetic counseling is performed by trained genetics counselors compared to physicians not formally trained in genetics, a greater number of genetic risks that result in clinical decision-making are identified.³
- Health plans like Aetna have taken the lead in the development of policies that incorporate the promotion of formal genetic counseling into predisposition genetics testing to insure appropriate member education about this test.

A promising new area of genetic testing will utilize test results to guide pharmacogenetic therapies or the duration of therapy. So called “selected” or “targeted” therapies hold out the promise of directing therapies to individuals who may benefit from them while avoiding harmful side-effects and costs associated with treatments that will not benefit individuals. Examples of this model exist today for breast and colon cancer. Viral genotyping is used to determine the appropriate duration of treatment for Hepatitis C. Adherence to evidence-based guidelines for Hepatitis C therapy is not optimal. Health plans have demonstrated success in improving member adherence with evidence-based recommendations for Hepatitis C and other drug therapies.

¹ Menasha J, Schechter C, Willner J, *Genetic Testing: A Physician's Perspective*, Mt. Sinai Journal of Medicine, 67(2):144-151 (March 2000).

² Public Awareness and Attitudes About Genetic Technology, Princeton Survey Research Associates for Genetics and Public Policy Center, (November 5, 2002) www.dnapolicy.org/research/index.html

³ Koscica K, Canterino J, et al, *Assessing Genetic Risk: Comparison Between the Referring Obstetrician and Genetic Counselor*, American Journal of Obstetrics and Gynecology, 185:1032-34 (November 2001)

As the science of genetics advances, concerns over protecting genetic information from inappropriate uses have escalated. There is a growing public awareness about the health benefits that can be derived from genetic information and concern about the potential for misuse of such information. We must, however, engage in responsible policymaking on these issues and not unnecessarily restrict the use of information needed to promote appropriate health care decision-making.

I would like to address my remarks today to these key issues: (1) the current use of genetic information by health insurance plans and health care providers; (2) the role of health plans in providing decision-support information for patient and physicians; and (3) the adequacy of existing laws that prohibit the misuse of medical information, including genetic data.

How Health Insurers Use Genetic Information: Their Role in Providing Decision Support

Health insurance plans are committed to helping their members and insureds access the highest quality care possible through the provision of appropriate information, risk assessment, disease management, and other quality assurance systems. Genetic information is just one of many forms of medical data that clinicians and plans have been using to assist in the coordination of health care services.

- For example, screening tests are available for Hereditary Non-Polyposis Colorectal Cancer (HNPCC), an inherited predisposition to develop colorectal cancer. Individuals with this syndrome have an 80 percent lifetime risk of developing the disease and require earlier and more frequent screening tests to prevent cancer. Access to this information allows health plans to create deviations in standard preventive service benefits and insure that at-risk enrollees have access to earlier and more frequent colorectal cancer screening services than

that which is recommended for the population at large. This is an added value to these members.

- Another common example of the use of genetic data by health plans today is in the coordination of laboratory services. Physicians and members frequently call to inquire where specific laboratory tests can be performed within contracted networks.

Health insurance plans have learned that a key component of health care delivery is to make certain that patients and health care providers have the information they need to make informed decisions. Genetic information is increasingly relied upon by clinicians and patients in their decision-making. Health insurance plans are facilitating this information sharing in a number of ways:

Encouraging Appropriate Testing and Counseling: Individuals at risk of certain genetic conditions for which there are specific interventions for prevention or treatment are encouraged to undergo counseling to determine whether genetic testing is appropriate for them. Appropriate genetic tests are those that provide information that may positively affect the course of an individual's treatment. Health insurance plans can play an important role in facilitating access to genetic counseling (by their physician or other appropriate professional) to assist these individuals in determining the best course of treatment based on the results of genetic tests.

Aetna has been a leader in the promotion of genetic counseling services to assist members in the understanding of genetic conditions, the interpretation of test results, and their options based on test results. As an earlier adopter of BRCA testing, for example, Aetna included genetic counseling services as an integral part of testing. Our plan covers genetic testing for a family member not insured if the results affect the course of treatment of our member.

There is, however, a critical shortage of qualified genetic medicine clinicians in the United States. This scarcity may threaten the optimal use of these value services. Health

plans have created innovative approaches to improving access to quality genetic counseling and information such as telephonic counseling.

Using Genetic Test Results to Promote Preventive Screening and Disease

Management: Health insurance plans play a vital role in providing access to appropriate preventive screening and disease management programs for individuals who have tested positive for a genetic disease or predisposition. As noted, by promoting more frequent preventive screenings, health insurance plans are able to improve outcomes for individuals who have the gene for the familial form of colorectal cancer. Similarly, women who test positive for BRCA may be appropriate candidates for MRI screening of the breast and not conventional mammography. MRI screening is not recommended for low-risk women. As scientists acquire a greater understanding of the role genes play in disease and developing more genetic therapies and possibly even cures, preventive screening and disease management programs will become even more important.

Supporting Consumer Education and Patient Awareness: It is essential to translate our growing genetic knowledge into practice through the provision of medically appropriate health care. A number of studies have demonstrated that patient knowledge of health care – and in particular the impact of genetics on health care – is lacking. Health insurance plans can partner with the health care provider community to help consumers understand the appropriate use of genetic testing and its results. Many plans have created high quality electronic genetic information to fill the gap.

Laws That Prohibit the Inappropriate Use of Genetic Information

AHIP and its member companies strongly believe in the importance of protecting genetic information from illegal or inappropriate uses. One of the unfortunate myths about genetic information is that health insurance plans may use such information to deny insurance coverage or may disclose genetic information inappropriately. In fact, health insurance companies have many years of experience handling genetic information of their enrollees (for example, results of screening tests such as amniocentesis or neonatal

screening), with little, if any, empirical evidence that genetic information has, or is being, misused.

As a matter of practice, health insurance plans do not use or disclose personal health information for purposes except as required for payment activities such as claims payment or health care operations. Moreover, federal and state laws currently prohibit the inappropriate use of genetic information.

Laws Prohibiting Genetic Discrimination

The Health Insurance Portability and Accountability Act (HIPAA) prohibits employers and health insurance plans in the group market from using the results of a genetic test to deny coverage or set premium rates for group health plans. The federal law, which was passed by Congress in 1996, was based on a number of long-standing state laws which prohibit medical underwriting in the group health insurance market. The majority of Americans with health insurance coverage receive such coverage through employer-sponsored plans that are subject to these HIPAA protections. HIPAA specifically prohibits group health insurance plans from:

- Refusing to cover employees or their family members based on genetic information.
- Refusing to renew coverage based on genetic information.
- Charging employees and family members higher premiums based on genetic information.
- Imposing pre-existing condition waiting periods based on genetic information.
- Canceling coverage based on genetic information.

The individual health insurance market is considerably smaller than the group market and, as a result, works much differently. Given the voluntary nature of individual health insurance and the smaller risk pools that are typically covered by an individual health

insurance plan, most states allow underwriting of individual health insurance coverage to protect the viability of the market. It is important, however, to understand that:

- Health insurers in the individual market do not ask people seeking coverage to provide presymptomatic genetic test results.
- Health insurers in the individual market do not use genetic information in the underwriting process.
- Once issued, an individual health insurance policy cannot be cancelled for any health-related reason including genetic predisposition to a disease. State insurance rating laws prohibit singling people out for renewal rate increases based on health status-related factors, which includes genetic information.

Health Information Privacy Laws

Protecting the confidentiality of all health information – not just genetic information – is critical to preserving open and honest communications between health care providers and their patients. We believe consumers should be able to benefit from coordinated, integrated, health care delivery systems while being protected against unlawful disclosures of genetic information. Health insurance plans have long standing policies and procedures in place to protect the privacy of health information.

The HIPAA privacy rule, enforced through the Department of Health and Human Services' Office for Civil Rights, strictly limits the collection, use, and disclosure of genetic information by all health insurers (group and individual) and health care providers for purposes other than treatment, payment, and health care operations without an individual's written authorization. In addition, a number of state privacy laws impose similar restrictions on the use and disclosure of health information by health insurance plans.

Conclusion

AHIP and its member companies believe that genetic information can help providers and their patients make informed health care decisions. Today, there are genetic tests that, based on sound scientific evidence, have been shown to improve clinical outcomes. Health insurance plans have an important role to play in promoting the appropriate use of genetic tests by encouraging evidenced-based counseling and testing, supporting consumer education and patient awareness, and using genetic test results to enhance preventive screening and disease management.

For many decades, health insurance companies have demonstrated responsible use and management of genetic information. Health insurance plans strongly support protecting all patient-identifiable health information, including genetic information, from unauthorized disclosure and other illegal uses. As the science of genetics advances, we are committed to facilitating access to genetic counseling and appropriate genetic testing, and guarding against the misuse of genetic information.

Society for Human Resource Management

**Testimony of
Michael P. Aitken
Director, Governmental Affairs**



Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS)

Monday, October 18, 2004

Good morning, my name is Michael P. Aitken and I am honored by the invitation and grateful for the opportunity to provide commentary to the Secretary's Advisory Committee on Genetics, Health, and Society regarding the scope and nature of genetic discrimination.

I appear today on behalf of the Society for Human Resource Management (SHRM). SHRM is the world's largest association devoted to human resource management. Representing more than 190,000 individual members, the Society's mission is to serve the needs of HR professionals by providing the most essential and comprehensive resources available. As an influential voice, the Society's mission is also to advance the human resource profession to ensure that HR is recognized as an essential partner in developing and executing organizational strategy. SHRM serves the needs of the human resource management professional by providing the most essential and comprehensive set of resources available. In addition, the Society is committed to advancing the human resource profession and the capabilities of all human resource professionals to ensure that HR is an essential and effective partner in developing and executing organizational strategy.

SHRM is well positioned to provide unique insight on the probable effect that genetic non-discrimination legislation will have on the workplace. I have served SHRM as the Director of Government Affairs for little over a year. Previously, I was the Associate Executive Director for External Relations for the College and University Professional Organization for Human Resources for 13 years.

My remarks will focus on the potential impact genetic nondiscrimination legislation will have on employers, employees, and their organizations. SHRM believes that employment decisions should be based on an individual's qualifications, including education, experience, and demonstrated competence and ability to perform a job, not on the basis of characteristics that have no bearing on job performance. Therefore, SHRM strongly opposes employment discrimination on the basis of an individual's genetic information. The Society also believes however, that any legislative remedy proposed must be carefully drafted so as not to be overly broad, thereby leading to unintended consequences. These unintended consequences would include the interaction of new genetic nondiscrimination legislation with existing federal and state employment and benefits laws as well as existing nondiscriminatory employer practices.

As a result, I will discuss the interplay that any proposed legislation is likely to have on current federal laws such as; the Family and Medical Leave Act (FMLA), the Americans with Disabilities Act (ADA), Title VII of the Civil Rights Act of 1964, and the privacy regulations of Health Insurance Portability and Accountability Act (HIPPA).

Introduction

The completion of the Human Genome Project has resulted in the mapping of the 23 pairs of human chromosomes, and the identification of numerous genes responsible for various medical conditions. Access to this important information holds great promise for the early detection, treatment and prevention of many human diseases. Yet, at the same time many legal concerns have surfaced about the potential misuse of genetic information.

Genetic tests currently may be used to detect predispositions to certain medical conditions and diseases, and may soon identify genetic links to innate behavioral and personality traits. Concern

exists that individuals who have a predisposition to certain diseases or conditions, or with medical conditions in their family background, may find themselves at a risk of being stigmatized as an economic or safety risk for employment, and thus face discrimination in employment decisions.

As stated above, SHRM believes that all employment decisions should be made on the basis of an individual's qualifications such as education, experience and demonstrated competence, not on the basis of characteristics that have no bearing on job performance. For this reason, SHRM would oppose employment policies that permit employment decisions to be made based on an individual's genetic information.

Current Law

Despite the fact that there is no evidence to suggest widespread possession or use of genetic information by employers, there is interest in enacting legislation to that would codify current protections against genetic discrimination offered by the ADA (as articulated by the Equal Employment Opportunity Commission's (EEOC) 1995 Guidance on Disability), as well as to fill the gaps left un-addressed by current law. Legislation banning genetic discrimination in the workplace may help to increase participation in clinical trials and research.

Under the current federal framework, there are several statutes that could potentially provide protection against genetic discrimination. However, these laws remain largely untested in the courts. Given that some genetic diseases have been found to be more prevalent in certain racial and ethnic groups (such as sickle-cell anemia in individuals of African descent or Tay-Sachs disease in Ashkenazi Jews), Title VII of the Civil Rights Act of 1964 may serve to prohibit genetic discrimination against a member of these groups. Since Title VII prohibits employment discrimination against members of a protected class, using genetic information which is highly correlated with the race, ethnicity, national origin or gender of an employee may be prohibited under this law. To date, at least one case supports employment discrimination claims based on genetic information under Title VII. The U.S. Court of Appeals of the Ninth Circuit held in *Norman-Bloodsaw v. Lawrence Berkeley Laboratory*, N.D. Cal., No. C95-3220 VRW that mandatory pre-employment genetic testing, performed without consent, may amount to an adverse impact under Title VII, since the claimants were tested for genetic markers based on their protected status. However, the protections afforded by Title VII are not comprehensive, and case law remains limited.

Although it does not explicitly address the genetics issue, another federal statute that many argue offers protection against discrimination is the Americans with Disabilities Act, or ADA. According to EEOC interpretation from its 1995 Guidance on Disability, genetic discrimination is prohibited under the third part of the statutory definition of the term "disability," which protects individuals who are "regarded as" having impairments that substantially limit one or more major life activities. This prong of the ADA is designed to protect against myths, fears and stereotypes about people with disabilities, and reflects recognition by Congress that the reactions of others to impairment or a perceived impairment should be prohibited the same way as discrimination based on an actual impairment.

In fact, the EEOC in 2001 filed a genetic discrimination suit against Burlington Northern Santa Fe Railroad (BNSF) in its genetic testing of employees who were filing claims for work-related carpal tunnel syndrome. BNSF workers were never told that they would be tested to see if they

had a genetic marker that made them more susceptible to carpal tunnel syndrome, and only learned of the program when one employee suffering from the condition visited the company doctor. The employee chose not to take the test and then was threatened by BNSF with dismissal if he continued to refuse to take the test. The EEOC sought a preliminary injunction to stop the testing of employees who filed claims for worker-related injuries, because it believed the tests were not job-related or consistent with any business necessity requirement under the ADA. Additionally, the EEOC believed that to base any employment action on the results of the tests would amount to unlawful discrimination. Although the case was not decided on the application of the ADA to a genetics issue, the suit was settled not long after it was brought.

Another case that might have some bearing on whether genetic discrimination is covered by the ADA is the 1998 U.S. Supreme Court ruling in *Bragdon v. Abbott* on ADA protection of asymptomatic medical conditions. In that case, an individual infected with the HIV virus who had not yet exhibited many of the manifest symptoms of AIDS was ruled to be “disabled” under the ADA. The Supreme Court reasoned that a physical impairment existed, based upon virus-related changes that occurred at the cellular and molecular levels after infection, even if the effects of these changes were not yet externally visible. Although it is unclear whether the courts will rule similarly in cases involving genetic issues, *Bragdon v. Abbott* may strongly influence future rulings in cases involving the presence of genetic markers for a currently asymptomatic genetic disorder.

On Feb. 8, 2000, President Bill Clinton issued an executive order banning genetic discrimination by federal agencies against their employees (Executive Order 13145). The order prohibits federal employers from obtaining, using or disclosing protected genetic information as a part of any hiring, promotion or discharge action. Such protected genetic information is defined to include information about the genetic tests of an individual or his/her family members, as well as information about the occurrence of a disease or medical condition in an individual’s family. Information about an individual’s current health status—including physical exams, or chemical, blood or urine analyses—is exempted from this definition. The order does provide limited exceptions to the privacy mandates, such as when such information would help ensure workplace health and safety, or if presence of a genetic condition could prevent the employee from performing essential job functions.

Potential Areas of Conflict with Proposed Genetic Nondiscrimination Legislation and Federal Law

Should a new federal genetic discrimination law be enacted, it is essential that it is developed to reflect the requirements and protections of existing employment statutes and that it not conflict with current laws or disrupt existing nondiscriminatory employment practices.

From a practical perspective, there is always concern that new employment legislation will be drafted in a vacuum; that consideration will not be given to its impact on and its interaction with existing laws. For example, the interrelationship and interaction among the ADA, FMLA and state workers’ compensation law, all of which impose different legal requirements—help demonstrate this problem. Because each law was passed at a different time and has a different policy objective, an employer’s efforts to comply with one law can easily cause it to violate a provision of the other laws.

Employment laws are most effective when compliance with one federal or state law does not contradict the purpose of other laws or does not require employers to violate one law to satisfy another. Any genetic nondiscrimination legislation must be balanced, objective, and developed with existing law in mind. It is imperative to the development of sound law, for public policy decision-makers to consider the requirements and implications of existing federal and state employment and benefits laws when considering various genetic nondiscrimination proposals. An opportunity does exist to enact legislation that provides meaningful protections for employees, that builds on employers' general support for such legislation, and does not further exacerbate the patchwork quilt of conflicting employment laws that currently exists at the federal and state levels.

As expressed above, SHRM is concerned about the interaction of proposed genetic discrimination legislation with previously enacted employment legislation, specifically but not limited to the ADA, FMLA, Title VII, workers' compensation, HIPAA privacy regulations, and the impact on the administration of employer provided health plans.

A fundamental element of each is the collection of an employee's medical information, which often may include documenting family medical history. This information is required for example, to ensure employer compliance with the FMLA. Under previous legislative proposals offered however, such information may be deemed "protected genetic information," and knowledge or use of the information may create liabilities for employers.

Americans with Disabilities Act

Under the ADA medical records may be used to help determine if an employee has an "impairment" that substantially limits one or more major life activities, or has a "record of" such a substantial limiting impairment. Moreover, medical information is often an integral part of determining a reasonable accommodation of disabled employees. Since employers are required to determine whether or not an employee or an applicant has "disability" within the meaning of the law, the employee's or the applicant's medical information is often required. HR professionals and employers would face an insurmountable challenge in making proper decisions without this information.

Yet previous legislative proposals would impede the collection of relevant medical information and could seriously impact an employer's ability to assist individuals with disabilities in the workplace.

Family and Medical Leave Act

The Family and Medical Leave Act (FMLA) creates a similar problem. As you know, the FMLA allows an employee to take up to 12 weeks of unpaid leave for a "serious health condition" or the serious health condition of a spouse or family member. Serious health condition is defined as an illness, injury, impairment, or physical or mental condition that involves one of the following: (1) inpatient care; (2) continuing treatment plus absence from work for more than three calendar days; (3) pregnancy, (4) chronic conditions requiring treatment; (5) permanent or long-term conditions requiring treatment; and (6) multiple treatments for non-chronic conditions. (29 CFR 825.114).

In order for an employer to determine whether an employee qualifies for FMLA leave for the treatment of a serious health condition—whether that condition is manifested by the employee or by a family member—the employer must collect relevant medical information on the nature of the serious health condition. This medical information may very well indicate a genetic based health condition.

For example, an employee may request intermittent leave to assist her ailing mother in receiving radiation treatment for diagnosed breast cancer—a “serious health condition”—and a disease with a known genetic component. In granting this leave request, the employer has just acquired genetic information, a feasible violation under several legislative proposals that have advanced previously at the federal and state levels. Or, if an employee seeks to take leave to care for a child suffering from sickle-cell anemia, a serious hereditary disorder, the employer upon receipt of the leave request will inadvertently obtain genetic information about that employee or spouse of the employee—again a potential violation under earlier previous legislative proposals.

Genetic discrimination legislation that restricts the flow of all medical information may put employers into a real bind—how do they grant FMLA leave needed by employees without collecting potential genetic information?

State Workers' Compensation Laws

The interplay of legislation and the various state workers' compensation laws will create more challenges for employers. Under state workers' compensation laws, medical information is necessary to file a claim and is used to determine whether or not an injury is work-related. Would employers still be able to collect this information? Any legislative proposal on genetic nondiscrimination must include an exception for workers' compensation related claims.

It is imperative that legislative efforts be focused on prohibiting the discriminatory use of genetic information, not on the flow of such information.

Title VII of the Civil Rights Act of 1964

The thrust of Title VII is to prohibit discrimination on the basis of sex, race, color, creed, national origin and religion. However, Title VII may incidentally provide protection against some forms of genetic discrimination. It is plausible to imagine that a claim could be brought alleging that genetic discrimination based upon racially or ethnically linked genetic disorders constitutes unlawful race or ethnicity discrimination.

Title VII covers all aspects of employment from hiring and advancement to benefits and advancement. Therefore, under the current scheme of Title VII, employers are already prohibited from using employees' gender or in calculating either the amount of benefits to which male and female employees are entitled or the amounts that male and female employees would be charged for those benefits. This applies to both health and retirement benefits; even though there is documentation that women live longer than men and that certain conditions afflict some genders or religious and ethnic groups more often than others.

While it is possible, it remains to be seen if a claim of genetic discrimination under Title VII will prove viable. As discussed earlier, there has only been a strong nexus between race and national origin for a few diseases, and this nexus would be essential for a genetic discrimination claim.

Title VII protection of genetic discrimination will be available only when an employer engaged in discrimination against a particular race or ethnic group based on a genetic trait that is substantially related to the race or ethnicity of the group.

Health Insurance Portability and Accountability Act and Employer Sponsored Wellness Plans

In 1996, Congress addressed the issue of genetic information for group health insurance in the Health Insurance Portability and Accountability Act, or HIPAA. HIPAA is the only federal law that directly addresses the issue of genetic discrimination and includes protections for coverage under group health plans. These protections limit exclusions for preexisting conditions and prohibit discrimination against employees and dependents based on their health status. In other words, the law prohibits denial of benefits or increase of premiums to individual members of a group health plan due to health status.

HIPAA currently permits a group health plan to disclose health information to an employer that sponsors the plan, provided the information is used only for plan administration purposes and the employer has in place certain specified safeguards. This ensures the employer or plan sponsor's legitimate need for access to some health-related information. For example, an HR professional may encounter genetic information in the administration of a health care plan, such as providing claims assistance to an employee. In this instance, an employer has acquired genetic information without intent to harm, simply through administering this employee benefit. For this reason, SHRM believes any legislative proposal that includes employer liability for possessing genetic information should include a "safe harbor" exemption for employers who receive genetic information inadvertently through the benefit administration of health care plans.

Employer-sponsored wellness programs are another instance where employers may uncover genetic information. Faced with their fourth straight year of double-digit increases in health care premiums, many employers have implemented wellness programs to improve the overall health of their workforce and control costs. Establishing a wellness program often involves a confidential, individualized health risk assessment for the employee that provides him/her with a roadmap on how best to lower health risks. However, in conducting the risk assessment, information is collected that may include family history, blood samples for cholesterol screening, and other potential genetic information. Employers offering wellness programs are not conducting these wellness programs to gather genetic information on employees but rather simply trying to improve the health and safety of their workforce.

A final area of concern for HR professionals with regard to legislative proposals to address genetic discrimination is the overly broad definitions of "protected genetic information." Definitions that include information derived from family histories, such as cholesterol, high blood pressure, and cancer, could limit health plans' abilities to carry out disease management, quality assurance, wellness, and other important programs.

Similar to the law and employer practice examples, an employer may also inadvertently acquire potential genetic information through the "water cooler" scenario. Proposals that include an overly broad definition of genetic information could turn the casual conversations about loved ones around the water cooler into a litany of costly litigation and workplace disputes. For example, it is not uncommon for colleagues to share personal information about the health status of their family members with each other in the workplace. Let's say an employee voluntarily

discloses to the employer the genetically related health care condition of the employee's family member. Or, the example of an employer reading a newspaper obituary or learning in the office that an employee's relative died due to an illness which may have genetically related health care implications.

In each of these instances, it was not the employer's intent to seek out the potential genetic information of the employees in these two above examples. Nevertheless, an employer that simply "possessed" this information whether or not the employer ever acts on the information could be exposed to future liability if legislative proposals to prohibit genetic discrimination focus on controlling information only and not on the "intent" of the employer. Therefore, legislative proposals must differentiate between the mere possession of genetic information and the use of this information for discriminatory purposes.

SHRM Recommendations

- SHRM believes that employment decisions should be based on an individual's qualifications and ability to perform a job, not on the basis of characteristics that have no bearing on job performance. Therefore, we strongly oppose employment discrimination on the basis of a person's genetic makeup.
- Possession of genetic information must be differentiated from the use of this information for discriminatory purposes. Any proposed statute should be directed at controlling discriminatory conduct, rather than attempting to regulate the flow of information. As we like to say, genetic discrimination is about discrimination, not genetics.
- We believe that genetic discrimination is wrong, and if a company intentionally discriminates, remedies should be available. However, SHRM opposes legislation that would provide unlimited punitive and compensatory damages for victims of genetic discrimination, or that would expose employers to baseless litigation.
- All other employment discrimination laws limit damage awards. While it is critical to protect those who have been intentionally discriminated against, these individuals should be covered by the same protections and offered the same remedies under the law as individuals affected by all other types of workplace discrimination.
- Legislative proposals should not impede employer efforts to protect the safety and well being of their employees through workplace wellness programs and other services currently allowed under state and federal statutes. Furthermore, employer efforts to make timely and accurate determinations regarding requests or claims brought under current law such as state workers' compensation, the Family and Medical Leave Act, or the Americans with Disabilities Act should not be inhibited. Information that could be considered "genetic" may be needed to determine whether injury or illness was caused by work, FMLA leave is permitted, or an accommodation is needed.
- Duplicative efforts to guard against genetic discrimination are costly and confusing. Any legislative proposals regarding genetic discrimination should take into account the protections already offered by the HIPAA and its regulations, the ADA, and other federal, state, and local statutes and regulations

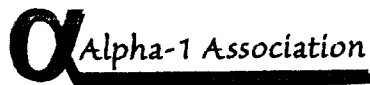
Conclusion

It is important to keep in mind when this body is providing recommendations to public policy makers that the collection and flow of employee information is an important issue for employers. In many respects, employment information is a double-edged sword. With proper information employers can make informed employment related decisions – decisions that they may be legally required to make; without such information decisions become more subjective, often miss the mark, and may subject the employer to claims of negligence. Thus as a general rule, employers should collect only information that they may legally use in making employment decisions, ensure such information is properly retained, and limit access to such information.

I would like to thank the committee again for the opportunity to appear before you today and will be pleased to respond to any of your questions regarding both my written and oral statements.

Thank you

Written Comments



Alpha-1 Association
275 West Street, Suite 210
Annapolis, MD 21401



Alpha-1 Foundation
2937 SW 27th Avenue, Suite 302
Miami, FL 33133

**Secretary's Advisory Committee on Genetics, Health, and Society
October 18, 2004**

These comments are submitted on behalf of the Alpha-1 Association and the Alpha-1 Foundation. The Alpha-1 Association is the patient advocacy and support organization representing the community of individuals affected by Alpha-1 Antitrypsin Deficiency (Alpha-1). The Alpha-1 Foundation is dedicated to providing leadership and resources that will result in increased research, improved health, worldwide detection and a cure for Alpha-1.

Alpha-1 is a devastating disorder, a pediatric liver disease that requires transplantation and an adult onset degenerative lung disease that strikes in the prime of life leading to repeated infections and progressive loss of lung function. The median age of survival is 54. The most common signs and symptoms of Alpha-1 are recurring respiratory infections, shortness of breath or awareness of one's breathing, non-responsive asthma or year-round allergies, rapid deterioration of lung function without a history of significant smoking, decreased exercise tolerance, chronic liver problems, and elevated liver enzymes.

Alphas need access to specialized healthcare without fear of retribution such as the loss of health insurance. In the absence of federal legislation, states have implemented a patchwork of laws that shield individuals from employment and insurance discrimination. We need national policy to ensure that all Americans have the same protections.

The Alpha-1 Foundation's Ethical Legal and Social Issues (ELSI) Working Group endorsed the recommendations of the American Thoracic Society/European Respiratory Society Standards Document on Diagnosis and Management of Alpha-1 Antitrypsin Deficiency. These recommendations are being implemented by the Foundation's National Targeted Testing Program and include testing symptomatic individuals or siblings of those who are diagnosed with Alpha-1. The absence of protective legislation has caused the ELSI to recommend against population screening and genetic testing in the neonatal population. Early diagnosis in Alpha-1 can significantly impact disease outcomes by allowing individuals to exercise preventative health measures, seek appropriate therapies, and engage in essential life planning. Unfortunately, gaining this information may lead to discrimination against individuals who have no control over their inherited condition.

The Alpha-1 Coded Testing (ACT) Trial, funded by the Alpha-1 Foundation and conducted at the Medical University of South Carolina offers a free and confidential finger-stick test that can be completed at home. The results are mailed directly to the participants. The ACT Trial has offered individuals the opportunity to receive confidential test results since September of 2001, to date over 2,400 test kits have been requested. The test is administered through a research study which evaluates perceived risks and benefits of genetic testing.

Of those returning the test kits and responding to the survey questionnaire:

Over 30% report fear of losing insurance as the reason for seeking confidential testing;

34% report concern about facing higher health care costs if results were public;

85% seek testing for the Genetic Knowledge. In fact, this was the most popular response to the perceived benefits of seeking testing.

We appreciate the opportunity to provide comments today and strongly encourage the Advisory Committee to make recommendations that will promote protective legislation for genetic conditions.



Alan I. Leshner
Chief Executive Officer and
Executive Publisher, *Science*

September 2, 2004

Ms. Amanda Sarata
Secretary's Advisory Committee on
Genetics, Health, and Society
6705 Rockledge Dr., Suite 750
Bethesda, MD 20892

Dear Ms. Sarata:

On behalf of the American Association for the Advancement of Science (AAAS), I want to express our support for your scheduled hearing on October 18, 2004, to examine the scope and nature of genetic discrimination. Founded in 1848, AAAS is the world's largest multidisciplinary scientific society and publisher of the journal, *Science*. The Association includes some 262 affiliated societies and academies of science, serving 10 million individuals. The non-profit AAAS is open to all and fulfills its mission to "advance science and serve society" through initiatives in science policy and education, international programs, and more.

The issue of how genetic information can be used to improve health without discriminating unfairly against people must be addressed if we are going to ensure the highest levels of public confidence in the application of genetic knowledge to medical care. We are pleased that SACGHS has identified genetic discrimination as an issue of "high priority."

In 1999, a AAAS working group issued a statement on genetic discrimination that included the following recommendations:

- Individuals should be able to gain information about their genetic makeup, but should be able to protect themselves against discrimination by controlling access to such information.

Ms. Amanda Sarata
September 2, 2004
Page 2

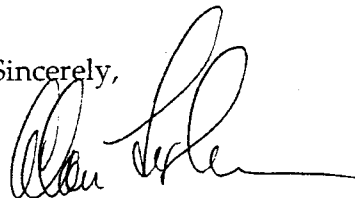
- Genetic information should be used only to enhance, not undermine, an individual's quality of life. Society, therefore, in pursuit of the common good, has a responsibility to protect citizens against the misuse of genetic information.
- Policies should be adopted to ensure opportunities for people to participate in research studies and clinical trials without fear that their genetic information could adversely affect their health insurance status.

The AAAS working group statement provides a sound set of principles for considering how to respond to the threats posed by genetic discrimination.

Those principles are as relevant today as they were when the statement was originally issued. We commend them to the attention of SACGHS, and request that the full AAAS working group statement be made part of the hearing record. The complete statement available on the AAAS website at <http://www.aaas.org/spp/dser/bioethics/resources/gdiscrim.shtml>.

Should you wish additional information about AAAS's activities related to genetic discrimination, feel free to contact Dr. Mark S. Frankel, director of our Program on Scientific Freedom, Responsibility and Law, at 202.326.6793, or you may call my office.

Sincerely,



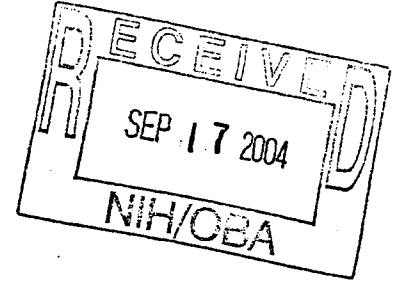
Alan I. Leshner



American Association of Clinical Endocrinologists

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September 15, 2004



Amanda Sarat
Secretary's Advisory Committee on Genetics, Health and Society
Office of Biotechnology Activities
Office of Science Policy
National Institutes of Health
6705 Rockledge Drive, Suite 750, MSC 7985
Bethesda, Maryland 20892-7985
FAX: 301-496-9839

RE: Genetic Discrimination

Dear Ms. Sarat,

The American Association of Clinical Endocrinologists (AACE) wishes to respond to the SACGHS request for information on potential genetic discrimination in anticipation of the October 18th SACGHS meeting. AACE represents more than 4600 practicing clinical endocrinologists in this country who care for patients with endocrine disorders including diabetes mellitus, osteoporosis, hormonal therapy, thyroid disorders, cholesterol disorders, hypertension and obesity. Genetic based testing for thyroid neoplasia and Multiple Endocrine Neoplasia (MEN 2) is in widespread use by AACE members.

AACE will survey their physician members through both the AACE web site and through the AACE First Messenger news letter for any cases of genetic discrimination known to AACE members. We will convey this information to SACGHS before the October meeting.

We look forward to working with SACGHS and sharing information in this critical area.

Sincerely,

Bruce Bower, M.D.
Chairman, AACE Genomics Committee



American Association of Clinical Endocrinologists

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October 6, 2004

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Re: Genetic Discrimination

Dear Ms. Sarat,

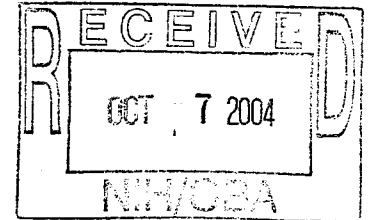
The American Association of Clinical Endocrinologists (AAACE) has posted the genetic discrimination inquiry on the AAACE website, the AAACE Online News and in the AAACE First Messenger publication. To date we have had no member reports of perceived genetic discrimination.

Please accept this preliminary information. A final report will be forwarded to SACGHS in approximately three weeks.

We look forward to working with SACGHS and sharing information in this critical area.

Bruce Bower, M.D.

Chairman, AAACE Genomics Committee





American Association of Clinical Endocrinologists

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November 2, 2004

Amanda Sarat
Secretary's Advisory Committee on Genetics, Health and Society
Office of Biotechnology Activities
Office of Science Policy
National Institutes of Health
6705 Rockledge Drive, Suite 750, MSC 7985
Bethesda, Maryland 20892-7985

RE: Genetic Discrimination

Dear Ms. Sarat,

The American Association of Clinical Endocrinologists (AAACE) has completed the genetic discrimination inquiry published on the AAACE website, in AAACE Online News and in *The AAACE First Messenger* publication earlier this month. To date, we have received no member reports of perceived genetic discrimination.

We appreciate that a survey of this nature is less than definitive although we also believe it is relevant to the question of genetic discrimination, perceived or real.

AAACE is an organization which represents over 5,000 physician endocrinologists. Endocrinologists have special expertise and training in the treatment of endocrine disorders including diabetes mellitus, osteoporosis, hormone therapy, thyroid disorders including thyroid cancer, cholesterol disorders, hypertension and obesity. AAACE is committed to the enhancement of the practice of endocrinology and the maintenance of the highest levels of care for patients with endocrine disorders.

We look forward to working with SACGHS in this ongoing important area of concern.

Sincerely,

Bruce Bower, M.D.
Chair, AAACE Genomics Committee

Comments from the American Board of Genetic Counseling to the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) on October 19, 2004.

The American Board of Genetic Counseling (ABGC) is the national accrediting and credentialing body for the profession of genetic counseling. The ABGC establishes minimum requirements for graduate programs in genetic counseling and develops the criteria by which individuals become eligible to sit for the certification examination. ABGC also recognizes the importance of demonstrating a life-long commitment to maintaining the knowledge and skills necessary to provide genetic services and oversees recertification of genetic counselors.

ABGC feels that resolution of the two issues discussed by SACGHS during this meeting – genetic discrimination and billing and reimbursement - are critical to the continuation and growth of the field of genetic counseling. One of the primary goals of the process of credentialing and accreditation provided by ABGC is to protect the public by ensuring access to professionals appropriately trained in genetics. Comments from presentations yesterday and from working genetic counselors imply that some individuals are fearful of genetic discrimination and afraid to seek the help of trained genetic professionals. Such individuals may feel that they must seek genetic information from other healthcare providers, non-medical caregivers, friends and family members. In requesting answers to important questions about their risk to develop medical conditions with an inherited component, they often receive incomplete or incorrect information. This could potentially result in an individual not obtaining information about optimal health care interventions and prevention programs. ABGC accredited training programs universally include the topic of discrimination in their curriculums and teach genetic counseling students how to discuss the advantages and disadvantages of obtaining genetic information. Legislation designed to reduce genetic discrimination and educational initiatives addressing the actual versus the perceived risk of genetic discrimination need to be developed so that individuals may freely discuss their concerns about genetic conditions with professionals who can knowledgeably provide accurate information. ABGC is willing to work with this committee and others to reach this goal.

ABGC also works to insure that the field of genetic counseling remains a viable and attractive career. The difficulties with billing and reimbursement for genetic

counseling services could impede the development of new genetic counseling programs and interfere with the ability of institutions housing clinical genetics programs to support the activities of genetic counselors. As discussed at the last SACGHS meeting, efforts must be made to increase the number and size of training programs. University leaders will assess the viability of the profession and the need for new programs and expansion of existing programs in genetic counseling before committing resources. Lack of reimbursement for genetic services could result in a decrease in these services, affecting not only our patients and their families who are dependent on these services but also decreasing the availability of clinical training sites for genetic counseling students. Lastly, potential students may be hesitant to enter the field of genetic counseling because of the uncertainty of reimbursement for services.

As this committee is well aware, the advances in medical genetics are forcing fundamental changes in the way health care providers practice medicine and think about health and disease. Knowledge about genetics and its social and ethical implications is becoming increasingly essential for many health care professionals. Genetics health care professionals, have been, and will continue to be, the ones who train and educate other health care professionals about the many complexities of genomic medicine - including the potential for discrimination. According to a professional status survey administered by the National Society of Genetic Counselors in 2002, a majority of genetic counselors are currently involved in the genetics education of physicians and medical students and other healthcare professionals. Many genetic counselors have developed and implemented innovative educational models that facilitate the genetics education of other health care professionals and students.

ABGC is committed to working with this committee to reduce the barriers of genetic discrimination and inadequate billing and reimbursement for genetic services, and encourage the public to seek information from trained genetic professionals. As certified genetics professionals, we recognize that the demand for genetic counseling services will continue to increase and we would like to insure that these services are provided by appropriately trained professionals. ABGC supports this committee in its efforts to recognize those qualified to provide genetic counseling services and hopes the committee will support the credentialing processes already in place for genetic

counselors, nurses in genetics and others. Clinical genetics services must be recognized by the health care industry and reimbursed appropriately – both so patients can receive quality genetics services and genetics professionals can be trained. There must be high standards for all genetics professionals and competency must continue to be assured through the accreditation of training programs and certification and re-certification of practicing genetic counselors.

Thank you.



September 17, 2004

Edward R.B. McCabe, M.D., Ph.D.
Chair
Secretary's Advisory Committee on Genetics, Health and Society
NIH Office of Biotechnology Activities
6705 Rockledge Drive, Suite 750
Bethesda, Maryland 20892

Dear Dr. McCabe:

As one of the nation's largest non-profit health organizations, the American Cancer Society is responding to the Secretary's Advisory Committee on Genetics, Health, and Society public comment solicitation concerning genetic discrimination.

Genetic research is one of the most exciting areas of scientific investigation today. The field of cancer genetics is rapidly growing and has already delivered great promise to patients who suffer from the disease. Advancements such as targeted drug therapies, classification of tumor aggressiveness, and cancer susceptibility based on biomarkers (e.g., BRCA1/2) have already become evident. Further, this technology is impacting all aspects of cancer management today, including prevention, screening, diagnosis, and treatment. However, fear of employment and insurance discrimination prevents individuals from taking advantage of these genetic technologies.

Currently, federal statutory protection against insurance and employment genetic discrimination is limited. Although the Health Insurance Portability and Accountability Act affords some group health insurance protection, insurers may still use genetic information to determine insurance rates and collect genetic information. Further, there is no federal anti-discrimination statute governing the individual market in which insurance companies may deny or cancel health coverage on the basis of genetic information and many other insurance products remain unregulated. Nor is there federal statutory employment protection that specifically prohibits genetic discrimination or that prohibits employers from requesting or requiring genetic testing. Although many states have enacted differing employment and health insurance anti-discrimination regulations, this patchwork protection is inadequate and may be negated by federal preemption of state law. The Health Insurance Portability and Accountability Act privacy regulations are protective of genetic information privacy but do not eliminate the continued reluctance of individuals to participate in genetic testing due to fears of discrimination.

The American Cancer Society is the nationwide, community-based voluntary organization dedicated to eliminating cancer as a major health problem by preventing cancer, saving lives, and diminishing suffering from cancer through research, education, advocacy, and service. The American Cancer Society supports the enactment of federal legislation to prevent employment and insurance discrimination based on genetic susceptibilities and cancer diagnoses. We look forward to participating in the Advisory Committee's public hearings on October 18th.

Sincerely,

A handwritten signature in black ink, appearing to read "Harmon S. Eyre". The signature is fluid and cursive, with a large initial "H" and "E".

Harmon Eyre, MD
Chief Medical Officer and Executive Vice President for Research and Cancer Control
American Cancer Society

October 19, 2004

Edward R.B. McCabe, M.D., Ph.D.

Chair

Secretary's Advisory Committee on Genetics, Health and Society

NIH Office of Biotechnology Activities

6705 Rockledge Drive, Suite 750

Bethesda, Maryland 20892

Dear Dr. McCabe and Esteemed Committee Members:

As one of the nation's largest non-profit health organizations, the American Cancer Society (ACS) is formally submitting additional comments regarding genetic discrimination to the Secretary's Advisory Committee on Genetics, Health, and Society. The American Cancer Society has a long legacy of advocating on behalf of cancer patients, their families, and our nation's health. Recent advances in the basic, applied, and clinical genetic sciences are beginning to offer Americans the ability to prevent and detect cancers in their earliest forms. These advances greatly increase the ability to save lives from cancer, diminish the suffering from cancer, and potentially eliminate cancer as a major public health problem. However, these advances in genetic evaluation and intervention pose many social, ethical, and legal issues that may lead to genetic discrimination and breaches of patient confidentiality, both of which are significant concerns to the American Cancer Society.

During the twentieth century, genetic science made monumental gains in the field of disease diagnoses and prevention. Historically, genetic disease, including inherited cancer syndromes, was largely identified by the collection of family history information, a tremendously effective screen for hereditary cancer; however, even today family history is largely under-utilized by practicing physicians. To that end, solely examining family history or one gene marker is an oversimplification of cancer etiology and progression. Cancer is a multi-factorial disease. A genetic test cannot account for age of disease onset or the synergistic effect of environmental factors. Behavior modification, such as regular exercise and a healthy diet, may prevent or delay disease onset and advances in medical and pharmaceutical interventions may also prevent or delay disease.

Molecular genetics, and its ability to analyze multiple factors simultaneously, holds great promise in the field of cancer prevention and control. The completion of the Human Genome Project helped identify thousands of genetic markers. Each year scientists are detecting new biomarkers for more diseases and further translating bench science discoveries to clinical bedside practice. These discoveries have enabled medical practice to enhance the quality of care before disease manifestation by monitoring disease progression, providing counseling options, improving lifestyle and dietary changes, and offering prophylactic surgery.

Today, scientists can analyze thousands of genes and determine patterns of gene activity simultaneously using a DNA chip, a technology quickly becoming integrated into clinical

practice settings. DNA chips are being used to diagnose cancers. These chips are already supplementing and may someday replace the practice of identifying tumors by visual appearance and thereby providing more accurate and earlier diagnoses, determining tumor aggressiveness, and predicting patient outcomes by varying treatment regimes.

Although DNA chips will revolutionize cancer genetics, their emergence into mainstream clinical practice has not yet taken place. Traditionally, there are four types of genetic tests available to patients and physicians: carrier, predictive, newborn, and disorder. Cancer genetic tests can largely be categorized as a predictive genetic test, which is a test that is used to identify those with a specific family history of disease and are at risk of developing disease (e.g. BRCA1/2). These genetic tests pose several potential problems to patients and other purveyors of this information.

First, a positive test does not mean the disease will occur, while a negative test does not mean an individual is without risk. Further, privacy protections are not adequately in place for many Americans and the potential for improper use of genetic results concerns many Americans, including President George W. Bush. In the summer of 2001, President Bush addressed the potential for genetic discrimination and the medical uncertainty a genetic test may yield during his Presidential Radio Address:

"Genetic discrimination is unfair to workers and their families. It is unjustified – among other reasons, because it involves little more than medical speculation. A genetic predisposition toward cancer or heart disease does not mean the condition will develop. To deny employment or insurance to a healthy person based only on a predisposition violates our country's belief in equal treatment and individual merit." - President George W. Bush

Despite the projected and current advances based on genetic information and test results, individuals are reluctant to be genetically evaluated. In 1997, Donna Shalala reported that over 20% of people with a genetic disorder stated that they, or a family member, had been refused medical insurance on the basis of their genetic profile. Numerous surveys have concluded similar results:

- ❖ A report published by the Johns Hopkins University Genetics and Public Policy Center in 2004 stated that 92% of individuals were opposed to employers having access to their genetic information, while 80% of individuals were opposed to health insurers having access to their genetic information. Further, 97% of college-educated respondents opposed either employers or health insurers having access to their genetic information.
- ❖ In a 2003 survey of participants in the Hereditary Colorectal Cancer Registry at the Johns Hopkins Hospital, nearly half reported a high level of concern regarding the potential for genetic discrimination. These participants also reported that they were less likely to undergo or consult with a health care professional about genetic testing and significantly more likely to pay out-of-pocket, use an alias, or ask for results to be excluded from their medical record.
- ❖ In a 2000 survey of genetic counselors, 68% of respondents reported that they would not bill their insurance company if they had a 50 percent or greater risk of carrying a BRCA mutation for fear of genetic discrimination.

Individual cases of genetic discrimination, due to a cancer diagnosis, have not yet been recorded; however, the Coalition for Genetic Fairness published a report in July of 2004 reporting several anonymous cases of genetic discrimination.

“Mary had a family history of breast cancer – both her mother and an aunt had been diagnosed with the disease. Concerned about her own future, Mary considered being tested for BRCA-1, hoping to take prophylactic measures to reduce her risk if the result was positive. Ultimately, she decided not to take the test because she feared a positive result would jeopardize her chances for promotion at her law firm.” – Anonymous

“A 28-year-old woman who tested positive for BRCA-1, one of the genes that indicates predisposition to breast cancer, was denied health insurance coverage because of her genetic status. Although she was not asked for genetic information when she applied for insurance, when the woman reported on her application that she had undergone prophylactic mastectomies and a hysterectomy, the insurance company requested her medical records, which included her genetic information. Her application for coverage was rejected and she was later able to determine that the denial was due to her positive BRCA-1 test result. Only after involving a lawyer, and after much time and effort, was she ultimately able to secure insurance coverage.” - Anonymous Email Submission

“It was extremely important to me to know that I could be tested and not dropped from my insurance or job if I were found out to have a BRCA1 or BRCA2 mutation.... The fear of possibly having a disease and either losing insurance or a job when I would need it most would be frightening beyond words. How sad if people like myself, who are most at risk, would not test and therefore possibly die an early and preventable death.” - Anonymous Memorandum Submission

“A patient advocate working at an oncology clinic had a telling encounter with a young woman whose mother and sister died of breast cancer. The young woman visited the clinic, but refused to sign in. The advocate explained that registration was required, and that the woman's genetic information would be kept entirely confidential. The woman became extremely emotional, saying that she believed she would expose herself and her children to the risk of discrimination if her visit were in any way documented. The advocate tried to encourage the woman to stay, but she left the office without testing or counseling, and without scheduling a screening.” - Anonymous Email Submission:

The fear of potential genetic discrimination demonstrated in these surveys and anonymous submissions are justified since current statutes are not adequate to prevent genetic discrimination. Specifically, there is no federal statute protecting against genetic discrimination for individual health insurance and gaps remain in group health insurance protection. Additionally, there is no federal employment statute specifically protecting against genetic discrimination and employment protections pursuant to the Americans with Disabilities Act (“ADA”) and other disability statutes are limited and uncertain. Further, various existing federal statutes require disclosure of genetically sensitive information that can inform employers and others of genetic susceptibilities and disease and individual state regulations vary making enforcement difficult.

Additional genetic discrimination health insurance legislation is needed because there is no federal anti-discrimination statute that covers individual health plans and the Health Insurance Portability and Accountability Act (“HIPAA”) does not completely eliminate genetic discrimination in group insurance coverage. HIPAA permits providers to request

or require genetic testing. Although genetic information is no longer treated as a preexisting condition for group plans and the use of genetic information to establish rules of eligibility or continued eligibility and for determination of premium contribution is prohibited, underwriters can use this genetic information to set premiums for the entire group. In addition, policy benefit caps and exclusions for specific conditions are also permitted. Further, although HIPAA preempts less stringent state law, a patchwork of varying and potentially more stringent state law exists that makes interpretation in specific discrimination cases more difficult.

Although the Standards for Privacy of Individually Identifiable Health Information promulgated pursuant to HIPAA protect medical record privacy, individuals fear that in certain circumstances this information can still be revealed. These regulations prohibit all forms of personal health information disclosure without voluntary, informed consent by both public and private health plans for HIPAA covered entities i.e. health insurers, providers and health care clearinghouses. Employers sponsoring health plans are prohibited from accessing personal health information for employment purposes without employee non-coerced consent. However, private health information may still be released in many circumstances including information necessary to protect public health, conduct medical research, improve the quality of care *i.e.* oversight and quality assurance, and to fight health care fraud and abuse. Genetic information can also be revealed in judicial and administrative proceedings, limited law enforcement activities, national defense and security, identification of deceased person or cause of death, facility patient directories, and emergency situations.

There is also no federal statute specifically protecting employees from genetic discrimination. Federal employees, less than five percent of the workforce, are protected from employment discrimination by Executive Order 13145 and disclosure exemptions of the Freedom of Information Act ("FOIA"). However, this non-statutory Executive Order protection is limited and can be withdrawn. This Order prohibits limiting, segregating or classifying employees to deprive them of opportunities and prevents discrimination in hiring and discharge. It defines protected genetic information; however, current health status information would not be protected unless it was derived from covered protected genetic information. Disclosure of genetic information is prohibited and these materials are considered confidential medical rather than personnel records. The federal government is prohibited from requesting, requiring, and collecting or purchasing protected genetic information. However, the employing entity may request or require information if the current condition would prevent the applicant or employee from performing essential functions of the job or where the information will be used exclusively to determine whether further medical evaluation is necessary for diagnosis. Genetic monitoring of biological effects of toxic substances is also permitted in specific circumstances. Although the Freedom of Information Act ("FOIA") permits public disclosure of government records, personnel and medical files maintained by agencies within the executive branch are excluded where the disclosure would be a "clearly unwarranted invasion of personal privacy." It also protects against disclosure impacting decedent's survivors. The protections afforded federal employees should be extended to

the remaining 95 percent of the workforce and a comprehensive genetic discrimination statute should protect federal and private employees.

Further, the applicability of federal disability statutes to genetic discrimination issues is limited and uncertain. The Americans with Disabilities Act (“ADA”) protects against employment discrimination in private sector employment, public services, public accommodations, and telecommunications; however, it specifically is “not intended to disrupt the current regulatory structure for self-insured employers...or current industry practices in sales, underwriting, pricing, administrative and other services, claims and similar insurance related activities based on classification of risks as regulated by the states.” Further, employers of less than fifteen employees are not even regulated pursuant to this statute. ADA statutory definitions and language also do not specifically include genetic discrimination. The ADA would, thus, require amendment to ensure applicability to genetic discrimination and all employees.

Moreover, court decisions have substantially limited the scope of the ADA. For covered individuals, the statutory definition of disability is limited to: (1) physical or mental impairment that substantially limits one or more of the major life activities of an individual, (2) a record of such impairment, or (3) of being regarded as having such an impairment typically of an illness, disease or other disorder. However, recent Supreme Court decisions interpreting the statutory language of the Act limit ADA protection to individuals who are presently – not potentially or hypothetically—substantially limited, and those who are mistakenly regarded as having a physical or actual impairment substantially limiting one or more *major* life functions. Proof of substantial limitation of a major life activity as a result of the impairment and the inability to work in a broad class of jobs are required. The Supreme Court’s holding in *Toyota Motor Manufacturing v. Williams* further limited protection by reducing the “central inquiry” to “whether the claimant is unable to perform the variety of tasks central to most people’s daily lives” such as “household chores, bathing, and brushing one’s teeth” in lieu of “whether the claimant is unable to perform the tasks associated with her specific job” and by also requiring that the “impairment’s impact must be permanent or long-term.” Reliance on being regarded as having a disability is also problematic. For example, in *Law v. Pect*, the Court held that an asymptomatic Huntington disease plaintiff did not provide sufficient evidence that her employer regarded her as disabled and discriminated against her on that basis.

Further, this disability statute does not prevent the acquisition of genetic materials. Employers may request or require genetic information from their employees if the employer can demonstrate that the information would be job related and consistent with business necessity. Employers may set medical standards for specific jobs since the ADA is superseded by existing medical standards for workplace safety. Employment in inherently risky occupations *i.e.* airline pilots, police officers, or firefighters may be denied based on medical evidence of the “likelihood” of harm or injury to others in the workplace although assessment of the genetic predisposition and the probability of developing symptoms affecting others must be made on a case-by-case basis and reasonable accommodations provided.

Employees are also unlikely to file an ADA claim. Individuals must reveal that they already have or are at risk of developing genetic diseases or abnormalities in order to file discrimination claims. Employees may feel that they have "too much to lose" by revealing information that has the potential for additional discrimination. The claim process is also lengthy and the decision uncertain.

Although the Rehabilitation Act of 1973 prohibits discrimination against handicapped individuals and those with "hidden disabilities," its application is limited to institutions and entities that receive federal funding or contracts. Furthermore, the definition of disability is similar to the ADA; that is, any person that has a physical or mental impairment that substantially limits one or more of that person's major life activities, a record of such impairment or someone that is regarded as having an impairment. The disability act would similarly require amendment to assure applicability to genetic discrimination issues.

Civil rights statutory protection is also not a refuge from discrimination for most individuals since the statute is limited to specific claimant classes. Title VII of the Civil Rights Act prohibits discrimination based on race, gender, color, national origin, or religion by employers of more than fifteen employees, labor organizations, employment agencies, and state and municipal governments. Genetic defects may be associated with specific races, national origin, religion or gender. Sickle cell anemia, Tay-Sachs, and breast cancer are just some of the genetic defects with significant prevalence in these protected classes although other genetic defects do not disproportionately affect these classes and so would not be protected pursuant to the Civil Rights Act. Testing may be permitted even for protected classes where the test is a legitimate job requirement or business necessity and its purpose is more compelling than any discriminatory effect. Discrimination may also be permitted for these classes where the immutable characteristic prevents the employee from effectively performing his job function.

Existing statutes require review to ensure that genetic information is protected and discrimination prevented including the Employee Retirement Income Security Act, Public Health Service Act, Social Security Act, Family, Educational Rights and Privacy Act, and Food, Drug and Cosmetic Act. The Family and Medical Leave Act, National Labor Relations Act, Environmental Protection Act, Federal Aviation and Federal Highway Administration rules and others may also require the use or disclosure of protected medical information in specific situations and require amendment. In addition, the Occupational Safety and Health Administration Act would require review to encompass genetic discrimination and privacy issues and to provide adequate health related standards for worker safety that incorporate genetic susceptibility information.

Federal legislation is also needed because state genetic discrimination laws do not provide equivalent protection against discrimination. Some states do not have any genetic discrimination laws while other states have varying definitions of genetic testing and information that may not provide adequate protection. Most define genetic testing or screening as limited to laboratory analysis rather than including the traditional evaluation

of physical or medical condition or history. In addition, some states permit insurers to use genetic information that can be actuarially justified thereby potentially negating their non-discrimination laws. Further, federal legislation may preempt even these limited state anti-discrimination laws. State disability law is not universal and is typically construed similarly to federal civil rights statutes and is, thus, similarly limited. In addition, individuals cannot be definitely assured that they will be afforded anti-discrimination protection even in states with anti-discrimination statutes. The contract law governing insurance is applied pursuant to varying choice of law statutes that impact the applicability of specific state genetic discrimination laws in individual cases. The applicability of specific state law is determined judicially during costly litigation.

The American Cancer Society supports the recommendations of the National Institutes of Health and Department of Energy Working Group on Ethical, Legal and Social Implications of the Human Genome Research and the National Action Plan on Breast Cancer. Recommendations regarding genetic information, health insurance, and employment insurance include the following:

Insurance Recommendations

- Insurance providers and employers should be prohibited from using genetic information, or an individual's request for genetic services, to deny or limit any coverage or establish eligibility, continuation, enrollment or contribution requirements.
- Insurance providers and employers should be prohibited from establishing differential rates or premium payments based on genetic information, or an individual's request for genetic services.
- Insurance providers and employers should be prohibited from requesting or requiring collection or disclosure of genetic information.
- Insurance providers and other holders of genetic information, apart from research settings governed by the Federal Policy for the Protection of Human Research Subjects (a.k.a., the "Common Rule"), should be prohibited from releasing genetic information without prior written authorization of the individual. Written authorization should be required for each disclosure and include to whom the disclosure would be made.
- Insurers should be required to cover all patient care costs for individuals who are enrolled in approved clinical trials on genetic research.

Employment Recommendations

- Employment organizations should be prohibited from using genetic information to affect the hiring of an individual or to affect the terms, conditions, privileges, benefits or termination of employment unless the employment organization can prove this information is job related and consistent with business necessity.
- Employment organizations should be prohibited from requesting or requiring collection or disclosure of genetic information prior to a conditional offer of employment, and under all other circumstances, employment organizations should be prohibited from requesting or requiring collection or disclosure of genetic information unless the employment organization can prove this information is job related and consistent with business necessity, or otherwise mandated by law.

Written informed consent should be required for each request, collection or disclosure.

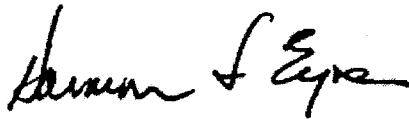
- Employment organizations should be restricted from access to genetic information contained in medical records released by individuals as a condition of employment, in claims filed for reimbursement of health care costs, and other sources.

Further, the American Cancer Society strongly supports additional genetics research and requests Congress to provide financial support and authorization to continue studies to examine the ethical, legal, and social issues related to protection of individuals, including those participating in genetic research and those seeking medical care involving genetic testing and counseling. Finally, it is our position that violators of these provisions should be subject to strong enforcement mechanisms, including a private right of action.

Advances in genetic technologies have allowed researchers to identify a growing number of genetic alterations that may indicate predisposition for developing cancer or other diseases. The ultimate goal of genetic testing research is the development of clinical applications for risk assessment, early detection, and appropriate interventions for individual risk reduction and disease prevention. The potential of such research raises questions about who will have access to genetic information and how this information might be used to discriminate or compromise individual privacy

The American Cancer Society is the nationwide, community-based voluntary organization dedicated to eliminating cancer as a major health problem by preventing cancer, saving lives, and diminishing suffering from cancer through research, education, advocacy, and service. In summary, we support the enactment of strong federal legislation to prevent employment and insurance discrimination based on genetic susceptibilities and cancer diagnoses.

Sincerely,

A handwritten signature in black ink, appearing to read "Harmon S. Eyre". The signature is fluid and cursive, with the first name being the most prominent.

Harmon Eyre, MD
Chief Medical Officer and Executive Vice President for Research and Cancer Control
American Cancer Society

American Psychiatric Association
Department of Government Relations
1000 Wilson Blvd, Suite 1825
Arlington, VA 22209
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Statement of

The American Psychiatric Association

to

**Secretary's Advisory Committee on Genetics, Health,
and Society (SACGHS)**

on

Genetic Non-Discrimination

October 18, 2004

The American Psychiatric Association (APA), the national medical specialty society, founded in 1844, whose over 35,000 psychiatric physician members specialize in the diagnosis and treatment of mental and emotional illnesses and substance use disorders, appreciates the opportunity to provide a statement on genetic non-discrimination. We thank the Committee for allowing us to provide this statement.

Genetic testing offers tremendous promise in identifying current and potential future health risks. At the same time, we have significant concerns that Americans' genetic information could be misused. Our concerns are shared by a strong majority of Americans: a U.S. Department of Labor survey showed that 63 percent of respondents would refuse to take a genetic test if insurers or employers could access their private results.

We believe the strongest possible enforceable genetic non-discrimination law should be passed. Employers and insurers should not be permitted to discriminate on the basis of a person's genetic profile and family history.

Our concerns extend beyond patients' reluctance to take a genetic test. Such reluctance means that people are disinclined to participate in clinical studies that require genetic testing, hurting our efforts to identify causes and new treatments for diseases, including mental illnesses. Worse, some patients' reluctance could keep them from getting a proper diagnosis today, as well as potentially life-saving treatment. Perhaps the most pernicious potential consequence of not enacting a ban on genetic discrimination is that Americans could lose their jobs or their health insurance, based simply on their family history.

Protecting patients' genetic information is essential to providing the highest quality medical care. We believe a patient's genetic information should only be used or disclosed by a health care plan, provider, or clearinghouse with the informed, voluntary, and non-coerced consent of the patient. As our knowledge of genetics grows, especially through the Human Genome project, the possible misuses of genetic information will expand unless enforceable safeguards are enacted.

The U.S. Senate voted 95-0 to pass the "Genetic Information Non-discrimination Act of 2003" (S. 1053), with the support of President Bush. Similar but stronger legislation (H.R. 1910) is now cosponsored by 242 members of the House. Both bills would ban employers and insurers from discriminating on the basis of a person's genetic profile and family history. APA urges Congress to pass and the President to sign the strongest possible enforceable genetic non-discrimination legislation into law.

Thank you for this opportunity to deliver this statement. Please call on the APA as a resource, as we would be happy to assist the Committee on the vital issue of genetic non-discrimination in any way.



PUBLIC POLICY COLLABORATION



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Secretary's Advisory Committee on Genetics, Health, and Society
6705 Rockledge Drive
Suite 750
Bethesda, MD 20892

September 17, 2004

RE: Request for Public Comment on Genetic Discrimination

The following comments are submitted by United Cerebral Palsy and The Arc of the United States in response to the Secretary's Advisory Committee on Genetics, Health, and Society's (SACGHS) request for public comments on genetic discrimination.

For more than 50 years, United Cerebral Palsy has been committed to people with disabilities and to effecting public policy that will enhance the ability of people with disabilities to live lives without limits in the communities, neighborhoods, schools and jobs of their choice. United Cerebral Palsy's national organization and our nationwide network of 105 affiliates in 37 states strive to ensure the inclusion of people with disabilities in every facet of society — from the Web to the workplace, from the classroom to the community. United Cerebral Palsy's Research and Education Foundation is an internationally recognized leader in research on cerebral palsy and related disabilities.

Founded in 1950, The Arc of the United States is the national organization of and for people with mental retardation and related developmental disabilities and their families. The national organization and our nearly 1,000 chapters nationwide are devoted to promoting and improving supports and services for people with mental retardation and other disabilities and their families. The association also fosters research and education regarding the prevention of mental retardation in infants and young children.

The disability community is, in general, highly supportive of genetic testing. Disability organizations, including The Arc of the United States, have participated in The Human Genome Project, a collaboration of scientists worldwide. Disability professionals understand that errors in our genes are responsible for an estimated 3000 to 4000 clearly hereditary diseases, disabilities and conditions. They play a part in cancer, heart disease, diabetes and many other common conditions, such as mental retardation.

On the other hand, genetic testing is clearly harmful if the information is used to deny jobs or insurance, or if it leads to other forms of discrimination. We already know that the threat of genetic discrimination leads people to decline genetic screenings and other health services to avoid revealing information that may be used against them. For example, the Journal of the American Medical Association reported in 2000 that only 57% of women at risk for breast cancer seek genetic testing, and 84% of those who decline the test do so because they fear genetic discrimination. Genetic scientists have warned repeatedly that progress in the field of genetic medicine depends on the willingness of individuals to submit to genetic tests without fear of discrimination.

Discrimination based on the presence of a disability has always been an issue of great concern to both The Arc and United Cerebral Palsy. People with mental retardation, cerebral palsy and other disabilities have long been discriminated against in both insurance and employment. Discrimination is an enormous challenge to people who are treated differently, given lesser opportunities, simply based on disability, and it is something that is faced every day in every community by thousands and thousands of people who are differentiated by disability.

Now families and individuals must fear a different type of discrimination, one that is emerging due to the increasing use of genetic testing for the purpose of exposing the presence of any abnormal or defective genes. This new phenomenon, genetic discrimination, extends the bounds of potential discriminatory activity against people to a new frontier that is based on genetic characteristics alone. It opens the door to excluding people from essential life activities based only on a gene, or genes, that predispose them or their offspring to disabilities, diseases or late-onset disorders. Genetic discrimination is even more insidious, in some ways, than other forms of discrimination, because it occurs when someone is treated differently based on having a gene that may or may not cause the person to manifest a disability sometime in the future. Also, people who are carriers for a genetic condition, who show no signs of the condition themselves, may be discriminated against because of their potential to have a child with the condition, again an event that may or may not ever materialize.

Family members of people with disabilities, for example, or carriers of a genetic condition associated with a disability, may be discriminated against based on their genetic make-up. Such individuals may include people who carry the gene for Fragile X Syndrome, the most common inherited cause of mental retardation. Twenty percent of people with this gene will never display any form of mental retardation. Yet, because they carry the Fragile X gene, they may be treated as though they had mental retardation, even though they do not.

While many states have enacted protections against genetic discrimination in health insurance or employment, or both, these laws are inconsistent and limited in what and who they cover. In the employment arena, they fail to ensure a

uniform floor of protections, and as they apply to health insurance, most state laws fail to ensure coverage for a sizable number of those with private health insurance coverage. Because of the Employee Retirement Income Security Act of 1974 (ERISA), many of these laws may not apply to as many as 131 million American workers and families covered under private, employer-based health plans.

At the federal level, with the passage of the Americans with Disabilities Act (ADA) in 1990, discrimination in employment, public accommodations and services based on disability is against the law. It is important to remember, however, that the ADA does not apply to the insurance industry.

Since the ADA applies not only to people with a disability, but to people who may be "regarded" as having a disability or who have a "history" of disability, the ADA *should* protect individuals against genetic discrimination by employers who may perceive genetic predisposition to disease as a form of disability. People who experience genetic discrimination may be "regarded" as having a disability (because they have an abnormal gene), even though they may not have a disability. These protections, however, are untested and uncertain. Although the Equal Employment Opportunity Commission (EEOC) has brought one suit to enforce these rights, a case against Burlington Northern Santa Fe Railways was settled without a trial, and it is unclear how a court would rule on the EEOC's interpretation of the law. Although the EEOC determined that "entities that discriminate on the basis of genetic predisposition are regarding the individuals as having impairments, and such individuals are covered by the ADA," they also said that "unaffected carriers of recessive and X-linked disorders, individuals with late-onset genetic disorders who may be identified through genetic testing or family history as being at high risk of developing the disease, are not covered by the ADA."

Also, proving employer bias under the ADA has been a difficult standard of proof for employees to meet, and recent decisions by the Supreme Court have limited the reach of the ADA and narrowed its protections. Thus, the ADA is not likely to provide adequate support for employees seeking to enforce *new* rights. In any event, the ADA does not protect workers from requirements to provide genetic information to their employers.

The only other federal protection against genetic discrimination is found in the Health Insurance Portability and Accountability Act of 1996 (HIPPA) which places limitations on the exclusion period for pre-existing conditions when people change jobs and prohibits discrimination against individuals based on health status, including their genetic information. While HIPPA extends coverage to people who have genes that predispose them to a disability or disease, or who have genes for a late onset disorder, it may not protect carriers of genetic disorders who do not yet manifest symptoms.

For Americans at risk for genetic discrimination, these gaps in the law pose a serious barrier to their security.

The Arc of the United States and United Cerebral Palsy appreciate the opportunity to comment on this vital issue. In summary, we believe that genetic testing holds enormous promise to prevent disability and health problems and can help people cope more effectively with conditions that are unavoidable. Present and future generations will benefit from the insight into disabling conditions genetic testing and genetic research can provide. Nonetheless, strong protections against genetic discrimination are critical to every man, woman and child in this country. We stand in firm support of federal legislation and/or policy that will clearly negate any American's having to fear that genetic information will be misused to prevent them from getting the jobs or insurance coverage they need.

Ms. Sarah Carr
National Institutes of Health
Executive Secretary
Secretary's Advisory Committee on
Genetics, Health, and Society (SACGHS)

September 10, 2004

Dear Ms. Carr:

Thank you for inviting the Council for Responsible Genetics (CRG) to submit comments to the SACGHS on genetic discrimination. In these comments, we highlight a few of the cases of discrimination identified in our research.

We would welcome the opportunity to be a part of the deliberations of the SACGHS on this important matter. Please feel free to contact me at (617) 868-0870.

Best regards,

Sujatha Byravan,
Executive Director, CRG

**Comments to the Secretary's Advisory Committee on Genetics, Health and Society
from the Council for Responsible Genetics (CRG)**

September 10, 2004

I appreciate this invitation to submit comments to the Secretary's Advisory Committee on Genetics, Health, and Society. The Council for Responsible Genetics is a national non-profit organization based in Cambridge, Massachusetts. Founded in 1983, the Council fosters informed public debate on the social and ethical consequences of developments in the life sciences. I wish to provide the Committee with an overview of empirical evidence of genetic discrimination gathered by friends and affiliates of our organization over the last two decades.

From the late 1980s to the mid-1990s, Paul Billings, the current Chair of the Council for Responsible Genetics and Vice President for Genetics and Genomics at the Laboratory Corporation of America, carried out an investigation with nine other colleagues (under the title of the Genetics Screening Study Group) to identify cases of genetic discrimination. A total of 27,790 questionnaires were sent to the membership of several genetic disease organizations. After screening initial responses, Dr. Billings and his colleagues interviewed 206 claims of discrimination involving individuals who were presymptomatic for a genetic disorder, individuals who were asymptomatic because of ongoing therapeutic interventions, and individuals who were carriers for an autosomal recessive genetic condition.

The Genetic Screening Study Group applied a rigorous methodology for carrying out its work. Over half of the initial 455 respondents claiming genetic discrimination were excluded for failure to meet strict criteria. Cases of differential treatment based on clinical disability or illness were automatically disqualified. Cases where inadequate information had been submitted to verify a claim of discrimination were also removed from consideration. Many responses included supporting documentation from employers and insurance companies. The results of the interviews were subsequently published in the *American Journal of Human Genetics* (1992),¹ *Journal of Public Health Policy* (1994),² and *Science and Engineering Ethics* (1996).³

¹ Paul R. Billings, et al., "Discrimination as a Consequence of Genetic Testing," *American Journal of Human Genetics* 50(1992): 476-482

² Joseph S. Alper, et al., "Genetic Discrimination and Screening for Hemochromatosis," *Journal of Public Health Policy* 15(1994): 345-358

³ Lisa N. Geller, et al., "Individual, Family, and Societal Dimensions of Genetic Discrimination: A Case Study Analysis," *Science and Engineering Ethics* 2(1996):71-88.

These case studies tell a compelling story. They provide some of the best direct evidence of discrimination in employment and health insurance against asymptomatic, presymptomatic, and carrier status individuals on the basis of real or perceived genetic risk factors.

Kim, for example, was a social worker with a large human services agency. During a staff workshop on caring for people with chronic illnesses, Kim mentioned that she was the primary caretaker for her mother who died of Huntington's disease. Kim herself had a 50% chance of developing this fatal genetic condition. One week after she revealed her risk status, Kim was fired from her job---even though she had received outstanding performance reviews in the months prior to the firing.

In another case, a physician reported that "an individual was found to have Gaucher Disease. His brother was screened and the results were consistent with unaffected carrier status [heterozygote]. The brother applied for a governmental job and included the history of his testing in the application. He was denied the job because of his being a 'carrier, like sickle cell'."

A significant number of cases involved asymptomatic individuals with hereditary hemochromatosis, an iron storage disorder that can be effectively controlled by an inexpensive regimen of phlebotomies. Two of these cases are particularly relevant:

"An asymptomatic 53 year old man had been diagnosed with hemochromatosis. His treatment consisted of phlebotomies at three month intervals. He applied for a position at an insurance agent and was accepted into the company's agent training program. In the course of an interview, he mentioned that he was being treated for hemochromatosis and agreed to submit a copy of his medical report based on a recent physical examination. A manager of the company told him that his illness might result in the company's inability to offer him medical benefits. However, the applicant was permitted to enter the training program with the expectation of a position when he completed it. After approximately five or six weeks of training, he was told that the home office of the company would not hire him because of his diagnosis of hemochromatosis. He was not paid for the week which he spent training."

"An asymptomatic 25 year old man was diagnosed with hemochromatosis by means of blood tests after a family member developed the condition. He suffered no symptoms of organ damage as a result of the disorder. After his diagnosis he initially underwent weekly phlebotomies and was subsequently phlebotomized bi-monthly. After he became ineligible to continue receiving health insurance through his parent's health insurance policy, he applied for an individual policy since his place of employment did not offer health insurance. His application was rejected because of his hemochromatosis."

These case studies also revealed steps taken to avoid discrimination. The authors note that "because of fear of discrimination, several respondents reported that they withheld or

'forgot' to mention potentially important medical or family history information to physicians, employers or insurers. Others reported that their insurance agents suggested that they give incomplete or dishonest information on insurance application forms." These findings are supported by a survey published in *Science* in 1996 revealing that many respondents had refused to undergo genetic testing, refused to disclose genetic test information to insurers and employers, and had paid out-of-pocket for genetic services to protect the privacy of their medical records.⁴

Descriptions of reported instances of discrimination have been excerpted from the three case studies cited above.

⁴ E. Virginia Lapham, Chahira Kozma, Joan O. Weiss, "Genetic Discrimination: Perspectives of Consumers," *Science* 274, 5287(1996):621.

Oral Testimony
Sharon F. Terry
Genetic Alliance

October 18, 2004

I am Sharon Terry, President and CEO of the Genetic Alliance – an alliance of 600 genetic disease advocacy organizations that represent over 14 million individuals. The Genetic Alliance is a founding member of the Coalition for Genetic Fairness – a coalition of organizations working together against genetic discrimination. We understand the promise of basic and medical research and are appalled that many families and individuals experience genetic discrimination, and fear both knowing their own risk and participating in research as well. You have heard some of their stories here today. The Coalition's "Faces of Discrimination" provides you with more stories.

We believe that all genetic information, including family history, deserves strong protections against misuse in health insurance and employment. Such safeguards will protect the rights and confidentiality of the individual and their family. While privacy is no longer possible, misuse of information can be prevented.

This is an exciting and hopeful time for medicine. It is imperative, however, that we, the public, take full advantage of new medical advances that could help prevent disease before it develops. Genetic nondiscrimination legislation will reduce the likelihood of genetic information being misused in health insurance or employment decision-making. As you well know, simply having a positive genetic test does not mean one will develop a disease -- thus this information should not be used to make decisions about insurance coverage or employment.

You have heard here, from both consumers and professionals, that as biomedical research advances, genetic testing will become a critical tool in the provision of healthcare. As a result, many more people will know about their own genetic makeup, putting them at risk of genetic discrimination. People who would like to avail themselves of genetic testing already have enough to worry about. They should not have the additional burden of genetic discrimination.

In addition, it is important that we who carry mutations for diseases are encouraged to participate in genetic research. A fear of discrimination discourages that participation – adding another hurdle to the pathway from basic science and health care services.

The Genetic Alliance and the Coalition for Genetic Fairness have worked for years on this issue. This past year we presented a letter to Speaker Hastert signed by hundreds organizations and hundreds of individuals. We held a press conference with Heidi Williams and Dr. Collins on Capital Hill. We continue to work together on this issue, and plan together to go forward until legislation is passed – in a spirit of cooperation and compromise.

Therefore, on behalf of millions of consumers and advocacy organizations, I convey to our strong support of genetic information nondiscrimination legislation. The Senate passed S.1053 95-0, and President Bush has said he will sign it.

We have come here today to ask that you be bold and clear in your communication with the Secretary. Please ask the Secretary to ask Speaker Hastert, Representatives Delay and Barton to move this legislation. In addition, you can help to ferret out the opposition to this legislation. We have not been able to get a clear picture of the reason this bill is not moving in the House. Your position, as a chartered committee of the Secretary, should afford you the leverage needed to understand the problem. We offer our support and the support of our member organizations to move this legislation.

Public policy must keep pace with scientific advances, and provide those advances with a climate conducive to their translation into health benefits for all. Thank you for this opportunity to bring these voices to the table. Thank you also for your leadership – we need you in this fight.

Testimony of
Sharon F. Terry
President and CEO, Genetic Alliance

Secretary's Advisory Committee on Genetics, Health and Society

October 18, 2004

The Genetic Alliance strongly supports legislation that prohibits genetic discrimination.

Policy Development Outstripped by Escalating Genetic Discoveries

Thanks to the Human Genome Project, these are remarkable and historic times. Scientists all over the world are using this genetic map to unravel the mysteries of heart disease, cancer, diabetes, mental illness, asthma, multiple sclerosis — since all diseases have a hereditary component. Already there are individuals and families whose lives have been touched in profound ways by biomedical tests and technologies never before imagined.

Genetic research is moving at breakneck speed, taxing our ability to construct timely public policies that safeguard the promise of genetics to improve health. Surveys and polls tell us that the public is worried about the balance between benefit and harm posed by these new technologies. Based on these concerns, growing numbers of individuals and families have decided not to pursue genetic tests or services — the hard-earned products of genetics research.

In the Midst of the Genetics Revolution, Healthcare Consumers Have No Safety Net

1. In the midst of the Genetics Revolution, people who could benefit from the new technologies are afraid to use them. They are afraid to have genetic tests or participate in research because they are losing their insurance and their jobs if their insurance companies and their employers learn the results of those tests.
2. Congress could put these fears to rest once and for all by enacting legislation that makes it illegal for insurance companies to deny coverage and for employers to refuse to hire.

promote or fire people based on genetic test results. This would encourage people to take advantage of the rapid advances in genetic testing and other new technologies that can improve public health, alleviate human suffering and extend productivity.

These protections will ensure true nondiscrimination and facilitate the future sustainability of the biotechnology and healthcare industries.¹

Speaking on Behalf of the Genetic Alliance, Families and the Public

I represent the Genetic Alliance – the largest international coalition representing more than 600 lay advocacy, research, health professional, public and private sector organizations and their millions of members. The mission of the Genetic Alliance is to leverage the collective voices of individuals and families living with genetic condition. Since 1986, we have worked to speed the translation of scientific and technological advances into quality healthcare and consumer-informed public policies. We are also founding members of the Genetic Fairness Coalition.

We are in constant contact with individuals struggling not only with the effects of life threatening conditions, but also with discrimination or with the fear of discrimination. Their struggles reaffirm the principle that “Genetic information is inherently personal and must be treated as confidential and proprietary.” (Alliance Guiding Principle)

We are All at Risk. “Genetics Is About ALL of US”

We also represent those who do not yet understand that ‘Genetics is about ALL of us.’ Because every man, woman and child has some genetic predisposition, condition or disease resulting from inherited or acquired genetic changes.

Tests are currently available for hundreds of genes, most of which are associated with relatively rare disorders. However, that number will grow to thousands with an understanding of the genetics of more common health problems. For the most part, these will be predictive tests, opening windows to early detection and prevention of diseases currently thought to be untreatable. For every person identified with a genetic disease, many non-symptomatic family members who would benefit from the knowledge gained from a genetic test should be identified as well. This will also create an explosion in the

ability to identify risk factors and make predictions for a broad range of health problems – from rare conditions to common complex diseases.

With this explosion comes a sense of greater risk for disease. Our real risk has not changed, but our awareness of risk has. We do not understand the exact implications of these newly identified risks. It may take us 100 years to determine whether certain risk factors are meaningful or not and to what degree, taking mitigating and co-mingling factors of other genes and the environment into account. It is now even more apparent that genetic conditions are universal and we are all increasingly at risk for genetic discrimination.

Does Genetic Discrimination Based on Predictive Genetic Tests Really Happen?

Through our Genetics Helpline, Discrimination Survey and hundreds of advocacy organizations, hundreds of people have come to the Genetic Alliance with their stories about the unauthorized use of genetic information in employment and insurance coverage decisions. This should not come as a surprise. We live in a society with a long history of discrimination based on ethnicity, class, gender, physical and mental impairment and now genetics. We already witnessed the tragic consequences of discrimination based on sickle cell trait test results in the 1970's.

If Genetic Discrimination Is A Serious Problem, Why Aren't People Coming Forward? Why Aren't There Any Test Cases?

First, without a sound scientific and social compass, the public is unable to assess the fairness of the situation in which they find themselves. Second, our legal rights are equally obscure. State and federal laws and regulations make up a complex patchwork of protections that vary by state, health plan and employment situation and create major obstacles to legal action. Third, there are cases out there that have not yet seen the light of day. When people lose their health insurance or employment, they hold tight to the last vestiges of privacy and anonymity at all costs and are reluctant to be burned twice. Fourth, we know that the door is wide open for discrimination and that this potential will certainly increase with all the new predictive tests on the horizon.

Is the Public Concerned about Testing and Research?

We know that people are deciding not to have predictive tests and not to participate in research based on fears that insurance companies and employers will use this information to cancel healthcare insurance and deny them jobs. ²

We Have All Heard Numerous Stories

Without a safety net, there was no way for individuals to use critical information about their own health without fear of discrimination and losing health insurance. It is hard to believe that in one of the most advanced nations on earth, we are driven to undergo anonymous genetic testing for fear we will lose our insurance and access to healthcare.

Why the Potential for Genetic Discrimination Based on Predictive Tests?

Genetic discrimination occurs because most state laws do not prohibit the use of predictive test information in health insurance determinations. In most cases, state protections are inadequate and do not address predictive information specifically. State laws are generally described as a colorful, complex and inconsistent patchwork of definitions, provisions and right to action and often do not address predictive information specifically.

Looking to existing Federal protections, HIPAA's protective jurisdiction is also variable and inadequate, depending on whether someone belongs to an individual or group health plan or his employer is self-insured. In the individual market, there are no protections whatsoever. The genetic condition can be excluded or the premium set as high as the market and consumer can bear. Because there are no restrictions or ceilings to the premium, access can be effectively blocked by pricing someone out of the market. In the small group market, the group member is protected to the degree that rate hikes – resulting from member medical treatments or increased risk – are spread across the group pool. The employer is responsible for how the increased tab for premium increases is covered or shared with employees. In the small business situation, these HIPAA protections result in serious potential vulnerabilities, for both the employee and the business owner. Increased premiums may threaten the solvency of a small business and put owners on the alert for employees whose medical condition and treatments are causing group rate hikes. In a small work environment, health and personal issues are sometimes common

knowledge and the identified employee known to all. As has been well documented in the EEOC case involving Terri Sergeant and her former small business employer, HIPAA regulations leave both the employee and small businesses vulnerable to the misuse of genetic information in making employment decisions.³

With regard to protection under the Americans with Disabilities Act (ADA), people with predictive genetic information will probably not fare too well, given the trend in the courts over recent years. When Congress passed the ADA in 1990, Congress intended that the law would cover individuals with a broad range of diseases, such as epilepsy, diabetes, breast cancer, heart conditions and mental illness. Indeed, some members of Congress even explained that the ADA would protect people who experience discrimination based on predictive genetic information, because such individuals would be “regarded” as disabled and hence covered under the law.⁴

Unfortunately, soon after the ADA went into effect in 1992 and culminating in a trio of cases by the Supreme Court in 1999, the ADA’s scope of coverage has been significantly restricted. Thus, in many cases, individuals with conditions such as cancer, epilepsy, diabetes, heart and respiratory conditions, mental illness, and a range of other health conditions, who have alleged discrimination based on such conditions, have been turned away at the courtroom door on the grounds that they are not sufficiently “disabled” to receive legal protection under the ADA.⁵ In essence, the courts have required that to be covered under the ADA, an individual must be so debilitated by his or her impairment that it is difficult for the person to function at all. Moreover, if such an individual can take medication or receive a device (such as a pacemaker) that will enable the person to function, he or she will not be considered “disabled” under the ADA. In addition, even if an employer refuses to hire an individual expressly because of a health condition, this will not be sufficient to claim that the employer “regarded” the individual as disabled unless the individual can also prove that the employer believes many *other* employers would act the same way. The same reasoning that has eliminated legal protection under the ADA for individuals with a range of health conditions will likely be used to deny coverage under the ADA for individuals with predictive genetic information or family histories regarding such conditions.

Predictive Tests Are Not Relevant to Decisions about Health Insurance Coverage

A person with a positive predictive test result may never manifest the condition. One's actual risk depends on interactions with other genes and with the environment.

We do not yet really know the exact level of risk indicated by the test results. The meaning of test results will evolve over time with longitudinal research that follows participants over their lifetime, assessing the interplay with other genes and the environment and the actual expression or incidence of the condition. While the current risk percentages reflect scientists' best guesses, this is not good enough if the results can be used to deny health coverage and employment and disrupt productive lives.

Test results do not translate directly into healthcare dollar costs for any one particular person. Everything in medicine today is measured in terms of evidence-based and outcomes research and cost benefit analysis. However, the use of predictive test results to make health insurance decisions does not fit this paradigm. Predictive tests are not linear, black and white measures of healthcare dollar liability; currently they may have some meaning for pools of people, but not individuals. The science is too new and the variability of expression for two identical genotypes too great. We cannot measure healthcare dollars or future productivity based on computations using genetic test results as the yardstick.

It is impossible to lump all predictive tests in one category. Health dollars could even be saved through the development of preventative treatments that forestall the occurrence of expensive chronic conditions. In the case of hemochromatosis, for example, early identification could lead to phlebotomy treatments that stop the development of an otherwise insidious, chronic, expensive and possibly fatal condition.

Finally, we all have flawed genes. With so many predictive tests already on the radar screen, we will all be at risk for genetic discrimination.

The Use of Predictive Tests in Health Insurance Determinations Puts People at Increased Risk for New Social and Medical Harms and Poses New Societal Burdens.

The use of predictive tests in health insurance determinations affects individuals and their families in the most personal ways – loss of privacy, healthcare, and employment. That is why people are choosing not to have genetic tests that could, in some cases, save their lives. We know that this strategy, while logical, can put the individual at medical risk,

the family at financial risk, and sometimes results in serious, even fatal, health consequences.

Employers may fear hiring, promoting, or retaining someone whose test results or recommended treatment threatens to raise the group insurance rate. We have already seen this happen in the case of Terri Seargent who was essentially symptom-free – jogging several miles every day – but was fired from her job after her employer learned about her positive genetic test results and preventative medical treatment.

Falling public confidence impacts everyone. We are all waiting for the benefits of biomedical research. However, without nondiscrimination assurances, people will not participate in the very studies that could lead to more precise interpretations of ‘risk’ measures, better understanding about interplay between gene and environment and other genes, and the development of preventative treatments – sometimes for their own condition.

The real measure of genetic discrimination is the potential for broad societal impact and burden. If we systematically exclude individuals and families from healthcare and jobs based on genetic make-up, we are risking the creation of an uninsurable and unemployable genetic underclass at enormous public, moral and economic cost.

Genetic Alliance Recommends These Core Principles to Guide Policy Decision-Making

- We all possess mutations that will become equally and increasingly transparent with tomorrow’s technologies.
- Health insurance and employment in this country are intrinsically linked. They are inseparable.
- It is important to take a broad view of the implications and impact of predictive genetic test results for individuals and families and for the small business employer.
- Without protections by Federal law, genetic discrimination will affect increasing numbers of individuals and families and pose unfathomable social harms. The focus of civil rights advocacy in the 21st century will be genetic discrimination.
- Risk-based health insurance may not work in this new genomics age. How can we have a risk-based health insurance system when the meaning of the risks that are being identified through new genetic tests is unknown? The science is literally galloping ahead of our ability

Genetic Alliance

to understand this new information. This distorts the usefulness of information resulting from genetic tests.

Genetic Alliance Advocates for Comprehensive Federal Nondiscrimination Legislation

Looking to S1053 as a Model for Legislation

- Legislation must cover all genetic information – including family history, medical tests and healthcare service records – which can be used to predict future health risks in healthy individuals.
- Legislation must ensure that those entities holding genetic information about individuals will not disclose it to third parties without the written permission of the individual.
- Legislation must prohibit both health insurers and employers from collecting predictive genetic information and from using it to discriminate in the health care system and the workplace.

Opposition to Unwarranted Discrimination in Health Insurance and the Workplace

Finally, I want to point out that this testimony has focused on the hardships faced by those who experience discrimination based on predictive genetic information or family histories. However, we urge you to consider the fact that if these individuals are eventually diagnosed with a medical condition at some future point (whether such health conditions are genetically caused or not), they should also not be subject to unwarranted discrimination in health insurance and the workplace. As noted above, the reason people with predictive genetic information cannot rely on the ADA is because their brothers and sisters with actual medical conditions cannot rely on the ADA either.

This is why we cannot stop at only legislation for genetic nondiscrimination without clarifying the ADA and considering the need for additional protective legislation. When a healthy individual tests positive for a gene that could cause a condition like Alzheimer's or bipolar disease, it is not always clear if signs of that condition have occurred. To ensure that people will not be afraid to seek treatment and receive a diagnosis, we need to assure them that, if a condition does manifest, their access to healthcare and employment will be protected.

Safeguarding the Potential of Genetics to Improve Health.

Completion of the sequencing of the genome is a wonderful and inspiring scientific accomplishment; however, it has also accelerated the need for universal protections of genetic information that help to describe future risks for health and disease. Assurances against the abuse of personal genetic information will safeguard our hopes for improving public health through new genetics knowledge and technologies.

Congress demonstrated extraordinary vision in funding the mapping of the human genome. It requires an equal measure of vision and courage to pass the legislation that makes it possible for people to benefit from the new tests and technologies and creates a safety net for healthcare consumers. Otherwise, the remarkable achievements of the Human Genome Project will be slow to translation to health applications.

In a country founded on precepts that offer protections against discrimination, based on sex, race or religion, we certainly have room for perhaps the most basic factor of all – our genes, representative of both our shared inheritance and the essence of our diversity.

The Genetic Alliance calls for the unequivocal prohibition of genetic discrimination in health insurance and employment, and all other aspects of life. Every American – regardless of genetic inheritance – is entitled to the protection that Congress alone can provide.

¹ Insurance Industry Sustainability:

Research focusing not only on survival rates and the probability of future disease, but also on future healthcare needs and the availability, effectiveness, and potential cost savings of early intervention, is of great potential benefit. Not only would patients better understand their prognoses, but also physicians could improve treatment modalities, and plan sponsors and insurers could better evaluate the appropriateness of covering specific tests, their likely impact on insurance costs, and their potential implications for risk classification in the individual market.

Some of the key questions that remain are:

- How accurately will genetic tests predict future health care needs?
- Will meaningful interventions be available for genetic disease?
- Will genetically based treatments become available?
- What impact will genetic technology have on overall medical care expenditures?

Policy-makers need a clear understanding of these issues so that proposals regulating the use of genetic testing information can find the best balance between the concerns of the public, the predictive ability of genetic test results, and the affordability of health insurance.

² Genomics and Managed Care: Preparing for the Revolution By: Carl Peterson

[Healthplan 41(5):14-20, 2000. © 2000 AAHP "Concern among consumers is high. In a mid-June Time/CNN poll of 1,200 U.S. adults, three-quarters of respondents feared having health insurers gain access to disease predisposition data. An even greater number (84 percent) were concerned about government access to personal genetic information."

Genomics Research— However, Knowledge and Understanding Remain Modest Release Harris Interactive Polling Date:6/19/01 1,000 Adults polled June 2001 When asked what their greatest fears are, the answers given most often are that genetic information may be misused (45%)”

In genetic testing studies at the National Institutes of Health, 32 percent of eligible people who were offered a test for breast cancer declined to take it because of concerns about loss of privacy and potential for discrimination in health insurance. May 2001

³ Congressional Research Service Report for Congress

RL30006: Genetic Information: Legal Issues Relating to Discrimination and Privacy

The Health Insurance Portability and Accountability Act of 1996

P.L. 104-191, the Health Insurance Portability and Accountability Act of 1996, has been hailed as taking "important steps toward banning genetic discrimination in health insurance" but has also been criticized as not going far enough. The Act prohibits a group health plan or issuer of a group health plan from using genetic information to establish rules for eligibility or continued eligibility and provides that genetic information shall not be treated as a preexisting condition in the absence of the diagnosis of the condition related to such information. It also prohibits a group health plan or issuer of a group health plan from using genetic information in setting a premium contribution. However, the Act would not prohibit group health plans or issuers of plans (i.e., insurers) from requiring or requesting genetic testing, does not require them to obtain authorization before disclosing genetic information, and does not prevent them from excluding all coverage for a particular condition or imposing lifetime caps on all benefits or on specific benefits. In addition, this Act does not address the issues of the use of genetic information in contexts other than health insurance such as employment.

⁴. See, e.g., 136 Cong. Rec. H4627 (statement of Rep. Waxman).

⁵ Footnote: For a comprehensive discussion of how the ADA's coverage has been significantly restricted, see Feldblum, Definition of Disability under Federal Anti-Discrimination Law: What Happened? Why? And What Can We Do About It? 21 Berkeley Journal of Labor and Employment Law 91 (2000)

Genetic Alliance



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April 1, 2004

The Honorable J. Dennis Hastert
Speaker
US House of Representatives
Washington, DC 20515

Dear Speaker Hastert,

We urge you to consider and pass the Genetic Information Nondiscrimination Act [S. 1053], to prohibit discrimination on the basis of genetic information with respect to health insurance and employment. This bill was passed unanimously (95-0) in the Senate last year, is strongly supported by the President and currently sits at the desk in the House.

We believe that all genetic information, including family history, deserves strong and enforceable protections against misuse in health insurance and employment. Such safeguards will protect the rights, privacy and confidentiality of the individual and their family.

This is an exciting and hopeful time for genetic medicine. It is imperative, however, that we, the public, can take full advantage of new medical advances that could help prevent disease before it develops. Genetic nondiscrimination legislation will reduce the likelihood of genetic information being misused in health insurance or employment decision-making. Genetic information is merely predictive information. Simply having a positive genetic test does not mean one will develop a disease -- thus this information should not be used to make decisions about insurance coverage or employment.

As biomedical research advances, genetic testing will become a critical tool in the provision of healthcare. As a result, many more people will know about their own genetic makeup, putting them at risk of genetic discrimination. These issues will affect you, your family members, neighbors and colleagues. We urgently request that you pass a genetic nondiscrimination bill that truly protects all of us from that risk now and into the future.

Thank you for all your hard work and efforts on this critical issue. We look forward to continuing to work with you to ensure that the Genetic Information Nondiscrimination Act passes and can be signed by the President this year.

Sincerely,

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American Association on Mental Retardation – Washington, DC
American Autoimmune Related Diseases Association (AARDA) – Eastpointe, MI
American Cancer Society
American College of Medical Genetics – Bethesda, MD
American College of Preventive Medicine – Washington, DC
American Health Information Management Association (AHIMA) – Washington, DC
American Osteopathic Association – Washington, DC
American Psychiatric Association – Washington, DC
American Society of Clinical Oncology – Alexandria, VA
American Society of Human Genetics – Bethesda, MD
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The Arc of the United States – Washington, DC
Association of American Medical Colleges – Washington, DC
The Association of Women’s Health, Obstetric and Neonatal Nurses – Washington, DC
The Barth Syndrome Foundation, Inc. – Perry, FL
B'nai B'rith International – Washington, DC
BCCNS Life Support Network – Burton, OH
Boston Cure Project for Multiple Sclerosis – Arlington, MA
BPEI Family Network – Pullman, WA
Building Resources Corp. – Minneapolis, MN
CARES Foundation, Inc. – Short Hills, NJ
Caring Voice Coalition – Meridian, ID
Chromosome 18 Registry & Research Society – San Antonio, TX
Citizens for Quality Sickle Cell Care, Inc. – New Britain, CT
The Coalition for Heritable Disorders of Connective Tissue – Washington, DC
Colorectal Cancer Network – Kensington, MD
Commission on Social Action of Reform Judaism – Washington, DC
Cornelia de Lange Syndrome (CdLS) Foundation, Inc. – Avon, CT
Cystic Fibrosis Foundation – Bethesda, MD

The Digestive Disease National Coalition – Washington, DC
Duke University Medical Center – Durham, NC
Dysautonomia Foundation, Inc. – Washington, DC
The Dystonia Medical Research Foundation – Chicago, IL
Ehlers-Danlos National Foundation – Los Angeles, CA
Familial Dysautonomia Hope Foundation – New York, NY
Family Voices – Albuquerque, NM
FORCE: Facing Our Risk of Cancer Empowered – Coral Springs, FL
Foundation for Ichthyosis & Related Skin Types, Inc. – Lansdale, PA
GeneDx, Inc. – Gaithersburg, MD
Genetic Alliance – Washington, DC
Genetic Alliance BioBank – Washington, DC
Global Health Initiatives, Inc. – Potomac, MD
Gluten Intolerance Group of North America – Seattle, WA
Hadassah, the Women's Zionist Organization of America – Washington, DC
The Hemophilia Federation of America – Lafayette, LA
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Hunter's Hope Foundation – Orchard Park, NY
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International Rett Syndrome Association – Clinton, MD
International Society for Mannosidosis & Related Diseases – Baltimore, MD
International Society of Nurses in Genetics, Inc. (ISONG) – Newton, IA
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National Coalition of Health Professional Education in Genetics – Lutherville, MD
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Spina Bifida Association of America – Washington, DC
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The University of Iowa
Iowa City, IA

Lori Wills
Pharmaceutical Representative
Radford, VA

Dayna J. Wolff, Ph.D.
Director, Clinical Cytogenetics and Molecular
Pathology
Associate Professor, Department of Pathology and
Laboratory Medicine
Medical University of South Carolina
Charleston, SC

Jacqueline Wolff, CRRN, CHPC, M.ED
Newton, MA

Patricia Wood, President
NBIA Disorders Association
El Cajon, CA

Sarah Wright
Sr. Project Specialist
Marriott International
Arlington, VA

Huiying Yang, M.D., Ph.D.
Director, Genetic Epidemiology Program
Cedars-Sinai Medical Center
Associate Professor of Pediatrics and Epidemiology,
UCLA
Los Angeles, CA

Colleen Yinger
Thousand Oaks, CA

Joan Young
Platelet Disorder Support Association
Potomac, MD

S. Robert Young, PhD, FACMG
Professor, Obstetrics & Gynecology
Director, Cancer and Research Genetics
Founding Director, M.S. Genetic Counseling
Program
Univ. of South Carolina School of Medicine
Columbia, SC

Jonathan Youse
Nashville, TN

Kathleen Zeitz
breast cancer survivor
Omaha, NE

Katherine Crotty Zuback
Woodbine, MD

Audra Zuckerman
Democrat
New York, NY

Janice Zurich, M.D.
Director, Genetics Center
Indiana University School of Medicine
Northwest Center for Medical Education
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International Signers

Yann Joly, lawyer
Research Associate
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Montreal, Quebec, Canada

Dalia Mikha
Melbourne, Victoria
Australia

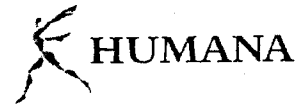
Loraine Oman-Ganes MD, FACMG
Clinical Geneticist
American Society of Human Genetics affiliate
Toronto, Ontario, Canada

Patricia Pearsell
Canadian Representative of the Basal Cell
Carcinoma
Nevoid Syndrome Life Support Network.
Whitby, Ontario, Canada

John Sulston
Human Genome Project member
Vice-Chair of the UK Human Genetics Commission
Cambridge England

Humana Inc.
500 West Main Street
Louisville, KY 40202

December 8, 2004



Ms. Sarah Carr
SACGHS
Office of Biotechnology Activities
National Institutes of Health
6705 Rockledge Drive, Suite 750, MSC 7985
Bethesda, MD 20892-7985

Dear Ms. Carr:

Humana Inc., headquartered in Louisville, Kentucky, is one of the nation's largest publicly traded health benefits companies, with approximately 6 million medical members located primarily in 15 states and Puerto Rico. We offer coordinated health insurance coverage and related services - through traditional and Internet-based plans - to employer groups, government-sponsored plans, and individuals. As of January 2004, Humana serves over 350,000 Medicare beneficiaries in markets across the nation.

The purpose of this letter is to provide comments for the record with respect to the October 18th meeting of the Secretary's Advisory Committee on Genetics, Health and Society. We are concerned that one of the participant's testimonies, specifically the testimony of Ms. Heidi Williams, may lead others to wrongly conclude that Humana underwrites new applicants for insurance on the basis of genetic information. It has never been Humana's policy to make a coverage determination based on someone's status as a carrier for genetic disease or based on the results of a genetic test. We make coverage decisions based on someone's diagnosed condition.

On February 19, 2004, we discovered that an underwriter had incorrectly declined Ms. William's application for dependent coverage with us. Lorie Hoekstra, of our company, contacted Ms. Heidi Williams on this date to apologize for this error and offered to extend coverage retroactively to the original effective date she requested of 9/1/03. Because of our mistake, Ms. Hoekstra stated that Humana would cover Ms. William's family premium costs through March 31, 2004. Ms. Williams indicated that she wanted to discuss this offer with her husband and would get back with us. We have attached a copy of the letter that was sent to Ms. Williams outlining this offer. She did call Ms. Hoekstra several days later accepting this offer. Coverage was issued for one month and it was then canceled.

Since then, our company has also undertaken an extensive training program to alert all of our underwriters to the appropriate written procedures to follow on genetic carrier status to ensure that this error will not happen again.

Page Two
December 8, 2004

Finally, Humana strongly supports S. 1053, the Genetic Information Nondiscrimination Act. In essence, this bill outlines the business practices we encourage all health plans to follow.

We appreciate the opportunity to provide these comments. If you have any questions, please do not hesitate to contact me.

Sincerely,

Heidi Margulis

A handwritten signature in cursive script that reads "Heidi Margulis".

Heidi Margulis
Senior Vice President, Government Relations
Humana Inc.

Enc

February 23, 2004

Heidi Williams
623 Slaughter Ln
Cecilia, KY 42724

Dear Ms. Williams:

This letter confirms our telephone conversation on Thursday, February 19 regarding your recent appeal of decision declining coverage for your children, Jesse and Jayme. As I explained, the decision was made in error as your children while carriers, have not manifested any of symptoms of the condition AAT. It is not Humana's practice to deny coverage based on carrier status of a genetic condition.

As we discussed, we would like to offer retroactive coverage on Jesse and Jayme effective September 1, 2003. We will cover back premiums from September 1, 2003 through March 31, 2004. Starting April 1, 2004, your monthly premium will be \$105.87. This premium is guaranteed through August 31, 2004 unless you physically change your resident address or add/delete family members.

Enclosed is the application that you originally completed by phone for Jesse and Jayme. Please return the application and any related forms to our office to my attention and I will make sure it is processed immediately. Coverage is not in force until the enclosed documents are returned. If you prefer, please feel free to return this information via facsimile to 920-632-0457, attention Lorie Hoekstra.

Again, I apologize for this error in processing your children's application for coverage. We appreciate your interest in HumanaOne and thank you for your patience. Please call me at (262) 951-2512 if you have further questions.

Sincerely,

Lorie Hoekstra, RN, MHP
Director of Underwriting
Humana Individual Underwriting Department



LABCORP PUBLIC POLICY STATEMENT

LABCORP SUPPORTS GENETIC NON-DISCRIMINATION LEGISLATION

Issued September 7, 2004

Statement of the Issue

Major medical advances based on the science of genomics have the potential to dramatically transform and improve health care. Genomics, the scientific discipline of mapping, sequencing, and analyzing the genome, is now transitioning to an emphasis on genome function and clinical applications. This foreseeable and natural shift in research and development has exposed the increased importance and benefits of genetic and genomic testing.

Genetic or genomic testing involves the analysis of DNA, RNA, chromosomes, proteins, or metabolites to detect genotypes, mutations, or chromosomal changes associated with a predisposition to develop or a capacity to carry a disease. These innovative testing technologies can identify and support treatment of the cause of disease, rather than merely its symptoms. Performed at the molecular level, this testing provides the opportunity for early intervention through prediction of disease predisposition. Genetic testing can also increase the potential for successful therapy by allowing a physician to individualize treatment; tests are now available that allow monitoring of therapy effectiveness, and avoidance of toxicity or adverse reactions.

However, many individuals are concerned that obtaining genetic information important to their health care is not worth the risk of discrimination by insurers or employers.¹ Insurance companies and employers have a potential interest in promoting genetic screening to identify individuals carrying disease-associated genes.² Employers have a strong economic incentive to identify potential employees who will likely remain healthy, thus reducing labor costs.³ Insurance companies assert that they should be free to use genetic information to avoid the risk that people who know they will become ill will try to obtain insurance at regular rates.⁴ There are also concerns that discrimination could occur when individuals decide to forego genetic testing requested or demanded by an employer or insurer.

The fear of discrimination in employment and insurance practices based on genetic information is not unjustified or irrational; while such practices may not be widespread at this time, it is clear that unfair and discriminatory uses of genetic data already occur under current conditions, and have occurred in the past.⁵

While many states have enacted some type of genetic non-discrimination law, these laws vary widely with respect to their application and level of protection.⁶ Although Federal law provides some limited protection against genetic discrimination through the Americans with Disabilities Act of 1990 (ADA), Title VII of the Civil Rights Act of 1964, and the Health Insurance Portability and Accountability Act of 1996 (HIPAA), there is currently no comprehensive Federal law which prohibits genetic discrimination in employment and health insurance. Consequently, currently enacted state and federal laws are inadequate to prevent some forms of genetic

¹ Guttmacher A and Collins F. Welcome to the Genomic Era. *N Engl J Med.* 2003; 349(10): 996-99.

² Billings PR, Kohn MA, deCuevas M, et al. Discrimination as a Consequence of Genetic Testing. *Am J Hum Genet.* 1992; 50:476-482.

³ Council for Responsible Genetics. Genetic Discrimination. Available at <http://www.genewatch.org/programs/privacy/genetic-disc-position.html>. Accessed September 7, 2004.

⁴ Clayton EW. Ethical, Legal, and Social Implications of Genomic Medicine. *N Engl J Med.* 2003; 349(6): 562-69.

⁵ Billings PR, Kohn MA, deCuevas M, et al. Discrimination as a Consequence of Genetic Testing. *Am J Hum Genet.* 1992; 50:476-482.

⁶ Clayton EW. Ethical, Legal, and Social Implications of Genomic Medicine. *N Engl J Med.* 2003; 349(6): 562-69.

discrimination,⁷ and many people believe that only the passage of Federal legislation mandating uniform national protection against the misuse of such information will lead to full use of genetic testing.⁸

Policy Position

Laboratory Corporation of America® Holdings (LabCorp®) encourages and supports legislation designed to prohibit discrimination on the basis of genetic information, particularly with respect to health insurance benefits and employment. The benefits of genetic testing can only be fully realized when the fear of genetic discrimination, and its actual practice, are eliminated from the health care system. Federal legislation establishing a national and uniform basic standard for genetic non-discrimination is the best way to accomplish this goal.

Supporting Information

In a study among genetic counselors, 92% of the genetic counselors interviewed stated that adult patients seeking counseling for presymptomatic conditions had some level of awareness and concern about the potential for insurance discrimination, and 67% of the counselors interviewed said adult patients have a high level of concern.⁹ According to 38% of the counselors interviewed, many patients decline testing due to insurance discrimination concerns.¹⁰

Another study involving 29 responses to an advertisement soliciting cases of possible genetic discrimination described 41 separate incidents of possible discrimination, including 32 insurance-related claims and 7 employment-related claims.¹¹ Problems cited included difficulties in obtaining insurance coverage and finding or retaining employment.¹²

In a survey of 1,000 individuals who were at risk for genetic conditions, 22% said they had experienced some form of genetic discrimination.¹³

In 1982, 1.6% of companies surveyed were using genetic testing for employment purposes.¹⁴ A 1989 survey by the Congressional Office of Technology documented at least 5 Fortune 500 companies that were conducting genetic screening on their employees.¹⁵ In 1997, the American Management Association found that the number of companies using genetic testing for employment purposes had increased to 6-10%.¹⁶

Pre-employment genetic screening at Lawrence Berkeley Laboratory led to a court decision in favor of employees who were the victims of genetic discrimination.¹⁷ In that case, genetic tests were performed on female employees for the purpose of testing for pregnancy and on black employees for the purpose of testing for sickle-cell trait, a condition present almost exclusively in the African-American population; genetic testing becomes a straightforward violation of Title VII of the Civil Rights Act of 1964 when employees or applicants are singled out based on race or sex.¹⁸

⁷ Billings PR, Kohn MA, deCuevas M, et al. Discrimination as a Consequence of Genetic Testing. *Am J Hum Genet.* 1992; 50:476-482.

⁸ Guttmacher A and Collins F. Welcome to the Genomic Era. *N Engl J Med.* 2003; 349(10): 996-99.

⁹ Hall MA and Rich SS. Patients' fear of genetic discrimination by health insurers: the impact of legal protections. *Genet Med.* 2000; 2(4): 214-221.

¹⁰ *Id.*

¹¹ Billings PR, Kohn MA, deCuevas M, et al. Discrimination as a Consequence of Genetic Testing. *Am J Hum Genet.* 1992; 50:476-482.

¹² *Id.*

¹³ National Workrights Institute. Genetic Discrimination in the Workplace Fact Sheet. Available at http://www.workrights.org/issue_genetic/gd_fact-sheet.html. Accessed September 7, 2004.

¹⁴ *Id.*

¹⁵ Council for Responsible Genetics. Genetic Discrimination. Available at <http://www.genewatch.org/programs/privacy/genetic-disc-position.html>. Accessed September 7, 2004.

¹⁶ National Workrights Institute. Genetic Discrimination in the Workplace Fact Sheet. Available at http://www.workrights.org/issue_genetic/gd_fact-sheet.html. Accessed September 7, 2004.

¹⁷ *Norman-Bloodsaw v. Lawrence Berkeley Laboratory*, 135 F. 3rd 1260, 1269 (9th Cir. 1998)

¹⁸ French S. Genetic Testing in the Workplace: The Employer's Coin Toss. 2002 *Duke L. & Tech. Rev.* 0015.

In the Equal Employment Opportunity Commission's first case challenging genetic testing of employees under the Americans with Disabilities Act (ADA), Burlington Northern Santa Fe Railway admitted to conducting undisclosed genetic testing on its employees after the workers complained of carpal tunnel syndrome stemming from work-related activities; the defendant admitted no wrongdoing, and the case was settled on May 6, 2002.¹⁹ Although the company's motive for pursuing tests to detect a mutation associated with hereditary neuropathy with liability to pressure palsies was never made clear, it seems reasonable to suspect that the company would have tried to deny disability benefits to any employee who had such a mutation, arguing that the mutation, and not the job, caused the carpal tunnel syndrome.²⁰ While the EEOC has sought to classify genetically predisposed individuals as possessing an "impairment" qualifying for protection under the ADA, and has stated as its policy that basing employment decisions on test results revealing genetic predispositions violates the ADA, the ADA has not been formally amended to include such a provision; thus, the ADA does not prevent employers from requiring pre-placement medical exams, which may include genetic tests, and employers are still not prevented from requiring workers to consent to a general medical record release or disclosure of family history including genetic information.²¹ The ADA was created to protect only those who are presently disabled, and interpreting it to include all persons with a potential to become disabled in the future, including those who are genetically predisposed to becoming disabled, would violate the original intent of the ADA.²²

Most people who have health insurance are insured through their employers, and many employers have self-funded insurance plans which are governed by the Employee Retirement Income Security Act (ERISA), which preempts state laws and does not protect against adverse insurance actions on the basis of genetic information.²³ In passing HIPAA, Congress banned certain uses of genetic information in determining insurance eligibility, but it placed no limits on rate setting,²⁴ does not preclude limiting benefits, and does not prevent insurance companies from asking people to be tested or from asking for test results.²⁵

Since genetic conditions occur at a fairly stable rate and are already reflected in the actuarial tables used by insurance companies, it is misleading for insurance companies to suggest that they would be negatively affected by insuring people at risk for genetic conditions.²⁶

For further information related to this policy statement, please contact:

LabCorp Public Policy & Advocacy Department
430 South Spring Street
Burlington, North Carolina 27215
Phone: (800) 222-7566 extension 65040
E-mail: hortond2@labcorp.com

For more information about LabCorp, please visit our website at www.labcorp.com.

¹⁹ *Id.*

²⁰ Clayton EW. Ethical, Legal, and Social Implications of Genomic Medicine. *N Engl J Med.* 2003; 349(6): 562-69.

²¹ French S. Genetic Testing in the Workplace: The Employer's Coin Toss. 2002 *Duke L. & Tech. Rev.* 0015.

²² *Id.*

²³ Steinberg K. Risks Associated with Genetic Testing: Health Insurance Discrimination or Simply Business as Usual? *J Amer Womens Assoc.* 2000; 55(4): 241-2. Available at www.cdc.gov. Accessed September 7, 2004.

²⁴ Clayton EW. Ethical, Legal, and Social Implications of Genomic Medicine. *N Engl J Med.* 2003; 349(6): 562-69.

²⁵ Steinberg K. Risks Associated with Genetic Testing: Health Insurance Discrimination or Simply Business as Usual? *J Amer Womens Assoc.* 2000; 55(4): 241-2. Available at www.cdc.gov. Accessed September 7, 2004.

²⁶ Council for Responsible Genetics. Genetic Discrimination. Available at <http://www.genewatch.org/programs/privacy/genetic-disc-position.html>. Accessed September 7, 2004.

Secretary's Advisory Committee on Genetics, Health and Society
Fifth Meeting
October 18-19, 2004, Bethesda, MD

Public Commentary on behalf of Myriad Genetic Laboratories, Inc. re: Perspectives on Genetic Discrimination, scheduled for Monday October 18th between 4:30-5:00pm EST (5 minutes allowed for comment).

Myriad representative: Gary Martucci

Good afternoon.

It is clear that the fear of genetic discrimination is preventing high-risk patients from accessing genetic services. My name is Gary Martucci; I am the Director of Strategic Alliances for Myriad Genetic Laboratories. Myriad began providing clinical genetic testing for common hereditary cancer syndromes 8 years ago. In 1996 two of the greatest barriers to genetic services were insurance coverage and the fear of genetic discrimination. Since 1997, I have been responsible for securing coverage and reimbursement for genetic testing from health insurers and managed care organizations nationwide. Myriad's experience is such that genetic testing for common hereditary cancer syndromes is paid for by insurers 90% of the time at an average of 90% coverage. Therefore, the insurance coverage barrier has been effectively eliminated. Unfortunately, the fear of genetic discrimination has not.

For almost 8 years I have had the opportunity to discuss genetic services with hundreds of medical directors, physicians and patients across the United States. The concern and fear about discrimination arises in virtually every

discussion. To reduce the anxiety around genetic discrimination, Myriad has implemented a policy that patient test results are not released to anyone other than the ordering healthcare provider or designee without the patient's express written consent. Insurance plans representing approximately 200 million covered lives comply with this policy because they recognize the clinical value of cancer genetic testing, which leads to the most effective medical interventions. Our policy, along with numerous state and federal laws that prohibit employment and health insurance discrimination, result in numerous protections for consumers of cancer genetic tests, yet there still remain gaps. The fear of genetic discrimination remains the most commonly cited reason for both patients and healthcare providers to not utilize genetic services to prevent life-threatening cancers.

We find ourselves in an awkward place. A large body of literature demonstrates the benefits – both clinical and psychological – of cancer genetic testing. While, peer-reviewed literature, suggests that actual genetic discrimination is not a significant problem the media continue to portray genetic discrimination as a common risk to individuals poised to take advantage of the health benefits offered by genetic services. Roth et al echo many experts' opinions when they state, quote, "Unless these people believe that they and their families will be adequately protected from discrimination and from the possibility of losing or being denied health insurance, many will choose not to be tested for genetic conditions or predisposition to disease," end quote. Therefore,

comprehensive legislation is a necessity, or the media and other uninformed stakeholders will continue to use the fear of genetic discrimination to dissuade patients from appropriate healthcare.

In the arena of hereditary cancers, genetic services and testing offer the hope to reduce the burden of disease that many families suffer. Fortunately, tens of thousands of individuals have benefited from the power of genetic tests to guide their providers in the most appropriate medical management. While this number may seem impressive, there are over a million people in the United States who carry mutations predisposing them to cancer, yet fewer than 2% know it. While we know that there are several key issues that underlie these statistics – for example, the lack of awareness of genetic tests, and the need for educational and clinical support for healthcare providers – we have consistently found that the fear of genetic discrimination is a top reason for refusing genetic services and testing. To integrate the promise of the Human Genome Project into clinical care, patients, clinicians and insurers need the best available information to coordinate medical management. Without the information available from genetic risk assessment, patients and healthcare providers are left with only limited knowledge of how best to manage the risk of disease. Not only does this dilute the benefits of medical management for the patient, it often results in poor allocation of resources: truly high-risk patients may not pursue risk-reducing options, while truly low-risk individuals may overuse the medical system due to their fear of developing cancer.

It is our experience that patients interested in obtaining potentially life-altering genetic services sit idle in fear of discrimination. The science and technology to positively influence a patient's outcome are with us today. It is our responsibility to make sure that patients are confident that there will be no negative consequences in insurance coverage or employment for pursuing this important information.

Perception is reality, and the public's perception is that genetic discrimination is a serious threat. People have allowed an essentially nonexistent or limited risk for discrimination to prevent them from managing a very real risk of developing cancer. We must eliminate the fear of genetic discrimination to allow the public to participate in the benefits of genetic medicine. Comprehensive legislation will reassure the public and get media coverage to spread the word. Comprehensive legislation will eliminate the confusion and mixed messages sent to individuals who need these technologies the most. Ladies and gentleman of the Committee, comprehensive federal legislation banning and prohibiting genetic discrimination is the answer.

Thank you.

Reference referred to in comment:

Genetic discrimination in health insurance: an overview and analysis of the issues.
Roth MT, Painter RB, Nurs Clin North Am. 2000 Sep;35(3):731-56.

The problem of genetic discrimination in health insurance will increase as genetic knowledge expands and the number of genetic tests proliferates. Unless appropriate legislative protections are developed and enforced, a consequence of the genetic revolution may be that more people are put at risk for losing their health insurance. The current situation requires people to make difficult choices about taking tests that could save or prolong their lives. **Unless these people believe that they and their families will be adequately protected from discrimination and from the possibility of losing or being denied health insurance, many will choose not to be tested for genetic conditions or predisposition to disease.** Solutions to this problem require continuing research and debate and the creation of new policies and laws that protect the people while maintaining the economic viability of insurance companies. This article explores the problem of genetic discrimination as it relates to health insurance in the United States. The goal of this article is to assist nurses and other health care professionals to better understand the important and complex issues and concepts related to genetics, genetic testing, and genetic discrimination in health insurance.

NBCC

NATIONAL BREAST CANCER COALITION

grassroots advocacy in action

**Testimony of
Fran Visco, President
National Breast Cancer Coalition
Secretary's Advisory Committee on Genetics, Health, and Society
OCTOBER 18, 2004**

Since its founding in 1991, the National Breast Cancer Coalition (NBCC) has changed the world of breast cancer – in public policy, science, industry and advocacy – by empowering those with breast cancer, their families and friends and creating new partnerships, collaborations, research funding opportunities and avenues for access to quality care. NBCC has grown to more than 600 organizations representing several million patients, professionals, women, their families and friends. Coalition members include cancer support, information and service groups, as well as women's health and provider organizations.

The mapping of the Human Genome has brought with it the promise of reducing human suffering by targeting interventions to those at risk of disease. The National Breast Cancer Coalition (NBCC) believes strongly that legislative and regulatory strategies must be established to address the protection of individuals from the misuse of their genetic information at the national, state and local levels of government. Genetic information is uniquely private information that should not be disclosed without authorization by the individual. Improper disclosure can lead to significant harm, including discrimination in the areas of employment, education, health care and insurance.

The 1996 Health Insurance Portability and Accountability Act (HIPAA, P.L.104-191), also known as HIPAA, was the first federal law that took significant steps toward extending protection to individuals from genetic discrimination in the health insurance area by creating privacy standards, but this law does not go far enough.

The time is now to extend protections against genetic discrimination to everyone. The release of the working draft of the human genome sequence in June 2000 and the development of new genetic tests necessitate legislative and regulatory strategies to address the issue of how to protect individuals from the misuse of their genetic information.

Furthermore, the fear of potential discrimination threatens both a woman's decision to use new genetic technologies and to seek the best medical care from her physician. Women are also afraid to enroll in research and clinical trials, and this in turn threatens the ability of the scientific community to conduct the research necessary to understand the cause and find a cure for breast cancer. Many of the women testifying and present in the audience today have experienced exactly these concerns.

NBCC strongly supports the enactment of legislation that would protect millions of individuals against discrimination not only in health insurance but also in the workplace, and that would provide strong enforcement mechanisms that include a private right of action. For these reasons, NBCC supports H.R. 1910, the Genetic Nondiscrimination in Health Insurance and Employment Act authored by Congresswoman Louise Slaughter (D-NY). This legislation prohibits health plans from doing several things including: requesting, requiring, collecting or disclosing genetic information without

prior *specific* written authorization of the individual; using genetic information, or an individual's request for genetic services, to deny or limit any coverage for established eligibility, continuation, enrollment or contribution requirements; and establishing differential rates or premium payments based on genetic information, or an individual's request for genetic services.

This legislation also prohibits employers from: using genetic information to affect the hiring of an individual or to affect the terms, conditions, privileges, benefits or termination of employment, unless the employment organization can prove this information is job related and consistent with business necessity; requesting, requiring, collecting or disclosing genetic information prior to a conditional offer of employment; or under all other circumstances, requesting or requiring collection or disclosure of genetic information unless the employment organization can prove this information is job related and consistent with business necessity; from accessing genetic information contained in medical records released by individuals as condition of employment, in claims filed for reimbursement for health care costs, and other services; and from releasing genetic information without *specific* prior written authorization of the individual.

Most importantly, H.R. 1910 contains strong enforcement language and provides individuals with a private right of action to go to court for legal and equitable relief if they are a victim of genetic discrimination, whether they are subject to discrimination by their health plan or their employer.

NBCC does not support the Genetic Nondiscrimination in Health Insurance and Employment Act (S.1053) passed by the Senate on October 14, 2003 because it does not contain sufficient enforcement provisions.

Unlike H.R. 1910, S. 1053 does not provide individuals with a private right of action should they become a victim of genetic discrimination in the individual insurance market.

NBCC believes that a right with no enforcement is really no right at all. It is for that reason that no matter how carefully a bill is worded, no matter how much effort is put into including "protections" that breast cancer patients need--if that bill does not have a strong enforcement mechanism, then NBCC simply will not support it.

As we can clearly see from the witnesses here today, genetic discrimination is a real and growing problem that needs an immediate solution—not one that should wait until we have further cases of women and men who have experienced this type of discrimination that is so detrimental to their ability to seek quality health care.

Thank you for the opportunity to share the views of the National Breast Cancer Coalition.



National Partnership
for Women & Families

Public Comments

Secretary's Advisory Committee on Genetics, Health and Society

Hearing on Genetic Discrimination

October 18, 2004

On behalf of the National Partnership for Women & Families and the Coalition for Genetic Fairness, thank you for holding this hearing to gather information about the scope and nature of genetic discrimination. The Senate has taken an important step in advancing genetic testing and research by passing the Genetic Information Nondiscrimination Act in October 2003. This legislation will provide much needed protection for all Americans from genetic discrimination in health insurance and in the workplace. We thank President Bush for his support of this legislation, and join with you in the hope that the evidence presented today will encourage the House of Representatives to take action to enact this legislation.

With the completion of the Human Genome project last June, the possibility for genetic testing and research is expanding rapidly. There are now genetic tests for hundreds of disorders, and some of the most widely available tests are for women. Women and families stand to benefit from improved prevention, detection, and treatment of diseases like breast and ovarian cancer. However, all the advances in the world will not help women and families if – by participating in genetic research or taking a genetic test – they can, or fear that they can, be denied job opportunities, health care, or both, based on their genetic information.

In addition to being longtime proponents of genetic nondiscrimination legislation, the National Partnership for Women & Families leads the Coalition for Genetic Fairness, a diverse group of disability, women's advocacy, and civil rights groups that recognizes the need for meaningful protections against genetic discrimination. The scope of this group reflects the impact that this issue has on all Americans.

To illustrate the impact of genetic discrimination and the fear of genetic discrimination, the Coalition developed a report, *Faces of Genetic Discrimination*, which is included in your briefing book. The report notes telling statistics, including the overwhelming opposition of individuals to allowing employers and insurers to access to their genetic information, but also shares the stories of individuals like Heidi, Kim, and Mary. Heidi

was denied health insurance for her children because they were carriers of a gene for a lung condition. Medical professionals knew that the children would never develop the disease themselves, but the insurance company denied coverage because they carried the genetic marker. Kim, a social worker for a human services agency, was fired because of her employer's fears about her family history of Huntington's Disease, which she revealed during a staff workshop on caring for people with chronic illnesses. Mary has a family history of breast cancer, but decided against being tested for the genetic mutations that make women more susceptible to breast cancer, because she feared a positive result would jeopardize her chances for promotion at her law firm.

To allow individuals like these three to realize the full benefit of genetic testing and keep genetic discrimination from standing in the way of improvements in public health, strong meaningful federal protections must be enacted.

The Coalition has developed four core principles that we believe must be part of any legislation:

- All genetic information that predicts future health risks, including family history, must be protected.
- Health insurers and employers must not be allowed to collect predictive genetic information and use it to discriminate in the health care system and the workplace.
- Individuals who experience genetic discrimination must have the right to seek redress through legal action, with access to meaningful remedies.
- Entities holding genetic information about individuals must be prohibited from disclosing it to third parties without the individual's permission.

As science progresses ever more swiftly, it becomes more critical that Congress act to ensure that Americans are protected from genetic discrimination.

Thank you.

**SACGHS testimony: National Society of Genetic Counselors
October 18, 2004**

Good afternoon. I am Kelly Ormond, president of the National Society of Genetic Counselors (NSGC). As you are aware, the NSGC is the leading voice, authority and advocate for the genetic counseling profession, and represents over 2,000 members. Together, our members provide genetic counseling for prenatal, pediatric and adult genetic indications, as well as work in academia, research and biotechnology companies. A high percentage of our clinically practicing members offer some form of pre-dispositional genetic testing on a regular basis, whether carrier testing or presymptomatic testing for adult onset disorders. Today we would like to primarily address two issues related to the provision of genetic services: genetic discrimination, and coverage and reimbursement of genetic counseling services.

NSGC would first like to address the issue of genetic discrimination by employers and insurers, and the related topic of genetic non-discrimination legislation. We have testified on this issue at past SACGHS and SACGT meetings. NSGC has also provided testimony to other organizations including the National Conference of Insurance Legislators (February and July 2004), and is an active member of the Coalition of Genetic Fairness. We have also recently collaborated with FORCE, a cancer advocacy organization, to develop an educational brochure on genetic discrimination. Our organization is disappointed that Senate Bill S1053 was not taken up by the House for discussion in the past year, and we are committed to working with all stakeholders to develop policies that are equitable and fair to the American public.

We would like to address three points in regards to genetic discrimination, beginning by reflecting upon the current status of documented genetic discrimination. It is clear that there are few documented cases of genetic discrimination in either the insurance or employment setting, but the oral testimonies this morning, written testimonies and cases presented in other resources, including the "Faces of Genetic Discrimination" booklet published by the Coalition of Genetic Fairness, have reinforced that it is clearly an ongoing problem for at least a small percentage of families with inherited disorders. In a paper that is currently in press (Apse, 2004), 7% of survey respondents at risk for colon cancer perceived that they or a healthy family member had experienced genetic discrimination based on genetic testing or family history; these reports were primarily around difficulty or denial obtaining health or life insurance coverage or in denial of screening coverage. It remains unclear, since the bulk of these anecdotes remain unpublished, whether the individuals are experiencing discrimination due to a specific disability as compared to discrimination occurring solely based on genetic status, and the extent to which discrimination may or may not be occurring.

Second, regardless of the rate at which genetic discrimination occurs, data suggests that individuals want to keep their genetic information private, as they do all health information (e.g. Kass et al., 2004), and that individuals are afraid that they will be discriminated against on the basis of genetic information. As a result, the topic of potential discrimination is frequently discussed in genetic counseling sessions; as was

reinforced by the health professionals panel earlier, this is usually brought up by the client, rather than by the genetic counselor. We have also heard data, further backed up by published studies, that suggests a proportion of individuals who are candidates for genetic testing, and for whom medical management may be changed based on test results, decline testing on the basis of this fear of genetic discrimination. Specifically, two recent studies document that nearly half of surveyed individuals are highly concerned about genetic discrimination (Armstrong, 2003; Apse, 2004). This fear may result in at risk individuals declining genetic counseling as well as genetic testing, undergoing testing using an alias or in an anonymous manner, not billing health insurance for genetic testing, or obtaining life insurance or other policies prior to undergoing genetic testing (Armstrong, 2003; Shinanman, 2003; Apse, 2004). There are also studies that document that a high percentage of individuals at risk for breast or colon cancer do not tell their physicians or insurers about these risks, or that they ask that the information not be recorded in their medical records (Kass, 2004; Apse, 2004). Such behavior certainly has personal and public health implications on medical management if individuals do not undergo early screening, or if they choose not to share genetic test results with health care providers. While education through the media and health professionals will be useful in minimizing the perception that genetic information is different than other personal health information, fear related to genetic discrimination appears to be pervasive.

As was discussed this morning, it seems clear that both state laws specific to genetic discrimination around health insurance and/or employment discrimination and the federal ADA, HIPAA and Civil Rights statutes may not be comprehensive, and that there are gaps between state legislation which become relevant in our highly mobile society. One critical point that was not discussed earlier is that research data suggests that neither primary care providers nor general public are aware of the potential protections these bills provide (Apse, 2004; Nedelcu, 2004).

As noted in our 2002 position statement, "The NSGC opposes discrimination against an individual with regard to eligibility for or maintenance of employment, insurance coverage or medical benefits on the basis of genetic information. Genetic information includes the results of genetic testing, other tests which reveal genetic information and information gathered upon review of the family history. Consideration of this information is appropriate only when used to protect the individual's best interests." While the NSGC does not support a position of genetic exceptionalism, we strongly support the passage of federal genetic nondiscrimination legislation. Such legislation would likely alleviate the majority of concerns regarding genetic discrimination, and allow members of our society to use genetic information to help clients make informed medical and personal decisions. To quote Paul Miller from a publication several years ago, "whether it [genetic discrimination] is a huge problem or a small problem, it should be prohibited" (2001).

In summary, NSGC supports federal legislation for genetic non-discrimination, and we are available to work with SACGHS to further this matter until such legislation is passed. We are also committed to work with SACGHS and other medical and public policy organizations to educate the members of our society regarding the key issues around

genetic information and privacy, and to address the misconceptions which have, unfortunately, become prevalent.

Finally, I would like to state that in our quest to improve the access of our American society to high quality genomic medicine, it is critical that this committee consider not only the need to decrease risk of genetic discrimination, but also ways to increase access to both high quality and affordable genetic services. As such, I would like to conclude by addressing the issue of coverage and reimbursement of genetic counseling services, primarily addressing services provided by masters trained genetic counselors. Issues of billing and reimbursement are among the most pressing that face members of the NSGC, and it is one of three areas prioritized in our recent strategic plan. Through our past testimonies, this committee is already aware that coverage and reimbursement for genetic counseling services are limited by the lack of CPT codes and the ineligibility for non-physician provider identification. While some payers contract directly with health plans to include genetic counseling as a covered service, and some services are covered by Medicaid and Medicare when provided to individuals with disabilities, the bulk of genetic counseling services are not currently reimbursed. While we have only preliminarily reviewed the newest draft recommendations, NSGC is pleased to see that the SACGHS and the Secretary's office considers ways to address these two issues. In particular, we are heartened to see that SACGHS is promoting the development and funding of evidenced based studies around clinical genetic services, through any agencies. NSGC offers its strong support in developing and conducting such studies, and have repeatedly been told that studies documenting such value will be critical. We are also pleased that SACGHS is continuing to advocate for the inclusion of masters trained genetic counselors as recognized providers in both private health plans and national provider identification systems. If NSGC can be of additional help as SACGHS is working with these issues, including offering formal testimony on our efforts towards licensure or documentation of the value of genetic counseling, please do not hesitate to contact us. Thank you for your attention.

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A Philanthropic Foundation Supporting
Those At Risk For The Development Of
Cancer

September 16, 2004

RE: Call for Public Comments on Genetic Discrimination by Secretary's
Advisory Committee on Genetics, Health, and Society

Honorable SACGHS Committee Members,

As the executive director of the STAR (Supporting Those At Risk) Foundation, an organization dedicated to supporting individuals who may be genetically predisposed to developing cancer, I have met many individuals for whom genetic discrimination is a very real concern. Some of the STAR Foundation's clients have had cancer, but many others have not and are simply seeking to protect their health as best they can with the knowledge that they are at increased risk for cancer. The proactive efforts of these persons should be rewarded, not punished. Yet many are afraid to let their insurance companies know of their genetic status, so they struggle to receive the healthcare they need.

I have seen individuals pay out of pocket for expensive genetic counseling, genetic testing, and cancer screening procedures in order to keep their genetic information from their insurance companies. Others cannot afford to do this. I have seen individuals forgo genetic testing until their financial and insurance circumstances put them in a better position to pursue testing. I have met several individuals who have withheld their genetic information from their physicians or asked their healthcare providers to withhold the information from their medical records.

For these persons, genetic discrimination is a real and valid issue. It threatens their quality of healthcare and financial stability. When individuals know they are at high risk for cancer and can share this information with their healthcare and insurance providers they are able to use preventative and screening measures that can prevent cancer or diagnose it at its earliest, most treatable stages. These persons' knowledge of their gene status can save their lives, but the great promise of our growing knowledge in predictive cancer genetics can only be realized if it can be used without fear of genetic discrimination.

For those who feel that genetic discrimination is not an issue that affects enough individuals to make it a legislative priority, I submit that I alone know of numerous examples of individuals whose healthcare has suffered because of a fear of genetic discrimination. The documented cases of genetic discrimination and the lack of federal legislation to protect Americans from it have created a widespread climate of fear that inhibits many from receiving the best medical care. Because of this, I urge you to use your influence to support passage of the Genetic Nondiscrimination Act.

Sincerely,

Courtney Nichols
Executive Director
cnichols@supportingthoseatrisk.org

September 17, 2004

Amanda Sarata, MS, MPH
Secretary's Advisory Committee on Genetics, Health, and Society
6705 Rockledge Drive, Suite 750
Bethesda, MD 20892

On behalf of the Society of General Internal Medicine (SGIM), I am pleased to provide comments to the Secretary's Advisory Committee on Genetics, Health, and Society for the hearing on genetic discrimination.

SGIM is an international association of 3,000 physicians and other health professionals who combine treating patients with teaching and conducting research. SGIM is dedicated to improving patient care, medical education, and research in primary care and general internal medicine.

I am an active member of SGIM and serve as Chair of the Human Rights Subcommittee of SGIM's Health Policy Committee. I am a general internist with some expertise in genetics as it relates to primary care and clinical practice. I strongly support passage of genetic non-discrimination legislation during this session of Congress.

Since 1997 I have been active in several national efforts to bring genetics into primary care education through participation in one regional and two national programs. From 1998 through 2001, I served as faculty at The Johns Hopkins University School of Medicine and while there, participated in development of The Johns Hopkins Mid-Atlantic Cancer Genetics Network funded through grants from the NIH-National Cancer Institute. During this same period of time, I was invited to serve on the advisory board of the HRSA-funded Genetics in Primary Care Faculty Development Initiative (GPC) that was designed to enhance the competency of generalist faculties in internal medicine, pediatrics and family medicine to teach genetics and to deliver genetics services in the ambulatory care setting and on the in-patient wards. I continued to serve in a leadership capacity in the GPC through 2003 when the program was moved to the University of Washington. That year I was elected to the board of directors of the National Coalition for Health Professions Education in Genetics.

As an observer in the breast and colorectal cancer genetics clinics, and as a participant in weekly journal club and case discussions at Johns Hopkins Hospital from 1998-2001, I observed often patients requesting that the results of their genetic test be kept confidential. The women seen in the breast cancer genetics clinic were particularly concerned. Several used pseudonyms and many paid in cash to avoid any risk that the information would be obtained by an employer or insurance company. The records of the breast cancer genetics clinic were locked in a clinician/investigator's office and the results of patients' genetic tests and their pedigree were kept separate from the patient's primary Hopkins medical record to provide further protection of confidentiality.

The need for individual protection against health insurance and employment discrimination with regard to genetic testing results came to national attention more than a decade ago. In the mid-1990s, NIH found over 32 percent of eligible women, when offered genetic testing for breast cancer, refused testing because of concerns about health insurance discrimination and loss of privacy. These data have been replicated among first-degree relatives of patients with colorectal cancer who, following lengthy focus group discussions about the risks and benefits of genetic testing, decided two to one to not undergo genetic testing often because of cost and concerns about discrimination.

Many argue that genetic non-discrimination legislation is not necessary because of protection provided by the Health Insurance Portability and Accountability Act of 1996 (HIPAA). HIPAA did 1) prohibit excluding an individual from group coverage because of past or present medical problems, including genetic information; 2) prohibit charging a higher premium to an individual than to others in the group; 3) limit exclusions in group health plans for preexisting conditions to 12 months, and prohibits such exclusions if the individual has been previously covered for that condition for 12 months or more; and 4) state explicitly that genetic information in the absence of a current diagnosis of illness shall not be considered a preexisting condition.

HIPAA did not, however, 1) prohibit an insurer from denying coverage to individuals seeking health insurance in the individual market based upon genetic information; 2) prohibit the use of genetic information as a basis for charging exorbitant premiums for health insurance to individuals seeking coverage in either the individual or group market; 3) limit the collection of genetic information by insurers and prohibit insurers from requiring an individual to take a genetic test; and 4) limit the disclosure of genetic information by insurers.

In 2003, SGIM endorsed the need for genetic non-discrimination legislation and included the issue during its Hill Day visits to Senators and Representatives. SGIM's official policy recommends passage of legislation that includes:

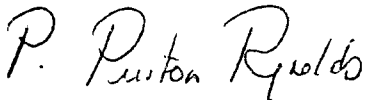
- Protection of individuals from being required to undergo genetic testing by health insurers and the use of this information in individual and group rating
- Protection of individuals from the use of genetic testing information by employers in hiring, promotion and job placement
- Protection of individuals against disclosure of genetic testing information by employers or health insurers that is not directly related to payment of claims or the provision of medical services
- Means for compensation for damages if an individual is harmed because of failure to keep genetic information confidential.

SGIM believes that with strong endorsement by President Bush and unanimous passage of genetic non-discrimination by the US Senate during this session of Congress, it is imperative the House of Representatives follow suit and protect Americans from

discrimination before widespread genetic testing is available for commercial and clinical use.

Thank you again for the opportunity to provide comments to the Advisory Committee. If you have any questions or would like more information, please contact Jennifer Brunelle, SGIM Government Affairs Representative, at (202) 261-4536.

Sincerely,

A handwritten signature in cursive script that reads "P. Preston Reynolds". The letters are fluid and connected, with a prominent initial "P".

P. Preston Reynolds, MD, PhD, FACP

I

II

III

IV

**Research Literature Referenced in Public Written
or Oral Comments on Genetic Discrimination**

Factors Influencing Patients' Decisions to Decline Cancer Genetic Counseling Services

Katherine P. Geer,¹ Mary E. Ropka,² Wendy F. Cohn,² Susan M. Jones,¹ and Susan Miesfeldt^{3,4}

Little is known about the factors influencing patients' decisions about whether to utilize cancer genetic counseling services. The purpose of this study is to identify potential barriers to broad utilization of such services. Of a total of 136 decliners of cancer genetic counseling services at our institution, 117 were deemed eligible to participate. Of these, 73 were randomly selected for study. A total of 37/73 (51%) agreed to participate in a semistructured telephone survey designed to assess the factors that impacted their decisions to decline cancer genetic counseling. An interview script, composed of both closed- and open-ended questions, was used to direct the survey. Interviews were audiotaped. Responses to open-ended questions were content analyzed. Of the participants, 34 were female and 36 were Caucasian. Seventy-two percent of the participants were between ages 36 and 55 years. Participants cited the following reasons for choosing not to proceed with cancer genetic counseling: concern over health insurability for self or family (n = 15); cost (n = 12); emotional impact on self or family (n = 11); no perceived benefit (n = 11); and time commitment (n = 9). These data provide an understanding of patient's attitudes and concerns impacting their decisions to decline cancer genetic counseling. This information provides guidance for the development of interventions designed to limit barriers among patients referred for such services.

KEY WORDS: attitudes; cancer risk counseling; genetic counseling; genetic services; hereditary cancer.

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INTRODUCTION

The identification of genes associated with inherited risk for adult-onset cancers of the breast, ovary, and gastrointestinal tract has presented health care providers and patients with novel tools for identifying increased cancer risk (Lindor and Greene, 1998). Individuals found to have or at risk to have mutations in cancer-associated genes are offered a number of management options based on the possible phenotypic expression of the mutant allele (Burke *et al.*, 1997a, 1997b). Although there is hope that genetic testing for cancer susceptibility will allow for improved risk stratification and targeted use of specific strategies for disease prevention and early detection, the assessment of inherited cancer risk also carries the potential for negative emotional and social impact on the individual and family. Due to the potential harm associated with molecular profiling for cancer susceptibility, genetics and oncology organizations have recommended that genetic testing for cancer risk be offered to patients only after they undergo genetic counseling and provide informed consent (American College of Medical Genetics, 1996; American College of Obstetrics and Gynecology Committee on Genetics, 1997; American Society of Clinical Oncology, 1996; American Society of Human Genetics, 1994; Geller *et al.*, 1997; Klimberg *et al.*, 1999; McKinnon *et al.*, 1997).

Cancer genetic counseling is a comprehensive process with both medical and psychosocial components (Peters and Stopfer, 1996). The medical aspect of this process begins with a clinical evaluation of the patient's personal and family history of cancer, so that an appropriate genetic differential diagnosis can be generated. Following this process, a discussion of relevant medical, genetic, and management implications can occur. This evaluation of the cancer-related history is necessary before DNA analysis is undertaken, so that the clinician may formulate a directed testing strategy. Such an evaluation also allows the patient to gain the knowledge necessary to make an informed decision about molecular testing. Some patients who undergo cancer genetic counseling will make informed decisions not to proceed with DNA analysis.

Despite a growing body of research addressing psychosocial issues associated with DNA testing for hereditary cancer risk (Bluman *et al.*, 1999; Croyle *et al.*, 1997; Lerman *et al.*, 1994, 1995, 1996, 1999; Loader *et al.*, 1998; Lynch *et al.*, 1997; Tessaro *et al.*, 1997; Struewing *et al.*, 1995), little research has been done regarding factors that influence decisions about whether to proceed with the evaluation and counseling process that necessarily precedes molecular testing. In addition, most work examining psychosocial aspects of inherited cancer susceptibility has been conducted in the research setting, where a degree of confidentiality is possible that may not be achieved in a clinic setting. Therefore, previous research may not be generalizable, a potentially important limitation given that most individuals referred for cancer genetic counseling in the future will be seen outside the research setting.

Why Patients Decline Cancer Genetic Counseling

Lack of information on the knowledge, attitudes and concerns of persons referred for clinical cancer genetics evaluation and counseling may impede the broad utilization of these services by at-risk individuals. To understand the potential barriers facing individuals referred for cancer genetic counseling, we surveyed a cohort of individuals who declined such services at our institution.

METHODS

Setting

The multidisciplinary Cancer Genetics Clinic (CGC) at the University of Virginia Health System (UVA-HS) provides genetic counseling and risk assessment for individuals with personal or family histories suggestive of inherited cancer susceptibility. Patients are evaluated through a multistep process. During an intake visit, the patient meets with a psychologist, a genetic counselor, and a medical oncologist; the personal and family histories are taken at this time. Before the second clinic appointment, the pathology reports of tumors in relatives are obtained to confirm reported diagnoses. Conclusions drawn from review of the histories and pathology reports are shared with the patient at the second appointment. This visit includes discussion of the disorder(s) suspected in the family, the mode of inheritance, the risk of occurrence of the disorder in children, risks of tumorigenesis in gene carriers, medical management options, DNA banking and testing, and the impact of this information on the patient and family.

Patients who wish to undergo DNA testing schedule a third visit, during which further patient questions about DNA testing are addressed, the impact of potential results on the patient and family are discussed, informed consent is obtained, and blood is drawn and shipped for analysis. Results disclosure, accompanied by psychological support, occurs at the fourth visit. In addition, patients are offered further visits if they wish to review new information or to ask follow-up questions.

CGC Referral Procedures

The CGCs general policy is that referral for services be made by a clinician who has evaluated the patient's personal and family medical histories. Otherwise, no specific history criteria are required for referral. Self-referrals occur rarely and are generally accepted. Following a patient's referral, a genetic counselor contacts the individual by telephone to discuss the multistep process of evaluation, counseling, and testing, and to schedule an appointment. The potential impact of this assessment is reviewed, and federal and state legislation protecting against genetic discrimination by health insurers is discussed. Individuals who arrange an

appointment receive a letter that similarly describes this process, and reviews the associated risks and benefits.

Design

A cross-sectional design was used for the telephone survey, which was conducted by a one-time interview.

Sample

The initial sampling frame was comprised of 136 individuals who were referred to the CGC between May 1995 and October 1998, but who declined services prior to sample selection. Individuals were excluded from the sampling frame for the following reasons: (1) deceased; (2) terminally ill; (3) known to have obtained genetic counseling services elsewhere; (4) prior indication that they did not want to be contacted further; or (5) cognitive impairment. The final sampling frame included 117 potential participants, from whom 77 (66%) were randomly selected to be invited to participate. Following randomization, four individuals were excluded because they were deceased or cognitively impaired, leaving 73 individuals.

Procedures

The proposed project was approved by the UVA-HS Institutional Review Board. All potential participants were mailed an introductory letter that explained the project, informed them that they would be receiving a call from the interviewer within 2 weeks, and offered \$12.00 for participation. Telephone interviews were conducted by a single investigator who was not a member of the clinic staff. The interviewer attempted to contact potential participants at a minimum of three different times a day over a 5-day period before discontinuing contact attempts.

Survey Instrument

A telephone interview script was developed by the investigators based on clinical expertise, review of the literature, and our own data from a qualitative study investigating factors that influence patients' decisions regarding whether to undergo cancer genetic counseling (unpublished). The interview consisted of both closed and open-ended questions. A member of the research team pilot-tested the interview script with three individuals: two current breast cancer patients at the UVA-HS Cancer Center who were not eligible to receive the survey and one

Why Patients Decline Cancer Genetic Counseling

patient who had already undergone evaluation in the CGC. Participants were asked to provide feedback on the length, flow, and clarity of the interview. Changes were made in the interview script based on this pilot test. The interview script is available on request.

Data Management and Analysis

Interviews were audiotaped and written notes were recorded. Responses to open-ended items were content analyzed and categorized by one investigator into codes representing emergent themes that evolved from the interview data. A second investigator then independently categorized qualitative responses and compared the categories with the initial codes. The coding system was refined, based on input from both investigators. Quantitative data were entered, validated, and analyzed using SPSS for Windows.

RESULTS

Description of the Sample

Of the 73 individuals invited to participate, 54 (74%) were successfully contacted and 37 (51%) agreed to participate in the study. Demographic characteristics of those who agreed to participate are presented in Table I. The majority of these individuals were women (87%), as most referrals to the CGC during the study period were for suspected inherited breast and ovarian cancer predisposition. Seventy-two percent of the participants were between ages 36 and 55 years and 100% were Caucasian.

Twenty-four (65%) of study participants were referred based on family history indications alone and 13 (35%) were referred based on a combined personal and family history of cancer. None of the study participants was referred to the CGC based on a personal history of cancer alone. Among the participants nine (24%) were referred by a surgeon, eight (22%) by a primary care physician, seven (19%) by an oncologist, six (16%) by an obstetrician-gynecologist, four (11%) by self, two by a nonphysician health professional (5%), and one by a gastroenterologist (3%). Twenty-two (60%) participants elected not to schedule an appointment, 12 (32%) cancelled their scheduled appointment, and three (8%) did not present for their scheduled appointment.

Ten (27%) participants chose to receive the \$12.00 honorarium for completing the interview. The remaining participants did not accept the honorarium, and indicated that they preferred to consider participation in the project a donation of their time to cancer genetics research.

Table I. Demographic Characteristics of the Participants
(*n* = 37)^a

	<i>n</i>	%
Sex		
Female	34	92
Male	3	8
Race		
Caucasian	36	100
Age (years)		
18–35	4	11
36–55	26	72
>55	6	17
Highest education level		
<High school graduate	4	11
High school graduate	13	36
College graduate	19	53
Income level		
<\$20,000	3	9
\$20,000–\$59,999	16	47
≥\$60,000	15	44
Biologically related children		
Yes	28	78
<18 years	11	39
≥18 years	17	61
No	8	22

^aOne participant did not complete the section of the interview that assessed demographic information, including race, age, highest education level, and income level. Two additional participants also declined to provide income level information. Percentages were calculated using the total number of responses to each question.

Reasons for Declining Cancer Genetics Services

Each participant was asked, “Why did you decide not to come [for the cancer genetics evaluation]?” and, “Of these [reasons], what were the *most important* reasons for you in deciding not to come [for the cancer genetics evaluation]?” Table II displays the reasons reported by participants for their decisions to decline services. The five most frequent responses to the question “Why did you decide not to come?” were as follows:

Potential impact on insurability for self or family members or both. Fear about health insurance discrimination was given as the most frequent reason (15 participants) for declining services.

I was concerned about how my health insurance would be affected. My insurance company has always covered the things I get done . . . , exams to find out if anything has developed. I didn't want this to change

I thought it might make it difficult to get health insurance in the future, especially if I decided to work for a small company or start my own business

Why Patients Decline Cancer Genetic Counseling

Table II. Reasons for Declining Cancer Genetic Counseling ($n = 37$)

	Why? (n)	Most important reasons (n)
Impact on insurability of self or family members or both	15	14
Cost	12	6
Emotional impact on self or family members or both	11	9
No perceived benefit to having the evaluation	11	4
Time commitment	9	5
Difficulty in collecting family history	4	4
Perceived cancer risk to be low	4	2
Difficulty scheduling or getting to appointment	3	0
Privacy/confidentiality concerns	3	3
Not yet decided whether to have the evaluation	2	2
Other	6	4
Total	79 ^a	53 ^a

^aParticipants were permitted to provide more than one response to each of these questions.

None of these participants mentioned the potential impact of the evaluation on their ability to keep or obtain life or disability insurance.

Cost. Twelve individuals cited cost as a reason for declining services. Concerns over the cost fell into two categories: (1) cost to the individual due to feared or confirmed limited or absent coverage by health insurance ($n = 10$); and (2) cost to the individual due to patient unwillingness to notify the insurance company of the evaluation because of insurability concerns ($n = 2$).

My physician told me it might cost a lot. I was feeling good, so I didn't want the expense
 I would have gone in if my insurance company had covered it completely
 I would have had to pay for it myself because I didn't want my insurance company to know
 I was having it done

Thirty-four participants were covered by health insurance. Of these, nine (26%) contacted their insurance companies to find out if the cost for cancer genetic counseling would be covered. Six of these individuals reported learning that none or very little of the cost would be covered. The remaining three each reported one of the following reasons: not being able to get a "direct answer"; learning that most of the fee would be covered but remaining uncertain about coverage for DNA testing; or being unable to remember their insurance company's response.

Of the six participants who learned that little or none of the cost would be covered by insurance, four reported cost of services as a reason they declined the cancer genetic counseling.

Emotional impact on self or family members or both. Eleven participants reported that they declined cancer genetic counseling because of the potential emotional impact on themselves or their family members. For many of these individuals, the referral to the CGC occurred at a psychologically difficult time. Often these participants explained that they were undergoing treatment for cancer

themselves or experiencing an illness in their family. Due to the hardships they were already facing, they felt emotionally ill-equipped to confront the results of a cancer genetics evaluation.

My daughters weren't prepared for me to do it . . . I was being treated for cancer at the time and they were too scared for me. They couldn't think about themselves because they were focused on getting me through my treatment. It was a bad time

I needed a break from cancer. I didn't want to think about new cancers, new medications, new surgeries

I feared finding out I had the gene. There has been so much suffering in my family. My sister had just died from cancer

No perceived benefit to having the evaluation. Eleven individuals reported that they perceived no benefit to undergoing cancer genetic counseling. These individuals implied that they were comfortable with their current approach to managing their risk. Most of these participants considered themselves at high risk and felt that they had already adopted all of the recommended health care practices that they were willing to consider.

My doctor and I are pretty convinced I have the gene, so we are as aggressive as we can be with my care

I didn't need this knowledge. It wouldn't help me at all. I would prefer to just continue with regular mammograms and exams

Time commitment. Nine respondents cited time commitment as a reason for declining services. For these individuals, common concerns included the number of appointments required for the evaluation, distance from the clinic, and a busy personal schedule.

I live far away. Four trips . . . would be very time-consuming

I was having problems at work. I just didn't have time to drive there and back. I was really busy. I had a lot going on in my life, so I put this on the back burner

The six "other" reported reasons (Table II), each cited by one participant, included: participant resented requirement for pretest counseling; participant obtained information from another cancer genetics service; participant feared pressure to follow health care recommendations based on results; participant's plan to be evaluated for an at-risk relative's benefit was cancelled when the relative underwent DNA testing herself; participant perceived that she had insufficient information about her relative with cancer for evaluation to be useful; and no reason provided.

Timing of Decision to Decline Cancer Genetics Services

In answer to the question, "When did you actually decide not to come for the evaluation?" 12 (33%) participants reported that they decided against the evaluation after obtaining either verbal or written information from the CGC staff.

Why Patients Decline Cancer Genetic Counseling

These individuals indicated that information about the clinic's procedures and risks caused them to decline.

I wasn't sure what I was getting into until I received the letter. This is what made me decide not to do it. . . It became clearer to me that I would have to provide family medical records. . . I also learned that I would have to undergo psychological evaluation. I didn't need this—I had just had a mastectomy

The next most common decision points, each reported by three participants, included: after discussing the service with family members; after obtaining information from a health care professional; and while filling out family history forms. Four of the 37 respondents did not specify when they decided against having the evaluation. Among the remaining 12 participants, no single decision timepoint was cited by more than two participants.

Perceptions of Implications for Health Insurance

All participants were asked if they had investigated how the cancer genetics evaluation would affect health insurance for themselves and their family members in the future. Nine (24%) individuals responded "yes" to this question. These nine participants reported obtaining information from the following sources: a health care provider ($n = 4$), an insurance agent ($n = 2$), a state senator ($n = 1$), a relative ($n = 1$), another cancer genetics service ($n = 1$), or reading ($n = 1$) or both. Eleven (39%) of the remaining participants volunteered that they had not researched this issue because they were already concerned about health insurance discrimination based on prior knowledge. Table III summarizes concerns of the nine individuals who reported investigating this issue, as well as the concerns volunteered by the 11 participants not investigating the evaluation's potential impact on health insurance for themselves or their relatives.

Table III. Perceived Impact on Health Insurance and Insurability

	Investigated ($n = 9$), N	Did not investigate ^b ($n = 11$), n	Total, n
May face increased premiums	4	4	8
May lose current health insurance	3	4	7
May experience difficulty getting new health insurance	4	2	6
May lose coverage of screening procedures or cancer treatment or both	2	3	5
Nonspecific	2	5	7
Total	15 ^a	18 ^a	33

^aParticipants were permitted to provide more than one response to each of these questions.

^bIncludes only participants who volunteered reasons for not investigating the evaluation's implications for future insurability.

Perceptions of Legislation Protecting Against Health Insurance Discrimination

In response to the question, "Are you aware of any laws that protect people with a genetic risk for disease from losing or not being able to get health insurance?" 11 (30%) participants stated that they were aware of legislation providing such protection in Virginia. When asked for their perceptions of the legislation, eight of these individuals felt the law offered inadequate protection due to concerns over unenforceability ($n = 6$), lack of protection in some states outside of Virginia ($n = 4$), and concerns that the law could be changed or revoked at some future time ($n = 2$).

I think it would be hard to enforce . . . Any problems would require a legal battle

I know that insurance companies are not allowed to cancel you in Virginia, but my family is outside of Virginia. And will the law be around tomorrow? I don't think insurance is anything to play around with

Concerns Regarding Implications for Life Insurance and Disability Insurance

All participants were asked if they investigated how the cancer genetics evaluation would affect life insurance and disability insurance for themselves and their family members. Only one participant examined the evaluation's implications for her life insurance; none of the participants investigated the potential impact on the evaluation on their disability insurance. No respondents investigated the potential impact on their relatives' life or disability insurance.

DISCUSSION

We found that the most frequent reasons that at-risk individuals declined cancer genetics services were concerns over insurability, cost, anticipated emotional impact of the evaluation, no perceived benefit to the evaluation, and time commitment required for the evaluation.

Concerns Related to Insurability

Fear of health insurance discrimination represented the greatest barrier to utilization of services by our study participants. Concern over the potential impact of the evaluation on the future insurability of self and relatives was the most common response to both of the questions, "Why did you decide not to come [for the cancer genetics evaluation]?" and "Of these, what were the *most important* reasons for you in deciding not to come [for the cancer genetics evaluation]?" These findings are consistent with previous studies, which identified significant concern among research participants about health insurance discrimination based

Why Patients Decline Cancer Genetic Counseling

on cancer-related DNA testing (Bluman *et al.*, 1999; Lerman *et al.*, 1995, 1996; Loader *et al.*, 1998; Lynch *et al.*, 1997; Struewing *et al.*, 1995; Tessaro *et al.*, 1997).

Among our study participants, concerns related to potential health insurance discrimination included having to pay increased premiums, losing some or all coverage, and inability to obtain new coverage. Such genetic discrimination by health insurance companies has been prohibited in Virginia since 1996, with the passage of the Genetic Information Privacy Act, a temporary law that was to expire after 2 years. Legislators later made the law permanent as of July 1, 1998 by repealing the Act's "sunset clause." However, the majority of the participants who were aware of the Act did not feel adequately protected by it. Given recent research suggesting that evidence of widespread genetic discrimination by health insurers is lacking (Hall and Rich, 2000), the extent of our participants' concerns about this issue may not be warranted.

Research on factors influencing decisions regarding DNA testing for cancer susceptibility has focused concern on health insurance discrimination. This study also investigated insurability concerns related to life and disability coverage. Among our study population, potential implications of the evaluation for health insurability were of much greater concern than for life or disability insurability.

Concerns Regarding Cost

Much of the prior work in the area of genetic testing for inherited cancer predisposition has not addressed the issue of cost because research studies have offered genetic counseling or testing through a research trial for which patients were not charged. In contrast, in our clinical setting, cost was identified as an important reason why many declined services. Individuals who schedule an appointment at the CGC receive a letter that reviews the cost of the cancer genetic counseling; this issue is also discussed by telephone with the genetic counselor prior to the appointment if the patient inquires about fees. Learning this information appeared to have been a deterrent for some participants.

Our finding of cost as a deterrent to obtaining service is consistent with previous research that has been done regarding charges that patients would be willing to pay for various aspects of cancer genetics services. Earlier data found that a substantial proportion of patients initially inquiring about cancer genetic counseling did not keep a clinic appointment, partly because their insurance companies did not cover the visit or would not authorize the referral (Olopade, 1996). Additional studies investigating the fees that patients would be willing to pay for cancer-related DNA testing found that a substantial proportion would be unwilling to pay more than \$25 (Chaliki *et al.*, 1995), more than \$200 (Petersen *et al.*, 1999), or more than \$500 (Durfy *et al.*, 1999).

Our data suggest that cost issues may impact the use of cancer genetic counseling services. This is compounded by other economic issues, including the realities

that third-party payer reimbursement for genetic services has traditionally been low and the expense to an institution to offer such services can be significant (Bernhardt and Pyeritz, 1989).

Concerns Regarding the Cancer Genetics Evaluation's Emotional Impact on the Individual and Family Members

Previous research addressing the psychosocial aspects of DNA analysis for inherited cancer predisposition has shown that many individuals fear or experience negative psychological consequences if they test positive for a constitutional mutation predisposing to tumorigenesis (Lerman *et al.*, 1994, 1995; Loader *et al.*, 1998; Lynch *et al.*, 1997; Struwing *et al.*, 1995; Tessaro *et al.*, 1997). Our findings indicate that psychological concerns may also lead individuals referred for cancer genetic counseling to decline service. Concern regarding the evaluation's emotional impact, cost of services, perception of no benefit, and time commitment were equally common responses to the question, "Why did you decide not to come?" However, concern over the evaluation's emotional impact was cited far more often than these other reasons in response to the question, "Of these, what were the most important reasons for you in deciding not to come?" In particular, many participants felt psychologically unprepared to undergo cancer genetic counseling because of current emotionally difficult circumstances. Patients referred for cancer genetic counseling following their own or a relative's cancer diagnosis may be reluctant to increase stress for themselves or relatives by undergoing the evaluation. Such self-selection against genetic assessment by individuals who fear negative psychological consequences from the process has been described for another adult-onset disorder, Huntington's disease (Codori *et al.*, 1994).

Other Factors Impacting Decisions Regarding Cancer Genetic Counseling Services

Some individuals declined cancer genetic counseling because they did not perceive a benefit to the service. These participants felt the evaluation was unnecessary because they were already taking every measure they were willing to consider in terms of cancer prevention and early detection. Due to their family or personal history of cancer, they felt well aware of their risk and its implications for screening and management issues.

However, this raises two issues about the adequacy of these participants' understanding of the benefits of the service. First, it was not clear that decliners were, in fact, managing their increased cancer risks maximally. Our experience in clinic raises concerns that this might not be the case. For example, our familial breast cancer patients frequently arrive believing that their utilization of high-risk breast

Why Patients Decline Cancer Genetic Counseling

cancer fully manages their increased cancer risks. Given that the common inherited breast cancer predisposition syndromes are associated with increased cancer risks at multiple anatomic sites, this belief is not accurate. Second, patients' declination because of perceived lack of benefit suggests that they may not understand that the evaluation might demonstrate, through eventual DNA testing, that the patient has escaped inheriting a mutant allele that may be present in other relatives.

Patients' perceptions of nonbenefit from cancer genetic counseling may also have roots in emotional distress about the family history of cancer: "... I don't even care if I have the gene, I know breast cancer is in my future" (Pasacreta, 1999).

We did not anticipate the negative impact that time commitment would have on decisions regarding cancer genetic counseling. However, the multistep process of family history review, psychological assessment, and genetic testing that is offered by our service is time-consuming. The multivisit protocol may be especially difficult for patients who must travel a long distance to reach the clinic or those who have busy schedules.

Implications for Policy and Practice

Changes related to health policy and clinical practice may address the barriers to broad utilization of cancer genetic counseling services that were identified through this study. National, rather than state-by-state, legislation protecting individuals from genetic discrimination by health insurers might help patients feel that they and their relatives will not be harmed by seeking cancer genetic counseling. If genetic discrimination is confirmed to be less of a threat than anticipated (Hall and Rich, 2000), education of patients in this regard may also help to remove a barrier to services. In addition, increased coverage of cancer genetic counseling by third party payers may improve patient access to services.

Practical questions arise about how to address the barrier of anticipated emotional distress. Among patients for whom the referral has occurred at an emotionally difficult time, deferral of services until some of these circumstances have resolved may promote comfort on the part of the patient eventually proceeding with service. For other reluctant patients, ongoing telephone contact is not a solution, as provision of intensive and extended support by telephone to someone who has not agreed to be a patient is inappropriate and financially insupportable. In this circumstance the cancer genetics service could ask referring physicians to work with these patients to address their fears. Whether physicians are willing to do this, or are able to do this in light of HMO-imposed time constraints, may be quite variable. Furthermore, some physicians may not have sufficient knowledge about inherited cancer predisposition to fully comprehend the possible outcomes of the evaluation (Giardiello *et al.*, 1997; Jones *et al.*, 1997), thus limiting their ability to effectively counsel patients about the emotional impact of the evaluation.

Patients' perception of lack of benefit from cancer genetic counseling suggests that discussion about what the evaluation could accomplish might address this deterrent. It may be appropriate for the genetic counselor to communicate with the referring health professional about such a patient's perception of nonbenefit from the service and to have them review with the patient the reasons that the evaluation might provide useful information and support regarding the family history of cancer.

Finally, clinics offering cancer genetic counseling services must develop strategies to streamline the evaluation process to limit the time commitment required by the patient.

Study Limitations

There are several limitations to this study. First, our sample size was relatively small. Second, the participants were homogenous in regard to race and gender. Therefore, generalizability to persons with other racial backgrounds or to men may be incomplete. Third, recall bias may have been more problematic for some participants than for others, given that survey responses were based on participants' memories of their decision regarding a referral that occurred at varying lengths of time in the past. Finally, the CGC procedures may not be representative of all cancer genetics services. For example, time commitment may pose less of a barrier to utilization of cancer genetics services that do not require psychological assessment and involve fewer visits. The experience of clinics where a larger proportion of patients are self-referred may also be different.

Recommendations for Future Research

Future research is needed to compare the concerns of individuals who decline cancer genetic counseling with factors influencing those who choose to undergo evaluation. This research should involve multiple clinics and a diverse patient population to maximize generalizability. Future studies should also examine men's perspectives on undergoing these services, particularly because genetic counseling services regarding prostate cancer risk are becoming available and genetic testing for this malignancy is occurring in research settings. Finally, additional research is needed to further examine new issues raised by this study, including perceptions that there is no benefit to cancer genetics services and that the time required for counseling may be a deterrent to services.

Despite the rapid growth in cancer genetic counseling services, much remains unknown about the human dimensions of identification of inherited cancer susceptibility. The ability to determine genetically increased cancer risk will drive the need to expand understanding of patients' responses to the prospect of having their

Why Patients Decline Cancer Genetic Counseling

genetic status assessed both clinically and by DNA testing. Improved knowledge in this area will permit cancer genetics health professionals to design strategies to reduce barriers for patients to these services.

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Genetic Counseling and Testing in Families With Hereditary Nonpolyposis Colorectal Cancer

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Background: Genetic testing to refine cancer risk is available. However, little is known about factors affecting the uptake of testing for the most common hereditary colon cancer, hereditary nonpolyposis colorectal cancer. This study investigated attitudes, intentions, and uptake of genetic testing within newly identified families with hereditary nonpolyposis colorectal cancer.

Methods: Cohort study conducted at the National Institutes of Health between April 15, 1996, and November 20, 1999. Data were collected through questionnaires before semistructured education sessions, individual counseling sessions, and the offer of genetic testing.

Results: Of the 111 eligible first-degree relatives, 51% chose to participate in education and individual counseling sessions. Participation was associated with greater numbers of first-degree relatives with cancer; no association was found between participation and personal history of cancer. Before education and individual counseling sessions, 64% of participants had heard little

about genetic testing for cancers; however, most (97%) stated intentions to pursue testing. Fifty-one percent identified learning about their children's risks as the most important reason to consider testing. Thirty-nine percent identified the potential effect on their health insurance as the most important reason to not undergo testing. Of the 111 eligible first-degree relatives, 51% chose to undergo genetic testing. Participants' intentions to pursue genetic testing were significantly affected by concerns regarding the ability to handle the emotional aspects of testing and the psychosocial effect on family members.

Conclusions: Genetic counseling and testing offers the potential to focus cancer screening resources in individuals truly at increased risk, thereby reducing mortality and morbidity. Fears of discrimination and concerns about psychological and psychosocial issues may present barriers to the use of current cancer prevention strategies, including genetic counseling and testing.

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HEREDITARY nonpolyposis colorectal cancer (also known as HNPCC and Lynch syndrome) is the most common hereditary form of colon cancer. It is estimated to account for between 1% and 5% of the individuals who develop colon cancer.¹ It has been estimated that the prevalence of HNPCC mutation carriers among the general population in Western countries is 1 in 740.² This means that in the United States approximately 380 000 individuals carry an HNPCC mutation and have a greater than 90% lifetime risk of developing one of the cancers associated with HNPCC.³ Some evidence suggests that the risk of colon cancer may vary between men and women.⁴ In addition to colon cancer, individuals with HNPCC are at increased risk (compared with the general population) for developing cancers of the uterus,

small intestine, stomach, urinary tract, kidney, ovary, and other sites.⁵⁻⁷ Lifetime estimates for developing a cancer associated with HNPCC approach 85% for colorectal cancer⁸ and 40% to 60% for uterine cancer⁹ (by age 70 years). Risks for cancer of the small intestine, stomach, urinary tract, kidney, ovary, and brain are also elevated but lower compared with the risks for colon and uterine cancer. Accurate and age-related risks for these cancers are not yet available.¹⁰

Before the identification of the gene mutations, the diagnosis of HNPCC was primarily made based on clinical criteria and family history.¹¹ At a time when the genetic cause of HNPCC was not known, the Amsterdam criteria were developed for research purposes in an attempt to clinically identify individuals and families likely to carry mutations.¹² The Amsterdam criteria are as follows:

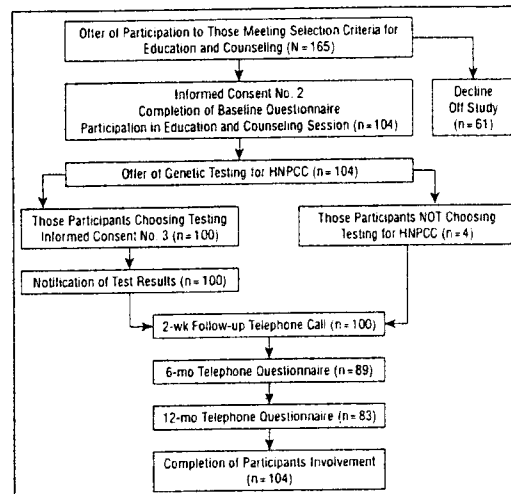
1. Histologically verified colorectal cancer in 3 or more relatives, 1 of whom is a first-degree relative of the other 2;
2. Colorectal cancer involving at least 2 successive generations; and
3. One or more colorectal cases diagnosed before the age of 50 years.
4. All of the above criteria must be met, and familial adenomatous polyposis must be ruled out as a cause for each colorectal cancer.

Subsequent molecular studies have identified several key genes that function in DNA mismatch repair and whose alteration leads to the increased risks for the cancers associated with HNPCC.¹³⁻¹⁵ Six genes have been identified to date, *MLH1*, *MLH3*, *MSH2*, *MSH6*, *PMS1*, and *PMS2*. Current estimates suggest that *MLH1* and *MSH2* account for 80% to 90% of these cancers, while *MLH3*, *PMS1*, and *PMS2* are much less frequent.¹⁶⁻¹⁹ More recent studies^{20,21} suggest a growing role for *MSH6* in families with HNPCC, accounting for 5% to 10% of families in which *MSH2* and *MLH1* mutations have been excluded. It has been suggested that families with mutations in the *MSH6* gene have, in general, a later age of onset (51-52 years), a family history of cancer that does not often meet the Amsterdam criteria, and more frequently occurring endometrial cancer than in families with mutations in *MLH1* or *MSH2*.²²

Understanding is growing about the factors affecting decision making regarding genetic testing and the psychosocial, medical, and behavioral effects of testing for mutations that predispose to cancer. Potential benefits to testing include informed decisions regarding cancer screening and reduced incidence of late-stage cancer following increased surveillance in at-risk individuals.²³ Despite the potential medical benefits, genetic testing also has the potential of adverse events, such as increased anxiety or depression, negative effect on family relationships, and loss of privacy and genetic discrimination.²⁴

Since the advent of genetic testing, research has shown that an individual's intention to pursue testing significantly overestimates the actual uptake of testing when offered.^{25,26} To date, 1 American study²⁷ and 1 Finnish study²⁸ have explored interest in genetic testing within families known to have HNPCC mutations. The American study found that a smaller proportion of family members with HNPCC (43% of family members eligible, 60% of those participating in the study) was likely to pursue genetic testing and receive results. Suggested barriers to test acceptance included higher formal education levels and the presence of depressive symptoms, especially in women. In the Finnish study, uptake of testing was higher, with 75% of eligible subjects choosing testing. Logistic regression analysis in the Finnish study identified employment status as the only significant factor predicting test acceptance; those employed were significantly more likely than others to choose genetic testing and receive the test results.

To further elucidate factors affecting decisions regarding genetic testing, we evaluated perceptions, intentions, attitudes, and uptake of genetic testing at baseline in individuals from families with newly identified HNPCC.



Flow diagram of study procedures. HNPCC indicates hereditary nonpolyposis colorectal cancer.

METHODS

STUDY POPULATION

The subjects were members of a cohort study conducted at the National Institutes of Health between April 15, 1996, and November 20, 1999. One hundred sixty-five adult men and women from 15 families identified with HNPCC mutations were considered eligible. Overall, 104 men and women chose to participate. Participants included individuals with HNPCC-associated cancers demonstrating microsatellite instability or a family history suggestive of HNPCC (index cases, $n=47$) and first-degree relatives at 50% risk of inheriting the family mutation ($n=57$).

PROCEDURES

The study was approved by the institutional review boards at the National Human Genome Research Institute and the National Naval Medical Center. A flow diagram of the study is depicted in the **Figure**. Probands were identified through cancer clinics at the National Cancer Institute and the National Naval Medical Center through the collection of family medical histories. Probands were also referred from unselected local and regional health care providers throughout the United States who became aware of the research protocol. Individuals with colon cancer identified through these means initially gave informed consent for the purpose of collecting a family medical history and to obtain tumor blocks for assessment of microsatellite instability.

GENETIC EDUCATION AND COUNSELING

Through telephone contact, those individuals meeting selection criteria were offered participation in the education and counseling portions of the study. Individuals whose families were previously known to have HNPCC mutations through other research or clinical efforts and are at 50% risk of inheriting the mutation were also eligible to participate. However, no participants were referred through these criteria or included in the data analyzed for this study.

An individual's decision to participate in the education and counseling portion of the study included the agreement to com-

plete a baseline questionnaire, receive genetic education and counseling pertaining to HNPCC, and participate in telephone interviews at 6 and 12 months after the intervention. All persons were reminded that participating in the education and counseling sessions did not obligate them to undergo genetic testing. Education and counseling sessions were conducted at the National Naval Medical Center, with partial reimbursement for the participant's travel expenses. Following consent to the education and counseling portion of the study, participants provided information pertaining to their demographics, knowledge, awareness, expectations, intentions, mood, attitudes, perceived risk, cancer worries, family relationships, spirituality, coping, and health beliefs through a baseline questionnaire. Participants were then provided with a standardized (scripted) genetic education session accompanied by slides. Depending on each participant's preference and consent, we provided an individual or a single-family education session. The sessions were presented by a board-certified genetic counselor (n=1; D.W.H.) or a cancer research nurse (n=2; J.J. or E.D.). Topics covered at the education sessions included (1) basic facts about the incidences of cancer and colon cancer; (2) risk factors for cancer; (3) inheritance of cancer susceptibility in families with HNPCC; (4) possible outcomes of genetic testing for HNPCC; (5) potential benefits, limitations, risks, and psychological or relational effects associated with genetic susceptibility testing; and (6) a review of cancer surveillance and screening guidelines recommended for families with HNPCC.²⁹ Information regarding surveillance and screening for uterine and ovarian cancer in women within these families was also provided, along with risk reduction options of prophylactic colectomy and hysterectomy. However, the lack of data about the efficacy of prophylactic surgery was noted. The potential benefit of early detection was discussed, including the potential of increasing life expectancy by the identification of premalignant or early lesions in mutation carriers.²² All participants were given a pamphlet that reviewed the information provided during the education session.

OFFER OF GENETIC TESTING

Following the education session, participants were provided a client-centered counseling session to consider the implications of genetic testing for HNPCC. All counseling sessions were undertaken individually to facilitate independent decisions regarding the option of genetic testing. The counseling sessions encouraged the participant to personally assess expectations of test results, review implications of test results on cancer risks, discuss plans regarding communication within the family, and identify motivating factors for or against testing. Following the counseling session, participants were presented with the options regarding genetic testing. Participants were offered the options of (1) taking additional time to further consider the option of testing, (2) not undergoing genetic testing at that time, or (3) undergoing phlebotomy for genetic testing. Those individuals who chose to be tested underwent an additional informed consent process focusing on the potential benefits, risks, limitations, and social and psychological issues pertaining to genetic testing. They then had a blood sample collected by an oncology nurse (J.J. or E.D.), which was sent for testing at a Clinical Laboratory Improvement Act–approved facility. The blood samples were labeled only with the participant's code number; no other identifying information was provided.

MUTATIONAL ANALYSIS

At the approved molecular diagnostics laboratories, blood samples provided by the initially affected individuals in each family (probands) had whole-gene sequencing of the 2 most

common HNPCC-associated genes, *MSH2* and *MLH1*. The individual exons (16 of *MSH2* and 19 of *MLH1*) of these genes were amplified using polymerase chain reaction, followed by direct DNA sequencing. The mutations were identified by forward and reverse sequencing and were confirmed by the allele-specific oligodeoxy-nucleotide (probes) or restriction fragment length polymorphism (patterns) polymerase chain reaction method. Individuals opting for genetic testing within families with known HNPCC mutations had directed mutational analysis completed. Payment for the gene sequencing was provided through the research budget.

PROVISION OF GENETIC TEST RESULTS

When results became available, the participants were notified by telephone. Those interested in receiving results were offered an appointment for a return visit to receive and discuss the results in person. Participants were also given the option of deferring the receipt of their results. All participants were encouraged to bring a support person with them to the results session. The participant, his or her designated support person, a clinical oncologist (I.K.), a board-certified genetics counselor (D.W.H.), and an oncology nurse (J.J. or E.D.) attended the results sessions. Results were provided within a standardized format and included the following topics: (1) thoughts and concerns experienced since the decision to pursue genetic testing; (2) expectations about the test results; (3) the genetic test results; (4) risk estimates for the cancers associated with HNPCC; (5) a review of cancer surveillance and screening guidelines; (6) plans and approaches for sharing genetic test results with others (family, friends, and health care providers); and (7) supportive counseling. Results sessions typically lasted 1 hour, with no other data collection occurring during that visit. The oncology nurse telephoned each participant 2 weeks after the results session to address questions since the last contact and to provide support or referral as appropriate. A follow-up letter was provided that summarized the results session.

RECRUITMENT OF FAMILY MEMBERS

In those individuals in whom HNPCC mutations were identified, participation in the education and counseling session was offered to relatives at 50% risk of inheriting the mutation. In cases in which the first-degree relative was deceased, the offer to participate was extended to second-degree adult relatives. The contacting of relatives and an offer to participate in the study could be accomplished in 3 ways. The options included (1) personal contacting of relatives; (2) providing the relative with a letter informing them of the identification of an HNPCC mutation within the family, with or without identification of the relative; or (3) our contacting the relative via telephone following notice by the participating relative. In all cases, participants chose to personally contact eligible relatives to inform them of the study.

MEASURES

All demographic and predictor variables were assessed through the baseline questionnaire before the education and counseling sessions and the offer of genetic testing. A broad array of key independent and dependent variables was elicited. Selected variables relevant to this analysis are listed in this subsection. The questions assessing awareness of genetic testing, risk perception, intentions regarding genetic testing, and pros and cons of genetic testing were adapted from previous research^{29,30,34} in individuals and families considering genetic testing for hereditary breast and ovarian cancer. In addition, all tools used in this study were included as part of a core set of instru-

Table 1. Study Participants (n = 104)*

Characteristic	Value
Median (range) age, y	43 (18-83)
Ethnicity	
White	87
African American	7
Hispanic	3
Asian American	2
Native American	1
Sex	
Female	57
Male	43
Annual household income, \$	
≥50 000	52
<50 000	48
Religious preference	
Protestant	51
Catholic	31
Jewish	3
Other	7
None	9
Cancer history	
Personal history of cancer	62
No cancer history	38

*Data are given as percentage unless otherwise indicated. Some percentages do not sum to 100 because of rounding.

ments for a consortium of genetic testing projects funded by the National Institutes of Health (Cancer Genetics Studies Consortium of the Ethical, Legal and Social Implications Program of the National Human Genome Research Institute).

Sociodemographics

Sex, age, marital status, employment status, income level, religious background, health insurance status, number of relatives with cancer (and degree of relatedness), and personal history of cancer were assessed through the questionnaire.

Awareness of Genetic Testing

A series of 4 Likert-style questions assessing awareness of genetic testing, in general, and cancer genetic testing for hereditary forms of breast and colon cancers was used.³⁰ Participants were asked, "How much have you read or heard about genetic testing" for (1) inherited disease, (2) cancer, (3) breast cancer, and (4) colon cancer? Possible responses to each of the questions were (1) almost nothing, (2) relatively little, (3) a fair amount, or (4) a lot.

Risk Perception

Participants' perceptions of having an altered gene associated with HNPCC were evaluated through their response to a 4-item, Likert-style question adapted from previous research.³¹ Participants were asked, "In your opinion, how likely is it that you have an altered HNPCC gene?" Possible responses to the question included (1) not at all likely, (2) somewhat likely, (3) very likely, or (4) definitely.

Intentions Toward Genetic Testing for HNPCC

Participants' intentions toward genetic testing for HNPCC were evaluated through their response to a single, Likert-style question adapted from previous research.^{24,32,33} Participants were

asked, "Which of the following statements best describes the way you are feeling right now?" Possible responses to this question included (1) I definitely do not want to be tested for HNPCC genes, (2) I probably do not want to be tested for HNPCC genes, (3) I probably want to be tested for HNPCC genes, or (4) I definitely want to be tested for HNPCC genes.

Pros and Cons of Genetic Testing

A series of 14 Likert-style items adapted from previous research^{33,34} was used to assess perceptions of the benefits, limitations, and risks of genetic testing. Participants read a list of benefits (7 items) and limitations and risks (7 items) of gene testing for HNPCC and were asked to rate the level of importance (not at all important, somewhat important, or very important). The 2 scales have been validated in previous research by Lerman and colleagues^{30,31,33} (Cronbach α coefficients, 0.73 [7 pro items] and 0.85 [7 con items]). In addition, the participants were asked to choose the single most important benefit and limitation or risk of genetic testing from the lists.

STATISTICAL ANALYSIS OF DATA

In general, data were analyzed via 2-dimensional contingency tables. When both variables represented unordered categories, the χ^2 test was used to assess the statistical significance of the association between 2 factors being evaluated. When any of the data in a table reflected ordered categories, a Cochran-Armitage test for trend, Kruskal-Wallis test for categorical data, or Jonckheere-Terpstra test for trend was used, as appropriate, according to whether 1 or both factors were ordered and the number of categories. Continuously measured factors were compared between 2 groups using the Wilcoxon rank sum test. All categorical analyses were performed with an exact procedure; thus, the reported P values, which are all 2-tailed results, are correct, even for sparse 2-way tables. This analysis was done in an exploratory fashion, with many associations investigated that are not reported. In view of the large number of statistical tests performed, only $P < .01$ should be interpreted as possibly being statistically significant, while P values between .01 and .05 would indicate a strong trend.

RESULTS

CHARACTERISTICS OF STUDY SAMPLE

Table 1 gives the characteristics of participating individuals. Fifty-seven percent of the participants completing the baseline questionnaire were female and 43% were male. Eighty-seven percent of the study sample were white, 7% African American, 3% Hispanic, 2% Asian American, and 1% Native American. The median age of participants was 43 years (range, 18-83 years); 50% of participants were between 34 and 52. Fifty-one percent reported themselves to be Protestant, 31% Catholic, 3% Jewish, 7% following another religion (not listed), and 9% had no religious affiliation. Forty-eight percent had an annual family income less than \$50 000. Sixty-two percent reported a medical history of cancer. Forty-five (96%) of 47 probands had colon cancer; the remaining 2 (4%) had HNPCC-associated cancers. Seventeen (30%) of 57 family members had experienced cancer before their participation in the study; 13 had experienced colon cancer and 7 had experienced multiple primary HNPCC cancers. Other cancers experienced by family members

included uterine (4 patients), ovarian (2 patients), cervical (2 patients), brain (1 patient), pituitary (1 patient), skin (2 patients), prostate (1 patient), and breast (1 patient).

BASELINE QUESTIONNAIRE

Of the 165 persons identified as eligible to participate, including index cases and first-degree relatives, 104 (63%) elected to participate. Because the identification of families in this study began with an affected individual (index case or proband) within a family suspected of having HNPCC mutations, we sorted probands from eligible family members. This identified 54 probands who were eligible to participate; 87% chose to participate, completed a baseline questionnaire, and received education and counseling. Of the 111 family members eligible to participate in the study, 51% chose to participate by completing the questionnaire and receiving education and counseling.

AWARENESS OF GENETIC TESTING

Sixty-five percent of participants had read or heard almost nothing or relatively little regarding genetic testing for cancer before their participation in this study. Likewise, 64% reported that they had read or heard almost nothing or relatively little regarding genetic testing for colon cancer.

In performing a cross tabulation of their awareness of genetic testing with other demographic variables, a statistically significant association was identified between participants' awareness of genetic testing and their household income. Specifically, those at higher household income levels were more aware of genetic testing for cancer ($P = .001$) and colon cancer ($P = .009$) than those at lower household income levels (**Table 2**).

No significant associations were found between participants' awareness of genetic testing and age, sex, intentions regarding genetic testing, personal cancer history, or number of first-degree relatives with cancer.

PERCEIVED RISK OF BEING A CARRIER FOR AN HNPCC MUTATION

Before education, counseling, and the offer of genetic testing, 72% of the participants thought that it was very likely (62%) or definite (10%) that they carried an HNPCC mutation. Twenty-five percent believed it was somewhat likely that they carried an HNPCC mutation, and 3% thought it was not at all likely. This study identified that participants' feelings about their chances of getting colon cancer are significantly associated with their beliefs about the likelihood that they carry a mutation ($P < .001$) (Table 2). In other words, those believing that they have a greater chance of getting colon cancer are also more likely to believe that they carry a mutation. Furthermore, participants' beliefs about whether they carry a mutation in a gene associated with HNPCC have a statistically significant association with their intention to pursue genetic testing ($P = .001$); those believing that they carry a mutation have greater intention to pursue genetic test-

ing. Cancer status (having had cancer vs no personal history of cancer) demonstrated a positive association ($P = .005$) with their beliefs about carrying an HNPCC mutation, with 50 (79%) of 63 of those who had cancer believing that they definitely ($n = 10$) or very likely ($n = 40$) carry a mutation, in contrast to 25 (61%) of 41 of those without cancer who definitely ($n = 0$) or very likely ($n = 25$) believed that way. In other words, those individuals who had already experienced cancer were significantly more likely to have a perceived risk of carrying an HNPCC mutation than individuals without cancer.

INTENTIONS TOWARD GENETIC TESTING

Ninety-seven percent of participants stated before education and counseling that they probably (28%) or definitely (69%) wanted genetic testing, while 3% probably or definitely did not want testing. The intention to pursue genetic testing was found to have a positive association with participants' beliefs that cancer may be explained by family heredity ($P = .006$) (Table 2). Furthermore, participants' concern about the psychosocial effect of genetic testing on the family demonstrated a negative association with their intention to pursue testing ($P = .001$). In addition, participants' concerns about their ability to handle the emotional aspects of genetic test results demonstrated a negative association with their intentions to pursue testing ($P < .001$). There was no association found between the participants' age, sex, or cancer status in regard to their intentions toward genetic testing.

REASONS FOR PURSUING GENETIC TESTING FOR HNPCC

One half of those responding believed that the most important reason for undergoing genetic testing was to learn about their children's risk; the second most important reason (17%) was to guide cancer screening; and third in importance (13%) was to confirm their belief that they carry a mutation.

With respect to the importance of genetic testing for reproductive decision making, a statistically significant difference was detected between those younger than the median age of 43 years vs older (dichotomized age, $P = .002$). Younger participants placed increased importance on using genetic testing for reproductive decision making, with 24 (51%) of 47 participants younger than the median age reporting genetic testing as very important ($n = 6$) or somewhat important ($n = 18$), in contrast to 11 (22%) of 51 participants older than the median age of 43 years who thought that it was very important ($n = 1$) or somewhat important ($n = 10$). This association was confirmed through the analysis of age as a continuous variable ($P = .001$). Furthermore, we identified a trend for male participants to place greater importance than female participants ($P = .02$) on using genetic testing "to make decisions about having (more) children." Twenty-two (50%) of 44 men believed that genetic testing was very important ($n = 4$) or somewhat important ($n = 18$) for reproductive decision making, in contrast to 13 (24%) of 54 women who thought that it was very important ($n = 3$) or somewhat important ($n = 10$).

Table 2. Selected Associations*

Awareness of Genetic Testing for Cancer						
Annual Household Income, \$	Awareness of Genetic Testing for Cancer				Total	P Value
	Almost Nothing	Relatively Little	A Fair Amount	A Lot		
<20 000	3	5	1	0	9	.001
20 000-35 000	5	14	3	0	22	
>35 000-50 000	5	10	4	0	19	
>50 000-75 000	8	6	9	4	27	
>75 000	1	11	12	1	25	
Total	22	46	29	5	102	

Awareness of Genetic Testing for Colon Cancer						
Annual Household Income, \$	Awareness of Genetic Testing for Colon Cancer				Total	P Value
	Almost Nothing	Relatively Little	A Fair Amount	A Lot		
<20 000	4	4	0	1	9	.009
20 000-35 000	6	8	7	1	22	
>35 000-50 000	3	13	3	0	19	
>50 000-75 000	6	10	9	2	27	
>75 000	2	10	10	3	25	
Total	21	45	29	7	102	

Beliefs About Likelihood of Carrying Mutation						
Risk Perception Regarding Colon Cancer	Beliefs About Likelihood of Carrying Mutation				Total	P Value
	Not at All Likely	Somewhat Likely	Very Likely	Definitely		
Much less	0	4	5	2	11	<.001
A little less	1	2	4	0	7	
About the same	0	9	7	1	17	
A little more	1	10	19	0	30	
Much more	0	1	30	7	38	
Total	2	26	65	10	103	

Beliefs About Likelihood of Carrying Mutation						
Intent to Pursue Genetic Testing	Beliefs About Likelihood of Carrying Mutation				Total	P Value
	Not at All Likely	Somewhat Likely	Very Likely	Definitely		
Definitely do not	0	1	0	0	1	.001
Probably do not	1	1	0	0	2	
Probably want	0	13	14	2	29	
Definitely want	2	11	51	8	72	
Total	3	26	65	10	104	

Cancer Is Explained by Family Heredity						
Intent to Pursue Genetic Testing	Cancer Is Explained by Family Heredity				Total	P Value
	Strongly Disagree	Somewhat Disagree	Somewhat Agree	Strongly Agree		
Definitely do not	0	0	1	0	1	.006
Probably do not	1	0	1	0	2	
Probably want	1	2	17	9	29	
Definitely want	0	1	34	37	72	
Total	2	3	53	46	104	

Effect of Genetic Testing on Family						
Intent to Pursue Genetic Testing	Effect of Genetic Testing on Family			Total	P Value	
	Not at All Important	Somewhat Important	Very Important			
Definitely do not	0	0	0	0	.001	
Probably do not	0	0	1	1		
Probably want	1	6	14	21		
Definitely want	9	32	15	56		
Total	10	38	30	78		

(continued)

Table 2. Selected Associations* (cont)

Intent to Pursue Genetic Testing	Ability to Handle It Emotionally			Total	P Value
	Not at All Important	Somewhat Important	Very Important		
Definitely do not	0	0	1	1	<.001†
Probably do not	0	1	1	2	
Probably want	2	15	10	27	
Definitely want	23	36	10	69	
Total	25	52	22	99	

*Data are given as number of subjects. Comparisons are by Jonckheere-Terpstra test unless otherwise indicated.
†Wilcoxon rank sum.

REASONS FOR NOT PURSUING GENETIC TESTING

The greatest concerns about genetic testing included worries about losing health insurance (39%), concerns about how it might affect the family (27%), and concerns about handling the results emotionally (10%).

A statistically significant difference ($P=.006$) was detected between those younger than the median age of 43 years (dichotomized age) compared with those participants older than 43 years with respect to concerns about handling the emotional aspects of genetic testing. Forty-two (86%) of 49 who were younger than the median age believed that their ability to handle the emotional effect of genetic testing was very important ($n=15$) or somewhat important ($n=27$), in contrast to 32 (64%) of 50 who were older than the median age who thought that the emotional effect of genetic testing on them was very important ($n=7$) or somewhat important ($n=25$) regarding their decision to pursue testing. Younger participants have greater reported concerns about their ability to handle the emotional effect of testing than older participants. Male participants tended to identify "I am concerned about the effect it (genetic testing) would have on my family" as an important issue more often than female participants ($P=.03$). Thirty-eight (97%) of 39 men reported that the effect on family members was very important ($n=18$) or somewhat important ($n=20$), in contrast to 30 (77%) of 39 women who believed that it was very important ($n=12$) or somewhat important ($n=18$).

TESTING DECISIONS

Table 3 gives the participation rates and uptake of genetic testing. Following education, counseling, and informed consent, 81% (44/54) of eligible probands eventually chose to undergo genetic testing for HNPCC. Nearly 51% (56/111) of eligible first-degree relatives chose to pursue genetic testing.

A significant proportion of those consenting to participate in the study chose to pursue genetic testing (Table 3), with 94% (44/47) of probands choosing to participate in the study eventually choosing to pursue genetic testing. Likewise, 98% (56/57) of family members, once consenting to participate in the study, chose to pursue genetic testing. Among the family members tested, 59% (33/56) received information indicating that they had

Table 3. Participation Rates and Uptake of Genetic Testing*

	Probands	Family Members
Participation		
Eligible	54	111
Choosing to participate	47	57
Participating, %	87	51
Uptake of genetic testing		
Choosing genetic testing	44	56
Declining testing	3	1
Eligible choosing testing, %	81	51
Participants choosing testing, %	94	98

*Data are given as number of subjects unless otherwise indicated.

HNPCC-associated mutations, and 41% (23/56) learned that they did not carry the family mutation.

COMMENT

In interpreting the results of this study, the limitations should be considered. First, participation in the study was voluntary and required considerable commitment on the part of participants, including travel to and from the study site and often an overnight stay. To enlist a broad group of participants and to remove cost as a factor affecting participation and testing, most of the travel costs were covered for those choosing to participate. However, because of the time commitment and travel away from home and family, we believe that our study attracted those individuals who had previous intentions toward pursuing genetic testing and, therefore, may not represent a general sample of family members at 50% risk of inheriting mutations.

Second, genetic education, counseling, and testing were offered free of charge to participants. This fact, in combination with participants' often stated perception that information and test results obtained within this study were less accessible to health insurers than if they pursued testing privately, seemingly lessened their concerns regarding insurance-related risks. These 2 issues may have resulted in certain selection biases, and we suspect that uptake rates of genetic testing within this study may overestimate those encountered in a clinical fee-for-service arena.

Third, because these were newly identified families and not part of an existing registry, the offer of par-

Table 4. Selected Characteristics of First-Degree Relatives*

Characteristic	Participants (n = 57)	Nonparticipants (n = 54)
Median (range), y	39 (18-83)	40 (18-78)
Sex		
Female	30 (53)	34 (63)
Male	27 (47)	21 (39)
Cancer status		
History of cancer	17 (30)	17 (31)
No history of cancer	40 (70)	38 (70)
No. of relatives with cancer		
First-degree	134	91†
Median (range)	2 (0-8)	1 (0-6)
Second degree	146	144
Median (range)	2 (0-7)	2 (0-9)
Total first- and second-degree	280	235

*Data are given as number (percentage) unless otherwise indicated.
†Significant at $P = .007$.

ticipation within families was dependent on participants' willingness to share information with other family members. Although this may mimic the diffusion of new information within families with newly identified disease-susceptible mutations, it does not provide an all-inclusive view of individuals who otherwise have reservations or concerns about genetic counseling and testing. Although we were aware of the number of eligible family members through the analysis of the medical family histories, we cannot be certain that all eligible family members were contacted and offered information about the study. Therefore, our comments provide insight primarily on those agreeing to participate in education and counseling sessions. Despite the noted limitations, we were able to analyze a few characteristics of first-degree relatives who did not participate, including sex, cancer status, and number of relatives experiencing cancer (Table 4). An association was identified between the number of first-degree relatives who had experienced cancer and participation in the study ($P = .007$), with participants having a median of 2 first-degree relatives who had experienced cancer, compared with a median of 1 first-degree relative with a history of cancer for nonparticipants. The number of second-degree relatives who had experienced cancer did not demonstrate an association. No significant association was identified between participating and nonparticipating first-degree relatives based on sex ($P = .34$), age ($P = .37$), or cancer status ($P = 1.0$).

Despite the suggested limitations, we observed an uptake of genetic testing by 51% of first-degree relatives of known mutation carriers. The uptake of genetic testing by family members was slightly higher in this study than that reported in a previous US study²⁷ (43%) investigating the uptake of genetic testing in families with documented mutations in genes associated with HNPCC. However, the difference in the uptake of testing between this study and the previous study does not represent a statistically significant difference ($P = .24$), with 56 (50%) of 111 undergoing genetic testing in this study and 90 (43%) of 208 undergoing genetic testing in the other study. In considering the Finnish study,²⁸ which also investigated the uptake of genetic testing in families with

known HNPCC mutations, we noted a significant difference in the uptake compared with our experience ($P < .001$), 51% uptake of testing in this study vs 75% (334/446) undergoing testing in the Finnish study. The difference between the uptake of testing in the United States and Finland was suggested to be primarily because of the basic differences between health care systems, with the US health care system relying on private insurance.²⁸ This is supported by noting that concerns about insurance as a reason to not undergo genetic testing were almost absent (<2%) among participants in the Finnish study. The authors of the Finnish study suggested that Finnish citizens have a greater level of trust in their health care providers and system, which they predict increases the uptake of genetic testing.

The general lack of awareness of the availability of genetic testing for colon cancer by participants before their enrollment in the study suggests that continued efforts are necessary to inform the general public and families suspected of having hereditary forms of colon cancer. Research efforts are needed to identify the most effective approaches to educate and disseminate information to the general public and to families at increased risk.³⁵ Furthermore, the significant difference identified through this study between the awareness of genetic testing by individuals of lower vs higher socioeconomic means suggests that concerted efforts should focus on research to determine effective methods and strategies for providing information to individuals with limited economic resources.

Our findings demonstrate that participants who believe that they have a greater chance of getting colon cancer are more likely to believe that they carry a mutation. In addition, those participants who believe that they carry a mutation have greater intention to pursue genetic testing. Based on this line of evidence, it may be conversely hypothesized that some individuals who do not choose to participate in genetic counseling have a lower perceived risk of getting cancer, which lessens interest (intentions) toward genetic counseling and testing, despite their a priori 50% risk of carrying an HNPCC mutation. Previous research²⁸ suggests that nonparticipants appear to decline genetic testing and cancer surveillance. However, it is possible that a proportion of those not participating are appropriately following cancer-screening guidelines but not choosing to pursue genetic testing. To understand factors that affect perceived risk and the potential effect on compliance with cancer-screening recommendations for persons at risk in families with HNPCC mutations, additional research would be informative in developing education, counseling, and screening programs for cancer genetics programs.

The data from this study indicate that younger participants are concerned about their ability to handle the emotional outcomes of genetic testing, fear the potential effect on family members, and place increased importance on genetic testing for reproductive decision-making purposes. Concerns about the effect on self or family that were reported in the questionnaire were generally not vocalized within the informed consent session. This may suggest uneasiness on the part of participants to express these concerns. These findings support

the inclusion of discussions between the health care providers and patients, particularly younger persons, regarding the potential effect of genetic testing on themselves, their family, relationships, and reproductive decision making. Health care professionals may assist younger individuals and couples by acknowledging the complex personal and social challenges such information presents and by encouraging referral to genetics professionals for additional counseling to facilitate informed (factual and psychological) decisions pertaining to these issues. Depression has been identified as a barrier to individuals' pursuing genetic counseling services.²⁷ Therefore, discussions with individuals in families suspected of having inherited forms of cancer should include this as a potential factor complicating informed decision making. Some individuals may be offered genetic counseling and the option of testing at a time that is difficult practically or emotionally. Therefore, periodic discussions about cancer risks and adherence to recommended cancer-screening guidelines may benefit the individual at increased risk of cancer and aid the health care practitioner in providing optimal care.

In considering participants' reasons to not pursue genetic testing, the most common concern was regarding the potential negative effect on their insurability. This concern was expressed through the baseline questionnaire (allowing quantitative analysis) and verbally by consenting participants. Before participation, the issue of potential risks to insurability was routinely and straightforwardly addressed in each informed consent session. Acknowledgment of the potential was accompanied by a brief discussion of the status of state and federal laws attempting to protect individuals from genetic discrimination. We acknowledged that such discrimination could have significant medical and financial consequences; however, we further noted that few cases of genetic discrimination had been reported to date. This concern may have been mitigated by the facts that this protocol (1) holds a Certificate of Confidentiality through the National Cancer Institute, (2) does not require participants' reimbursement for testing (through insurance or personal funds), and (3) uses numeric codes to anonymize samples sent to the Clinical Laboratory Improvement Act-approved facility that performed the testing. We acknowledged that these safeguards could not guarantee confidentiality but would reduce the chances of information getting back to insurers or employers. Nevertheless, most participants at the time of informed consent focused the discussion about potential risks of participating and genetic testing on the potential of insurance companies' obtaining information that could place themselves or their family members at risk for discrimination. The concern about genetic testing and its potential effect on insurability is foremost in participants' minds. Given such a high degree of concern among participants about the issues related to insurability, we hypothesize that this may also be a significant factor affecting participation among relatives who chose not to participate in the study. This hypothesis is supported by recent data³⁶ from individuals who declined genetic counseling services for hereditary breast and ovarian cancer after referral. In that study, 41% identified their concern about health insurability for

themselves and other family members as the most significant factor affecting their decision not to pursue genetic counseling services. The concern regarding discrimination may be extended, hypothetically, to individuals' efforts to obtain cancer screening, thereby increasing the number of individuals ultimately experiencing greater medical and psychological burden associated with the diagnosis of more advanced cancers.

A recent article³⁷ suggests that evidence for widespread discrimination by insurance companies is lacking and, therefore, may not warrant the level of concern expressed. However, until the public is reassured by legislation or additional evidence that discrimination is not occurring, the public's concern may override the uptake and use of information and testing to guide cancer screening. National and individual state legislation protecting individuals from genetic discrimination by health insurers may reassure patients and their families that they will not be harmed in seeking genetic counseling and testing services. Likewise, increased coverage of genetics services by third-party payers may improve patient access to services.

CONCLUSIONS

Our results suggest that the uptake of genetic testing for HNPCC among members of high-risk families may be lower than what was originally anticipated and more closely approaches those levels reported in a previous US study.²⁷ Nearly half of the individuals at 50% risk of inheriting an HNPCC mutation chose not to participate in the study. A clear understanding as to why such choices were made is unknown. Significant concerns are the potential that these individuals (1) do not perceive themselves at increased risk of cancer and therefore may not pursue cancer screening; (2) fear the potential consequences of genetic testing, eg, discrimination by insurers and employers, stigmatization, and the effect on themselves or family members; (3) are not aware of the availability of genetic testing; (4) experience financial or time constraints that limit their ability to pursue genetic counseling services; (5) perceive no benefit from genetic counseling services; or (6) are experiencing depression at a level that interferes with their potential to seek counseling and testing to clarify their risks and facilitate appropriate cancer screening.

These findings support the inclusion of psychological and psychosocial issues related to genetic testing as part of the informed consent process between health care providers and individuals considering genetic testing for HNPCC.

Future research is needed to identify more efficient means of reaching, educating, and counseling the general public, at-risk populations, and health care providers about medical genetics, genetic counseling, and testing. In particular, this research is relevant to understanding the extent to which lack of information, fear of insurance discrimination, and psychological or other barriers negatively affect cancer-screening behavior. However, until meaningful national legislative safeguards are established to address the concerns regarding insurance discrimination, we fear that a significant portion of persons will continue to live at increased risk, without the

benefit of information, counseling, and appropriate cancer screening to reduce the morbidity and mortality associated with cancer.

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38. Here are reasons some people give for wanting to be tested for HNPCC genes. Please indicate how important each reason is for you.

	Not at all Important	Somewhat Important	Very Important	Does Not Apply
a. To learn about my children's risk.				
b. To make a decision about surgery to have my colon or other organs removed before cancer arises.				
c. To know if I need to get cancer screening tests more often.				
d. To reduce uncertainty.				
e. To make decisions about having (more) children.				
f. To be reassured.				
g. Because you suspect you are a gene carrier.				

39. Which of these reasons would be your most important reason for wanting to be tested? (CHOOSE ONE)

- To learn about my children's risk
- To make a decision about surgery to have my colon or other organs removed
- To know if I need to get cancer screening tests more often
- To reduce uncertainty
- To make a decision about having (more) children
- To be reassured
- Because you suspect you are a gene carrier
- Other reason SPECIFY: _____

40. Here are reasons some people give for not wanting to be tested for HNPCC genes. Please indicate how important each reason is for you.

	Not at all Important	Somewhat Important	Very Important	Does Not Apply
a. I am concerned about the effect it would have on my family.				
b. I do not trust modern medicine.				
c. I believe that there is nothing that can be done to prevent getting cancer.				
d. I am concerned that I could not handle it emotionally.				
e. I am worried about losing my insurance.				
f. My chances of having an altered HNPCC gene are small.				

41. Which of these reasons would be your most important reason for not wanting to be tested for HNPCC genes? (CHOOSE ONE)

- Concern about the effect on your family
- Do not trust modern medicine
- Do not believe there is anything you can do to prevent getting cancer
- Concern about your handling it emotionally
- Worry about losing your insurance
- Chances of having altered gene are small
- Other reason SPECIFY: _____

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Genetic testing for Alpha₁-antitrypsin deficiency

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Purpose: The Alpha Coded Testing Study investigated the risks, benefits, and psychological impact of home genetic testing for α_1 -antitrypsin deficiency. **Methods:** In the study, 996 adult individuals requested and returned a home-administered, confidential, fingerstick blood test. **Results:** Individuals highly rated the benefits of establishing a diagnosis (82%), helping family members (86%), and anticipating peace of mind (79%). 78% of 239 current smokers reported a high likelihood of smoking cessation if diagnosed with AATD. After testing, more than 60% indicated that they would share the results with family and physicians but < 30% would share results with insurance companies. **Conclusions:** Confidential home testing for genetic disorders requires a comprehensive program of participant support. *Genet Med* 2004;6(4):204–210.

Key Words: α_1 -antitrypsin, genetic testing, smoking, genetic discrimination, confidentiality

α_1 -Antitrypsin deficiency (AATD [MIM 107400]) is a genetic condition that can lead to early onset emphysema and hepatic impairment in some individuals.¹ The gene that codes for α_1 -antitrypsin deficiency is located on chromosome 14 and more than 100 genotypic variants have been described. The serum levels of α_1 -antitrypsin are determined by each of 2 codominant genes. The most common phenotype in the US and world is PiMM (also called PiM) in which both AAT genes produce M protein. The two most common deficiency alleles S and Z produce proteins that are made in the liver but fail to fold properly for hepatic egress. At least one S or Z gene is found in 4% to 6% of the US population.²

Individuals with symptoms are usually severely deficient in AAT. The phenotypes associated with severe deficiency are most commonly PiZZ (also called PiZ) and PiSZ. PiZZ AATD is the most common cause of early onset emphysema with a mean age at time of pulmonary impairment at 35.³ Rarely, individuals with the carrier state PiMZ develop symptoms of lung or liver disease.⁴

Not all individuals with PiZZ AATD develop symptoms of lung and/or liver disease. In fact, there is substantial variability in the age of onset and severity of disease among PiZZ individuals even within families.⁵ Although some nonsmokers develop severe lung disease, the majority of pulmonary impairment occurs in ex-smokers or current cigarette smokers who develop emphysema. Therefore, identification of AATD allows

targeted intensive smoking cessation efforts. Intravenous augmentation therapy to restore serum AAT levels to higher levels remains costly,⁶ but has been shown to slow the course of emphysema in some patients.⁷

Because early onset severe emphysema is a devastating illness for affected individuals, interest in testing other family members for the Z or S allele has increased. Because of concerns about genetic discrimination in the absence of clinical disease and the lack of confidentiality of medical records, particularly from medical and life insurance entities, the AATD patient community has proactively supported a mechanism to provide confidential testing.

Despite potential benefits of genetic testing, some studies report hesitation and fear associated with genetic testing. Telephone interviews within the general population have suggested that the issues are complicated by a lack of understanding of the science and anxiety that is often unfocused.^{8,9} The possibility of genetic discrimination related to insurance has been cited as a major external factor concerning individuals considering genetic testing.^{10,11} A 1996 survey of 332 genetic support groups found that 40% of respondents had been asked about genetic diseases or disabilities on an application for health insurance. Of those 47% were denied coverage, and 25% were denied life insurance compared to 3% denied coverage in the general population.¹² The Health Insurance Portability and Accountability Act (HIPAA) prohibits certain uses of genetic information in determining insurance eligibility, but places no limits on rate setting.¹³ Uninsurability appears to be a valid concern for potential genetic test takers.

Other studies have shown public interest in genetic testing, especially among at risk populations.^{14–16} Eighty four percent of first-degree relatives of ovarian cancer patients who believed they were likely to be a gene carrier expressed interest in testing, compared to 63% of women who considered themselves unlikely to be carriers.¹⁷

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Research is emerging evaluating other factors that may influence interest in genetic testing. Some studies have found no influence of demographic factors on the decision to perform a genetic test.^{18,19} Cost and convenience may play a role in motivation for testing. In one study, over 90% of women considering testing for breast cancer susceptibility genes expressed interest in testing if it was free, but interest dropped to 60% if testing involved cost.²⁰ Among an HMO population, persons were more likely to undergo testing for cystic fibrosis if they could be tested upon initial approach with minimal effort.²¹

Although many studies have investigated interest in genetic testing, relatively few studies have evaluated specific beliefs about testing, such as anticipated risks and benefits. Furthermore, most studies examine beliefs a priori, without longitudinal follow-up after test results have been disclosed. The present study examines specific beliefs both before and after genetic testing for α_1 -antitrypsin deficiency (AATD), one of the most common yet misdiagnosed genetic disorders worldwide. The project was designed to provide a confidential, free, and convenient method of testing in order to reduce the potential confounding impact of these variables.

MATERIALS AND METHODS

Study design received input from individuals with AATD, physicians, psychologists, and genetic counselors knowledgeable about AATD. It was reviewed by the Ethical, Legal, and Social Implications (ELSI) Committee of the Alpha-1 Foundation, received approval from the Institutional Review Board for Human Subjects at the Medical University of South Carolina (MUSC), and carried a Certificate of Confidentiality from the Department of Health and Human Services. Patients signed written informed consent before testing.

The ACT Study utilized the infrastructure of the Alpha-1 Foundation, an organization founded by AATD-affected individuals to promote research. The study was advertised through a patient registry, regional meetings, and web site. Advertisements began in July 2001 followed by enrollment beginning in January 2002. Individuals requesting a test kit were mailed a study packet that included (1) informed consent, (2) a pretest questionnaire, (3) a fingerstick blood-spot test kit (Fig. 1), (4) a brochure discussing the testing procedure, and (5) a postage-paid preaddressed return envelope. Returned test kits were coded at MUSC and mailed to the University of Florida's Alpha-1 Genetics Laboratory. The blood spots were subjected to PCR analysis to determine if one or two copies of either the S or Z gene were present. Dried blood spot AAT concentration was matched to genotype. Results were returned to MUSC. Participants were then mailed a follow-up packet that consisted of (1) a letter detailing the results of the test, (2) a posttest questionnaire, and (3) a postage-paid preaddressed return envelope. In addition, all participants who tested either deficient for AATD (PiZZ or PiSZ) or were a carrier (PiMZ) received an informational support brochure addressing possible health concerns. Both deficient and carrier groups received an invitation to join the Alpha-1 Research Registry, and all participants

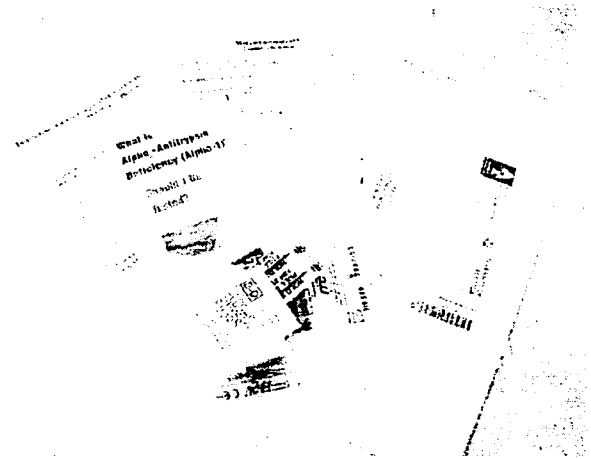


Fig. 1. Testing and educational materials used by the ACT study.

were offered free telephone support including consultation with a genetic counselor if desired.

Participants

Between January 2002 and February 2003, 3551 kits were requested. Individuals could request kits for themselves and/or other family members. Of these pretest packets, 1159 (33%) were returned with completed blood tests and pretest questionnaires. Some 163 individuals under the age of 18 years were excluded from pretest questionnaire analysis leaving a study cohort of 996 persons. After testing, these individuals were sent a posttest packet including questionnaire and statement of genotype status. To date, 700 (63% of the pretest sample) participants have returned the posttest questionnaire with 512 first time testers, age 18 or over included in posttest questionnaire analysis.

Survey materials

Pretest

Pretest questionnaires included items pertaining to demographics, smoking, reasons for seeking testing, and referral source. Six potential risks and six potential benefits were presented in a Likert format (1 = no risk or benefit, 5 = high risk or benefit). Other questions concerning beliefs (improved health, improved psychological well-being, increased health care costs, and expected discrimination) were presented in Likert format. Additionally, participants were asked to indicate the likelihood of a PiZZ child if both parents are PiMZ. Participants could choose among responses of 0%, 25% (correct answer), 50%, 75%, 100%, or "I don't know."

Posttest

The posttest questionnaire queried plans to divulge results of genetic testing to others. Additionally, participants were asked to rate anticipated effects from genetic testing. These effects included both potential benefits and harms.

Data Analysis

Demographic information is reported as a percent of the total response to each individual question. Likert scores of 4 and 5 were considered high. Values of $P < 0.05$ were considered significant for all analyses. Statistical analysis was performed using Stata (College Station, TX). Two sided t tests were used to compare age distributions between subgroups of participants. An overall median score for the risks and an overall median score for the benefits of testing at the pretest questionnaire were determined for those participants who answered all risk/benefit questions. The Wilcoxon signed rank test was used to compare the distribution of those median scores. The Wilcoxon rank sum test also was used to compare the distribution of responses to questions in Likert format between subgroups on the pretest questionnaire. The Chi-Square test for trend in binomial proportions was used to detect a trend in the proportion of correct answers to the genetic question with an ordered column variable. The Kruskal Wallis test and Wilcoxon rank sum test was used to compare the distribution of responses stratified by genotype to questions in Likert format on the posttest questionnaire. Fishers exact test was used to test for independence in 2 by 2 and 3 by 2 contingency tables.

RESULTS

The 996 participants averaged 42.4 ± 16.3 years (mean \pm SD) with a range of 18 to 82 years. Consistent with known demographics of AATD, 93% of participants were Caucasian. Sixty one percent of participants were female. A total of 84% of participants reported having health insurance, and 89% of participants report believing they will receive very important information about their genes reporting the highest score of 4 or 5 on the Likert scale. The primary referral source for recommending testing for AATD was family, and reasons that the test was suggested are reported in Table 1. Genetic testing results indicated 520 negative results of genotype MM(464), MS(53), or SS(3), 407 carrier results of genotype MZ, and 68 deficient results of SZ(27) or ZZ(40). Rare alleles requiring serum phenotyping (PiZNull) were suspected because of low protein concentrations and confirmed in 2 cases.

Perceived risks on pretest questionnaire

Less than 40% of participants anticipated a high risk of increased insurance premiums (Table 2). Other potential risks

included potential loss of insurance (31%), psychological risks (18%), and increased stress (13%). Younger participants were more likely to anticipate risks of losing health insurance ($r = 0.13, P < 0.001$), higher insurance premiums ($r = 0.14, P < 0.001$), losing employment ($r = 0.12, P < 0.001$), or psychological risks ($r = 0.16, P < 0.001$) than older participants. Education level was loosely correlated with anticipated risk of losing health insurance ($r = 0.08, P < 0.03$) and risk of higher insurance premiums ($r = 0.1, P < 0.004$). No other demographic variables predicted anticipated risks.

Perceived benefits on pretest questionnaire

In contrast to potential risks of genetic testing, participants anticipated significant benefits (Table 2). High scores were recorded for the effect of genetic knowledge on the family (86%) and for establishing a firm diagnosis (82%). Anticipated benefits did not significantly differ according to familial risk or demographic variables. Perceived benefits greatly outweighed perceived risks, as distribution of overall median scores for the six listed benefits of testing was greater than for the risks ($P < 0.0001$).

Confidentiality

Confidentiality was an important reason for testing through the ACT Study for the majority of participants with 61% rating its importance high. There was no difference in the rating of confidentiality depending on whether the test was recommended by a physician ($N = 73$), family member ($N = 586$), or other source(s). Confidentiality was more important to the participant if the test was suggested because a family member had AATD compared to those being tested because of symptoms ($P < 0.001$). Concern for confidentiality was inversely correlated with participant age ($P = 0.03$). Persons entering the test with high levels of concern about confidentiality were more likely to anticipate other risks than persons without this level of confidentiality concern (Table 3).

Smoking

Of 239 respondents who were current smokers (26% of the entire cohort), 78% report a high likelihood of quitting smoking if they were to be diagnosed with AATD. Current Smokers were younger (age 38 ± 14.5) than current nonsmokers (age 44 ± 16.6) ($P < 0.0001$). Thirty two percent of smokers did not have medical insurance compared to 11% of nonsmokers ($P < 0.0001$). Smoker's family members were more likely to have recommended testing as compared to nonsmokers ($P = 0.0003$). Smokers were more concerned about the risk of a change in the identity of their biological parent ($P = 0.03$) and more likely to rate availability of a drug treatment as an important benefit of establishing a diagnosis ($P = 0.048$).

Index participants

Index participants are those whose test was suggested because of liver or lung disease symptoms ($N = 136$). Index participants were older than non-Index participants ($N = 715$) with mean age 52 ± 15 versus 40 ± 16 , and were more likely to

Table 1
Reasons for testing in the Alpha-Coded Testing study

The test was suggested because:	N	%
I am at increased risk because of family members with disease	523	56.2
I have symptoms of lung or liver disease	103	11.1
I am getting a screening test	96	10.3
I am a partner of someone with Alpha-1	90	9.7
Combination of answers	118	12.7

Table 2
Risks and benefits of genetic testing

	N	Mean ^a	SD	% 4 or 5 ^b
Risks				
Losing health insurance	847	2.60	1.61	31
Higher health insurance	839	2.75	1.66	36
Losing your job	816	1.70	1.26	12
Psychological risks associated with genetic knowledge	818	2.22	1.37	18
Religious issues associated with genetic knowledge	809	1.28	.78	3
Knowledge concerning children's true parents	796	1.31	.90	5
Increased stress knowing that I have normal genes while a family member has abnormal AAT genes	816	1.90	1.31	13
Total	708	1.94	0.89	17
Benefits				
Establishing a diagnosis	882	4.34	1.11	82
Benefit of a drug treatment not available without a diagnosis	844	3.89	1.41	67
Genetic knowledge that may be helpful for family members	882	4.47	1.00	86
Networking with others who have the genetic condition	849	3.38	1.49	49
Screening for other manifestations of Alpha-1	847	3.87	1.35	65
Peace of mind if the genetic test is normal	876	4.28	1.19	79
Total	708	4.03	0.97	71

^a Rated on a 5-point Likert scale (1 = low risk or benefit, 5 = high risk or benefit)^b Percent who selected the highest score of 4 or 5 on the Likert scale**Table 3**Participants who selected confidentiality as a very important (Likert scale 4–5) reason for testing ($N = 577$) were compared to those in which confidentiality was scored 0–3 ($N = 378$).

Potential risk	Odds ratio	95% Confidence interval
Losing health insurance	3.4	2.4,4.8
Higher health insurance	3.1	2.2,4.3
Losing your job	2.9	1.7,5.1
Psychological risks associated with genetic knowledge	1.7	1.1,2.5
Religious issues associated with genetic knowledge	1.8	0.7,5.8
Knowledge concerning children's true parents	2.0	0.9,4.7
Increased stress knowing that I have normal genes while a family member has abnormal AAT genes	1.4	0.9,2.3

Odds ratios for rating other potential risks associated with genetic testing very important are listed.

be recommended by a physician only ($P < 0.0001$). Index participants rated the benefit of establishing a diagnosis and the availability of a drug treatment with a diagnosis significantly more important than non-Index participants ($P < 0.04$). Non-Index participants are more concerned about confidentiality, losing health insurance, higher health insurance premiums, and psychological risks associated with genetic testing ($P < 0.05$).

Knowledge of α_1 genetics

Each participant was asked to answer a question about the probability of having a child with PiZZ α_1 if both parents have PiMZ. Overall 39% of participants answered the question cor-

rectly, 28% answered incorrectly, and 33% responded, "I don't know." The question was answered correctly by more participants who performed research on AATD before participating (OR 3.14 [95% CI 2.35, 4.22]) and by participants recommended for testing by a physician (OR 1.65 [95% CI 1.00, 2.75]). Participants who answered the question correctly were more educated and younger than participants who answered the question incorrectly ($P < 0.0001$ and $P < 0.01$).

Posttest questionnaire

The subset of respondents who returned the posttest questionnaire ($n = 512$) did not significantly differ from the 404

respondents who only completed the pretest with regard to race, gender, or age. Participants returning a posttest questionnaire were more educated than participants who did not return a posttest questionnaire ($P = 0.005$). We compared responses to the post-test questionnaire between participants receiving a first time negative (PiMM, PiMS, $N = 277$), carrier (PiMZ, $N = 203$), and deficient (PiZZ or PiSZ, $N = 32$) test result. Table 4 demonstrates willingness to disclose test results with others.

Results of subgroup responses to 10 questions about participant expectations of events or feelings after the test result are shown in Table 5. Carriers and participants with a deficient test result were more likely to anticipate depression and anxiety compared to participants with a negative result ($P \leq 0.0001$). Of interest is that persons with deficient test results feel they rate the expectation of "improved health" more likely than persons with a carriers or negative test result.

DISCUSSION

The technology to provide home testing for genetic diseases remains in its infancy. In general, few participants reported difficulty with the use of home lancets to produce blood spots and most comments received at the coordinating center were highly favorable about the testing program. The high rate of nonreturned kits likely represents the combined effects of fear concerning the fingerstick, family members ordering test kits for other family members not interested in testing, anxieties concerning the testing process, and inertia related to the paperwork associated with testing. Technical aspects of the home test kit have been described elsewhere.²²

We were surprised that the majority of participants dismissed most of the proposed risks of testing and reported that risks were not in general discussed with family members. The greatest benefits were anticipated for the helpful effect of ge-

netic information for the family and for establishing firm diagnoses that assist with life planning.

Confidentiality was an important reason for testing through the ACT study. Persons most concerned about confidentiality were more likely to be concerned about risks associated with testing. Results from the current study indicated that younger age is related to greater perceived risk. This may reflect a belief or fear that a diagnosis of AATD could have long-term negative consequences. Many members of the AATD patient community are aware of the case of Terri Sergeant, recently awarded damages by the Equal Employment Opportunity Commission after being fired from her job as an office manager because she required extremely expensive medication to treat her AATD.¹³

The 24.4% prevalence of smoking among study participants is consistent with the current national statistic of 23.5%.²³ One of the most important outcomes of establishing a diagnosis is to effect smoking cessation because smoking is the most common environmental factor associated with developing AATD related lung disease. More than 75% of ACT participants report a strong likelihood of smoking cessation if they are diagnosed with AATD. Little is known about smoking cessation among a newly diagnosed adult population predisposed for lung disease. Followup of this cohort may provide insight to success in quit attempts because smoking cessation intent does not equate with cessation. Results from the National Heart Lung and Blood Institute's Registry of Patients with the deficiency of α_1 antitrypsin suggest that the AATD population is amenable to smoking cessation since only 8.3% of that cohort reported current smoking.²⁴ Also, previous studies indicate that screening at birth leads to a lower incidence of smoking among PiZZ individuals, suggesting that at-risk populations may benefit from early detection.²⁵

Although < 40% of participants rated the likelihood of losing insurance or increased premiums a high risk at the pretest

Table 4
Comparison of participant plans to tell others about their test result between those with a deficient (PiZZ or PiSZ), carrier (PiMZ), or negative (PiMM or PiMS) test result

	% Who will tell a deficient test result out of total (N)	% Who will tell a carrier test result out of total (N)	% Who will tell a negative test result out of total (N)	P
Current spouse	100.0 (21)	96.5 (144)	94.4 (199)	0.57
Ex-spouse	16.7 (6)	35.3 (34)	26.8 (41)	0.65
At least one sibling	92.0 (25)	94.9 (176)	77.3* (220)	<0.001
Children	100.0 (20)	95.4 (130)	84.9* (179)	0.003
Parents	95.2 (21)	94.7 (152)	80.8* (167)	<0.001
Employer	26.7 (15)	17.9 (106)	27.8 (126)	0.18
Best friend	66.7 (24)	68.9 (161)	63.4 (186)	0.55
Pastor or priest	30.0 (10)	30.2 (96)	30.0 (110)	0.99
Personal physician	82.6 (23)	59.9 (172)	61.6 (198)	0.11
Health insurance co.	10.5 (19)	17.3 (156)	27.3* (176)	0.05
Life insurance co.	6.7 (15)	15.2 (145)	25.7* (152)	0.037

* Different from carrier groups ($P < 0.05$).

Table 5
Differences in Likert (1–5^a) responses on the posttest questionnaire between participants receiving a deficient (PiZZ or PiSZ), carrier (PiMZ), or negative test result (PiMM or PiMS)

	Deficient test result (N = 32)		Carrier test result (N = 203)		Negative test result (N = 277)		P
	Mean	SD	Mean	SD	Mean	SD	
I will have improved health	3.72 ^b	1.30	3.11 ^b	1.40	2.43 ^b	1.50	<0.001
I will have improved psychological well being	3.09	1.5	3.27	1.23	3.42	1.46	0.15
I will become depressed	2.00	1.30	1.50	0.81	1.20 ^c	0.70	<0.001
I will become anxious	1.91	1.17	1.63	0.95	1.21 ^c	0.67	<0.001
The test will have a negative impact on my quality of life	1.56	1.11	1.60	0.98	1.33 ^d	0.93	<0.001
The test will have a positive impact on my quality of life	3.22	1.51	3.05	1.37	3.20	1.54	0.40
I will feel more in control of my life	2.96	1.48	2.97	1.36	3.00	1.55	0.98
I or my insurance company will spend more money on healthcare	2.86 ^b	1.75	2.05 ^b	1.22	1.43 ^b	0.91	<0.001
I will encounter social or workplace discrimination	1.28	0.77	1.35	0.84	1.09 ^c	0.44	<0.001
I have received important information about my genes	4.56	0.87	4.48	1.02	4.22	1.24	0.09

^a (Likert 1 = Not Likely, Likert 5 = Very Likely)

^b Different from all other groups in pairwise comparisons ($P < 0.02$)

^c Different from all other groups in pairwise comparisons ($P < 0.001$)

^d Different from carriers in pairwise comparisons ($P < 0.001$)

questionnaire, the posttest results were different. Overall 40% were unsure if they will tell their physician about their test result and 80% were unsure if they will tell their health insurance company. This discrepancy is interesting and worthy of further study to understand if it is unique to this study design.

Participants receiving a deficient test result rate the expectation of improved health higher compared to participants with a negative test result. This finding may suggest that knowledge of genetic status may increase a sense of control and therefore serve as a stimulus for healthy behavior change for these participants.

Past studies show an interest and willingness to pursue genetic testing, especially within specific at risk populations.^{16,17} Although self reported interest might be high, this does not always translate into actual behavior. Among individuals who initially showed interest in testing for Huntington's disease (about 2/3 of an at risk sample), only 15% engaged in testing.²⁶ Return rates from the ACT Study are similar as only 33% of requesters returned a completed test. Factors influencing the decision to follow through with genetic testing are not well understood. Cost, perceived risk, convenience, and education level have all been linked to the decision to go forth with genetic testing.^{17,20,21} Although such factors may be influential, perhaps other social, emotional, and psychological issues play a role in this decision.

The majority of ACT testers did not report depression and anxiety after knowledge of genetic status. However, a subgroup was different with 4 of 31 deficient participants reporting high scores for these questions. This finding suggests that some participants with a deficient test result may perceive themselves as more psychologically vulnerable. We found 25% of persons

with a deficient test result reported a moderate likelihood of depression with a score of 3% versus 12% of carriers and 7% of participants with a negative test result. Results were similar for anticipated anxiety. The findings from the present study indicate that knowledge of a deficient test does result in moderate distress for some people.

Limitations

The primary limitation of this study is its use of a self-selected sample. Participants sought out and/or volunteered for genetic testing, implying that they had some degree of comfort with the testing process and were perhaps well equipped to accept the results. Those truly anxious or fearful of genetic testing may have ignored the opportunity. The best methodology for studying interest and beliefs for genetic testing among the general population would use epidemiologic research.^{8,9} However, testing of the general population has not been recommended in the recently published Statement on Standards for the Diagnosis and Management of Individuals with AATD.²⁷

Another limitation was the use of nonstandardized, researcher-adapted assessment measures to examine perceptions and beliefs about genetic testing. Prior research on interest in and anticipated outcomes from genetic testing has predominantly included nonstandardized assessment measures as well, including qualitative data derived from focus groups. As genetic testing becomes more widespread, formal assessment procedures will become necessary and will allow for cross-study comparisons.²⁸

It is possible that risks and benefits of testing may have been scored differently if circumstances of testing were different. We provided a highly confidential testing environment in which

patient identifiers could be removed after testing. These protections were essential to determine if anxiety about the testing was focused on confidentiality or other aspects of testing.

We were disappointed with the posttest questionnaire return rates despite intensive efforts with telephone calls, mailed reminders, and monetary stipends to improve return rates. Not all participants answered all questions. Although we found few differences between the pretest questionnaires of those who returned and did not return their posttest paperwork, the 63% return raises the possibility of selection bias and limited the numbers of PiZZ and PiSZ participant responses included in the posttest questionnaire analysis.

Finally, the posttest questionnaire assessed anticipated but not actual effects. Posttest questionnaires were delivered at the same time as test results. At best, these anticipated effects reflect initial reaction upon knowledge of genotype. Future research should examine the intensity and duration of both psychological and physical impact of AAT deficiency longitudinally, although one study has shown that general quality of life among AAT-deficient individuals remains fairly stable over a two-year period.²⁹

Conclusions

As knowledge of genetics grows, so does the technology for quick and inexpensive testing. It is not inconceivable that the future will allow for home testing for a variety of genetic conditions. Public reaction and tolerance for the availability of such a market is unclear. Gaining more understanding toward motivators for genetic testing and the reaction to mailed results in the context of a comprehensive telephone support system will greatly influence the future of genetic testing. Evidence to date suggests that some people are receptive to home genetic testing.

This study sought to further examine perceived risks and benefits both before and after testing for AATD. Before testing, anticipated benefits appeared to outweigh anticipated risks. After testing, respondents who tested deficient or carriers for AATD anticipated some negative outcomes, including depression, anxiety, and increased health insurance costs. However, in agreement with expectations from the initial questionnaire, most reported feeling more in control of their lives and persons with an AATD diagnosis expected improved health. In the confidential setting of the ACT study, participants perceive the benefits of testing to greatly outweigh the risks.

ACKNOWLEDGMENTS

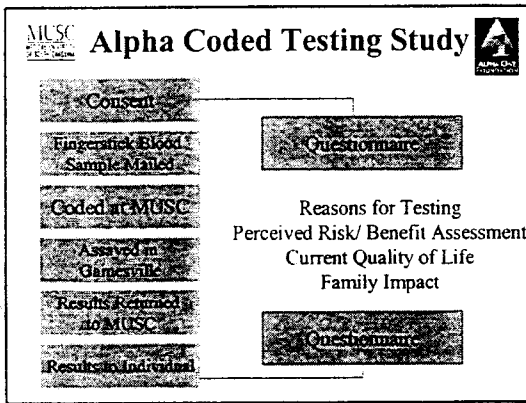
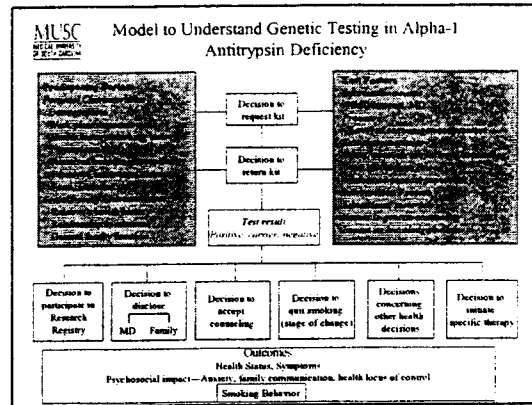
The authors extend their appreciation for funding to the Alpha-1 Foundation and to Kathy Valenti, Marc Yarborough, and Ann Knebel who assisted in study design.

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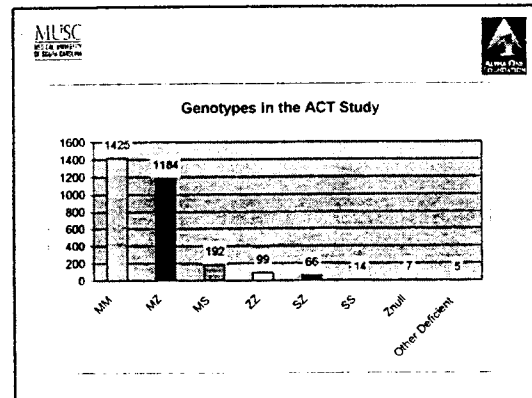
MUSC
 Alpha Coded Testing Study

- Study Aim:** Develop a model to explain the contributions of personal, family and test characteristics to decision-making concerning genetic testing. Understand the implications for education and counseling.



MUSC
 Alpha Coded Testing (ACT) Study
 1/1/02- 9/1/04

	N
Fingerstick Test Kits Mailed	6,268
Kits Returned	3,102 (49%)
Continue for 5 years	(67%)
Results Mailed	2,939



Risks and Benefits of Genetic Testing

Risks	N	Mean*	SD	% of total
1. Risk of discrimination	647	2.7	1.1	77
2. Privacy and confidentiality	77	2.7	1.1	9
3. Family conflict	77	1.7	1.2	22
4. Psychological distress associated with genetic test results	77	2.2	1.5	24
5. Embarrassment associated with genetic test results	77	1.2	1.1	15
6. Lack of understanding of genetic test results	77	1.7	1.1	22
7. Disclosure of test results to family members	77	1.7	1.1	22
8. Financial cost of genetic testing	77	1.7	1.1	22
Total	708	1.94	0.89	17
Benefits				
1. Knowledge of health status	432	4.34	1.11	62
2. Ability to make informed decisions about health care	444	3.89	1.41	67
3. Ability to take preventive health care	432	4.27	1.09	60
4. Ability to understand and manage health care	444	3.74	1.49	60
5. Ability to understand and manage health care	432	3.87	1.25	65
6. Ability to understand and manage health care	432	4.25	1.19	70
Total	708	4.03	0.97	71

* Mean score for each item on a scale of 1 to 5, with 1 being the lowest and 5 being the highest.

Post test: Who will you tell results?

	% Who will tell a Different Test Result (out of Total (N))	% Who will tell a Carrier Test Result (out of Total (N))	% Who will tell a Negative Test Result (out of Total (N))	P value
Current spouse	100.0 (23)	96.5 (14)	94.4 (19)	0.77
Employer	16.7 (15)	35.3 (13)	26.8 (11)	0.65
All next-of-kin	92.9 (22)	92.9 (17)	77.3 (12)	<0.001
Children	100 (20)	95.0 (17)	82.9 (16)	0.001
Employer	26.7 (15)	17.9 (10)	27.8 (16)	0.16
Best Friend	66.7 (21)	66.7 (11)	63.3 (10)	0.55
Personal Physician	82.6 (23)	84.6 (17)	81.9 (16)	0.11
Health Insurance Co.	16.7 (19)	17.3 (15)	17.3 (17)	0.95
Life Insurance Co.	6.7 (15)	15.2 (14)	25.7 (15)	0.04

ACT Study Research N=996

- ✓ Participants value receiving their results in a confidential setting
- ✓ Perceived benefits of testing outweigh the risks
- ✓ Participants who receive a positive test result are more likely to expect positive outcomes from testing as compared to participants with a carrier or negative test result
- ✓ Success of a home genetic testing service

Strange C. Genetics in Medicine 2004;6:204-10

Study of Non-participants 5/04

- 2345 Participants
 - 1196 (51%) did not return the test
- Random digit dialing
 - N=83 persons estimated sample size
 - Telephone administration of Family Assessment Device, Beck Anxiety Index, Impact of Events Scale

Odds Ratios and 95% confidence intervals for the logistic regression model to describe reasons for non-return

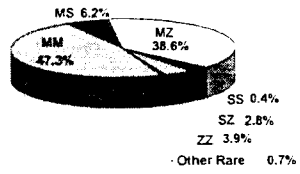
Variable	Odds Ratio	95% CI
BAI Score	1.0	0.98-1.03
FAD Score	0.98	0.94-1.01
IES Score	0.99	0.97-1.01
Gender	0.76	0.44-1.3
Age (10 years)	0.74	0.60-0.82
Finger Stick Fear	0.74	0.60-0.93
Too Much Paperwork	0.89	0.69-1.15
Concern for Confidentiality	1.26	1.01-1.57
Concern For What The Results Mean For Family	0.94	0.78-1.14

Smoking Cessation in ACT N= 105/480

Quit Attempts	Genes			Total
	MM/MS	MZ	SZ/ZZ	
Yes	14	16	8	38
No	46	18	3	67
Total	60	34	11	105

3 Month Cessation	Genes			Total
	MM/MS	MZ	SZ/ZZ	
Yes	1	4	2	7
No	59	30	9	98
Total	60	34	11	105

Alpha 1 Coded Testing Program
(n=3293)



- >1500 are either carriers or profoundly deficient
- >100 subjects request testing each month

Discrimination as a Consequence of Genetic Testing

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Summary

Genetic discrimination refers to discrimination directed against an individual or family based solely on an apparent or perceived genetic variation from the "normal" human genotype. We describe here the results of a case history study designed to assess whether or not genetic discrimination exists. Using the above definition of genetic discrimination and applying stringent criteria for case selection, we find that genetic discrimination exists and is manifested in many social institutions, especially in the health and life insurance industries. Stigmatization, and denial of services or entitlements to individuals who have a genetic diagnosis but who are asymptomatic or who will never become significantly impaired, is noted. Follow-up comprehensive studies on the significance and varieties of genetic discrimination are needed. In order to avoid creating a new social underclass based on genetic discrimination (the "asymptomatic ill"), existing and future genetic testing or screening programs need review by medical, scientific, legal, and social policy experts, as well as the public, and may require modification.

Introduction

The accelerating development of biochemical and DNA-based diagnostic tests for human genetic conditions in the last decade has engendered a revolution in genetic diagnosis. Numerous families faced with agonizing clinical or family planning decisions have been aided by information obtained through these tests (Phillips 1988). With further refinement of the human genetic map (Donis-Keller et al. 1987; McKusick 1988), the limited repertoire of genetic tests for the predisposition to common conditions such as cancer, cardiovascular diseases, and mental disorders may markedly expand (Lander and Botstein 1986; Scott 1987).

Insurance companies, private employers, govern-

ments and educational institutions all have an immediate or potential interest in promoting large-scale genetic screening to identify individuals carrying disease-associated genes (Motulsky 1983; Murray 1983; Uzych 1986; Hewitt and Holtzman 1988; Holtzman 1988, 1989; Nelkin and Tancredi 1989; Office of Technology Assessment 1990; Natowicz and Alper, in press). In addition, economic pressures to apply genetic tests to broader sections of the population may increase as biotechnology companies develop and sell genetic testing products and services (Hewitt and Holtzman 1988). Finally, the pace of development and application of DNA and biochemical genetic tests and their acceptance by the public may be accelerated by the recent widespread media coverage of the work of human geneticists.

As a result of the pressures to implement genetic technologies, problems engendered by their application may be overlooked. For example, with relatively few exceptions, our knowledge of how genes produce clinical illnesses is still quite limited. Yet an evaluation of the predictive value for clinical disease, utility, and impact is necessary before general use of genetic tests

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Genetic Testing and Discrimination

can be endorsed. In addition, some authors fear that an uncontrolled use of the tests may lead to a revival of social policies based on eugenics (Kevles 1985).

While there have been theoretical concerns about prejudices and discrimination surrounding genetic conditions, few investigations of these issues have been published (Hampton et al. 1974; Motulsky 1974; Kenen and Schmidt 1978; Murray 1991). Studies on the impact of genetic counseling have generally focused on subjects' understanding of genetic information and on family planning decisions (Leonard et al. 1972; Evers-Kiebooms and van den Berghe 1979); evaluations of the impact of genetic screening programs are typically concerned with issues of diagnostic sensitivity and specificity, medical efficacy, and cost-effectiveness and have usually not assessed long-term nonclinical outcomes. The personal costs of consenting to genetic testing and screening have not been studied and raise many issues of considerable significance.

Given this situation—powerful and attractive new techniques, social and economic forces pressing for their application, and an incomplete understanding of the potential negative social and personal consequences of genetic testing—concern about the burdens engendered by widespread utilization of genetic tests seems justified. One such issue is the problem of genetic discrimination.

This paper describes the results of a preliminary study of individuals labeled with genetic conditions. This study is not a survey; it does not purport to give statistically significant information about the extent of genetic discrimination. Instead, the aim of the study was to discover whether incidents which may reflect genetic discrimination are occurring in the workplace, in access to social services, in insurance underwriting, and in the delivery of health care. If the existence and range of genetic discrimination revealed by this study are confirmed by other investigations, then current policies and practices regarding the application of genetic testing and utilization of information obtained from such testing may need to be reconsidered.

Methods

A definition of genetic discrimination was developed for this preliminary study based in part on the work of the Genetic Screening Study Group, a Boston-based public interest group (Dusek 1987; Billings 1989; Beckwith 1991; Natowicz and Alper, in press). Discrimination stemming from supposed hereditary

transmission of a condition can be obvious, as in the case of an individual who was denied a job because a health record noted that the applicant's mother was "schizophrenic." In other instances, the distinction between discrimination based on clinical disability or illness, and that arising from genetic aspects of a condition, may be more difficult to determine. For the purposes of this study, genetic discrimination is defined as discrimination against an individual or against members of that individual's family solely because of real or perceived differences from the "normal" genome of that individual. Genetic discrimination is distinguished from discrimination based on disabilities caused by altered genes by excluding, from the former category, those instances of discrimination against an individual who at the time of the discriminatory act was affected by the genetic disease.

An advertisement to solicit cases of possible genetic discrimination was mailed to 1,119 professionals working in the fields of clinical genetics, genetic counseling, disability medicine, pediatrics, and social services in New England. This solicitation was also published in the *American Journal of Human Genetics* (Billings 1988). Similar appeals were reprinted in newsletters of organizations of individuals with genetic conditions, such as associations for persons with Friedrich ataxia, Charcot-Marie-Tooth disease (CMT), and muscular dystrophy.

Many responses included supporting documentation. Each described incident was reviewed independently by two of the authors (P.R.B. and M.A.K.), and a decision was reached as to whether it met the standard for inclusion in our study. This preliminary investigation was closed after receiving responses for 7 mo.

The most common reasons for excluding responses in this study were that (1) the differential treatment was based on a physical variation or disability (for example, individuals with Turner syndrome who were discriminated against in employment because of short stature), (2) there was a lack of evidence that the differential treatment arose from the hereditary nature of the condition, and (3) there was inadequate information submitted to determine whether discrimination had occurred.

Results and Discussion

Of the 42 responses received, 13 (31%) were excluded from further study because of failure to meet our strict criteria for genetic discrimination or because

insufficient information was provided to enable an accurate assessment. The remaining 29 responses came from all regions of the United States and Canada. A variety of genetic conditions were represented in the study group, including Huntington disease, Friedreich ataxia, CMT, hemochromatosis, phenylketonuria (PKU), and others. Most of the responses were elicited by the advertisement reprinted in newsletters.

The 29 evaluated responses described 41 separate incidents of possible discrimination. Of these 41 incidents all but two involved insurance or employment. Thirty-two incidents involved insurance (applications or coverage changes for health, life, disability, mortgage, and auto insurance), and seven involved employment (hiring, termination, promotion, and transfer).

The respondents described difficulties in obtaining desired insurance coverage, in finding or retaining employment, and in interactions with adoption agencies. Problems with insurance companies arose when individuals altered existing policies because of relocations or changes of employers. New, renewed, or upgraded policies were frequently unobtainable even if individuals labeled with genetic conditions were asymptomatic. Assessment of the natural history of the genetic condition or evaluation of the fitness of the individual by physicians had little or no influence on the adverse outcomes presented by the respondents. Because of fear of discrimination, several respondents reported that they withheld or "forgot" to mention potentially important medical or family history information to physicians, employers or insurers. Others reported that their insurance agents suggested that they give incomplete or dishonest information on insurance application forms.

The responses excerpted below illustrate several themes that appeared repeatedly in the data. These themes can be grouped into three categories: (1) "The Asymptomatic Ill," (2) "The Problem of Variability," and (3) "The At-Risk: to Test or Not to Test? Dilemma." While the incidents suggest discrimination by specific institutions such as insurance companies, the cases and types of genetic discrimination described below may reflect the attitudes and practices of an array of business, social, and political institutions.

The Asymptomatic Ill

Many individuals identified as having a hereditary condition are healthy. Some have undergone testing only because other affected family members have been identified with a genetic condition. As the number of genetic tests increase, and are widely applied, an in-

creasing number of individuals will discover that they harbor a disease-associated gene but have no identifiable clinical illness.

A respondent from a family with hereditary hemochromatosis wrote: "In 1973, at age 27 and 1/2, I was diagnosed as having excessive iron storage and was put on a regime of phlebotomies. . . .

"[After several years] I have never had the slightest symptom, in part because early detection [and appropriate treatment] of iron overload in my case avoided damage. . . .

"[After failing to get insurance because of my hemochromatosis] I have supplied doctor's testimonies to no avail. I might as well have AIDS. Even though I have proven that I prevented health problems by early detection and prophylaxis, they condemn me to the same category as lost causes. I run 10 Km races, etc. I am not a basket case, and will not be one, ever, because of iron overload."

With respect to a second case, a physician reported that "an individual was found to have Gaucher Disease. His brother was screened and the results were consistent with unaffected carrier status [heterozygote]. The brother applied for a governmental job and included the history of his testing in the application. He was denied the job because of his being a 'carrier, like sickle cell'."

With respect to a third case, a clinical geneticist caring for individuals with PKU wrote: "[Name withheld] is an 8 year old girl who was diagnosed as having PKU at 14 days of age through the newborn screening program. . . . A low phenylalanine diet was instituted at that time. . . .

"Growth and development have been completely normal. Height, weight, and head circumference all follow the 25th percentile. Routine developmental assessments done at 26 weeks, 53 weeks, and 54 months revealed skills solidly appropriate for age, and in many instances skills were above age-expected levels. The child continues to be developmentally normal and be healthy. The circumstances of the discrimination that this child has experienced involve rejection for medical insurance. She was covered by the company that provided group insurance for her father's previous employer. However, when he changed jobs recently, he was told that his daughter was considered to be a high risk patient because of her diagnosis, and therefore ineligible for insurance coverage under their group plan. She is currently being covered at the expense of her family, but this is a temporary solution at best. The family has written to the agency that administers

Genetic Testing and Discrimination

the group insurance plan to obtain details of the decision to deny coverage and also plans to write to the chairman of the large corporation for which the father works. All information will also be submitted to the [state] insurance commissioner.”

Comment

The first case illustrates both the promise and the burden of genetic testing. It was through genetic testing that this individual was diagnosed and successfully treated. Yet, because of his test results, he has been stigmatized and denied insurance as if he were severely ill. Similarly, the individual described in the second case was denied employment because of his genotype, despite the fact that he was asymptomatic and a heterozygote for an autosomal recessive condition.

The third case illustrates both the benefits of genetic testing in enabling the early detection and successful treatment of many children with PKU and constraints that can be imposed by genetic labeling. The cost of a phenylalanine-restricted diet, an effective treatment for PKU, is high. Without insurance, it is possible that the family would not be able to afford treatment for their child, with the consequent risk of developmental delays and permanent impairment. The family's life is restricted by the necessity for the father to maintain employment in the same state and at the same job in order to have access to insurance. Thus, the child's diagnosis has a major impact on the geographic and job mobility of other family members. These constraints, in turn, could cause economic or other potentially significant limitations.

By preparing to take their situation to a state agency, this family demonstrated “self-advocacy” abilities. The poor, the uneducated, foreign nationals, or those with fears about their job security may not be as willing or able to negotiate the complexities of our legal and regulatory systems in order to secure their rights.

All three cases illustrate instances of discrimination against individuals who are completely asymptomatic; their only “abnormality” lies in their genotypes. As large numbers of individuals submit to or are coerced into testing in order to obtain employment or insurance coverage, a new social class and category—the “asymptomatic ill”—may be constructed. Although they are healthy, persons in this new group may find that they are treated as if they were disabled or chronically ill by various institutions of our society (Marx and Sherizen 1986; Nelkin and Tancredi 1989).

All of the cases suggest that access to jobs, insurance, or social entitlements may be limited because of

genetic discrimination. Stigmatization and frustration (“I might as well have AIDS”) may accompany the test result. The financial and legal burdens of maintaining a reasonable standard of living and basic entitlements as a result of genetic discrimination could be significant.

The Problem of Variability

Several responses described situations in which people were victimized because of a misunderstanding of the clinical variability of many genetic conditions. One respondent with CMT, a nonfatal, clinically variable, and genetically heterogeneous neuromuscular condition, wrote: “I have been rejected for life insurance many times, but only once was CMT cited [explicitly] as the reason. . . . [I appealed, informing the insurance company] that people do not die from CMT and that they had declared me automatically eligible for accidental death insurance—the one risk that can be assumed might be greater for people with CMT. . . . [The insurance company's reply] repeated the statement that CMT is the reason for rejecting my application.

“In 1979 my daughter was denied employment by the [name omitted] Company because she has CMT even though the case is not really noticeable. She had indicated on the form that she had CMT and the examiner asked her what it stood for; then, he looked it up in a medical book and denied her a job which had been offered to her by the recruiter.”

Another respondent stated: “. . . My husband has a genetic disorder, Charcot-Marie-Tooth. We have just been turned down for automobile insurance with [name omitted] because of his disease. I have just recently sent them a letter from my husband's doctor . . . explaining that my husband is a far better driver than anyone I know. . . . My husband has had NO accidents, or traffic violations since he has been driving from the age of seventeen [twenty years of driving].”

Comment

In these examples involving CMT, the individual does suffer from a disease, but the symptoms are mild. The decisions regarding life and automobile insurance, and employment, were based solely on a diagnostic label, without regard to the severity of the condition for each individual. In these and other cases, having a particular genotype is equated with the presence of a severe illness and the lack of effective treatments. This evaluation of genetic conditions illustrates a lack of understanding of the concepts of incomplete

genetic penetrance, variable expressivity and genetic heterogeneity. In many cases, the worst possible scenario seems to be the standard used for policy decisions regarding at-risk individuals. Yet wide variation in clinical manifestations of a gene-associated disorder—individuality—is common. As highlighted in these examples, an individual may suffer serious consequences as a result of the inaccurate and unfair simplification of genetic conditions.

The At-Risk: to Test or Not to Test? Dilemma

The third category includes those individuals who are currently healthy but who may be at risk for a genetic disease. Discrimination may ensue as a result of a decision to forego testing and thereby not determine whether they (or their future children) will develop the disease. Discrimination may also occur if they opt for such testing and the results reveal a genotype associated with disease.

A respondent wrote: "I am at risk for Huntington Disease [age 31]. After many years of consideration, my husband and I decided not to bear our own children, but rather to adopt children, so as not to take the chance of passing on the Huntington gene.

"In 1987 we began investigating adoption. We encountered restrictions due to religion and availability of infants, but were finally invited to make application with [name and location of agency omitted]. We began our counseling process and our home visits, at which point the issue of my being at risk was discussed (I had disclosed in my original application the possibility of my developing the disease and why we had chosen adoption). Before completion of our home study, we were asked to withdraw our application, because of the Huntington Disease situation. . . .

"We understand the right to choose the BEST 50 couples out of some 500 applicants per year for placement. Availability of children is incredibly limited. And yet, should I be judged by a disease that I am only at risk for and that may not develop for some years to come? Does this make me different than anyone with diabetes or cancer, for example, in their ancestry?"

Another couple at risk for Huntington Disease sent a letter they received from an adoption agency: "We have decided, in your situation, not to proceed with your application because there is a fifty-fifty chance of your getting Huntington Disease. Though you would be likely to get the disease around the age of fifty, it could be sooner. You would not receive a child from us, if we could proceed with your application, for several years, and therefore we would be risking the

likelihood of not having you available to the child until he/she has reached adulthood. We feel that a fifty-fifty chance of getting a disease as serious as Huntington Disease is too great a risk, for our purposes and circumstances."

In a third case, a physician informed us that "a family with a child who has cystic fibrosis received health care through an HMO. When a second pregnancy occurred, prenatal diagnosis using DNA analysis was instituted. Fetal DNA tested positively for two copies of a mutation associated with cystic fibrosis. Nevertheless, the family decided to proceed with pregnancy. After disclosure of the test result, the HMO considered withdrawal of or financial limitations on the health care coverage for the pregnancy, postpartum and pediatric care, as well as for the already affected child. Threats of legal action were required before this situation was resolved."

Comment

In the first two cases, the adoption agencies' attitude illustrates how certain conditions categorized as "genetic" are viewed as special and handled differently in terms of social decision making. The use of genetic tests to ascertain the genotype in such at-risk individuals will not necessarily lessen the chances of subsequent discrimination, since a positive test will not be an infallible predictor of the burden a genetic trait may place on an individual, nor does it predict the abilities of affected individuals as parents or the qualities of a potential home.

These incidents also illustrate a large eugenic prejudice—the myth of genetic perfection (Billings 1989; Suzuki and Knudtson 1989). The agencies assume that the best possible family is the one least likely to face medical adversity—the "perfect" family with a disease-free genome. Unfortunately, all families are at risk. The comparison made by one respondent, of being at risk for Huntington disease with susceptibility to diabetes or cancer, highlights a prejudice—that the chance of developing a genetic condition is perceived differently from a similar probability of contracting an illness not produced primarily by a gene.

The third case raises several social and ethical problems. The family was faced with the possibility that bearing a child with a certain genotype might reduce their access to necessary health care benefits. These circumstances constitute a strong incentive for aborting a fetus on the basis of its genotype, a practice that might be interpreted as a form of eugenics. Finally, the couple was forced to threaten legal action to avoid

Genetic Testing and Discrimination

the abortion and loss of benefits, an alternative not available to people who cannot afford an attorney. In addition, pressuring a couple to have an abortion seems to infringe on a right many believe to be fundamental, the right to bear children.

Conclusions

This pilot study identifies multiple facets of genetic discrimination. It does not document the full range of the prejudices faced by individuals with genetic diagnoses, nor does it establish the prevalence of these attitudes or discriminatory practices. A comprehensive study of the significance and varieties of genetic discrimination is critical to design strategies to ensure the ethical and appropriate use of genetic testing in the future. The need for such studies is underscored by the large number of genetic tests currently performed each year. In addition, powerful business interests support larger applications of genetic techniques because of the revenues that tests and kits generate directly and indirectly.

The dominant theme noted in the responses in this study is that genetic conditions are regarded by many social institutions as extremely serious, disabling, or even lethal conditions without regard to the fact that many individuals with "abnormal" genotypes will either be perfectly healthy, have medical conditions which can be controlled by treatment, or experience only mild forms of a disease. As a result of this misconception, decisions by such institutions as insurance companies and employers are made solely on the basis of an associated diagnostic label rather than on the actual health status of the individual or family. The appropriate use of genetic testing information to restrict or limit access to public entitlements such as health care or employment has not been established and may not exist. The cost of such labeling is magnified by the fact that errors in testing and interpretation do occur.

Once labeled (possibly erroneously), an individual may suffer serious consequences, as highlighted in the examples. These include the inability to get a job, health insurance, or life insurance, being unable to change jobs or move to a different state because of the possibility of losing insurance, and not being allowed to adopt a child.

Furthermore, information related to genetic labeling may enter large-scale data banks now used to store personal health-related information. Individuals' health profiles, which can include genetic conditions, are

available privately and are generated in a manner similar to the ubiquitous credit checks encountered in business. Breaches of confidentiality and unauthorized uses of this information may arise (Dezell 1984; Norton 1989; Billings 1990; DeGorgey 1990). Genetic data on certain groups within our society are already being stored by governmental agencies (DeGorgey 1990).

These incidents suggest that there are very important nonmedical reasons why individuals may wish to avoid genetic testing. It is clear that the option not to know is being exercised now. Many persons at risk for Huntington disease, for example, have refused to be tested (Meissen et al. 1988; Brandt et al. 1989). The choice to refuse testing will become more difficult if such testing is required for employment, to obtain affordable medical care, or to obtain or renew health or life insurance. The personal decision to undergo genetic testing certainly requires accurate information about the clinical predictive value of genetic tests (data which may not be available currently), but the widespread utilization of genetic testing as a prerequisite for obtaining social entitlements may also influence an individual's right to choose not to know his or her genetic predispositions.

Various constraints exist to prevent or limit abuses made possible by access to genetic information. These include, for example, laws affecting privacy and civil rights (Russell-Einhorn and Rowe 1989; Natowicz et al. 1992). In addition, limitations exist on the uncontrolled expansion of genetic testing and screening. These include opposition by individuals and interest groups (such as labor unions and disability advocates) sensitized by the dismal history of eugenics movements and suspicious of genetic and scientific incursions into normal daily life, economic constraints (i.e., the poor profit yield of some tests), and state and federal regulations regarding testing and screening.

Nevertheless, it is clear that unfair and discriminatory uses of genetic data already occur under current conditions. Enacted state and federal laws are inadequate to prevent some forms of genetic discrimination, particularly that due to the health insurance industry (this report; Natowicz et al. 1992). The implementation of carefully considered legislation regarding, for example, the privacy of genetic information and the imposition of meaningful penalties on social institutions such as insurers that are found guilty of genetic discrimination are mechanisms to address aspects of genetic discrimination. Consideration of alternative medical care systems is another potential approach

toward solving some of these issues. Any approach should include the reaching of a broad-based public consensus on the appropriate use of genetic tests. Without further changes in social attitudes, legal protection, and/or changes in the prevailing American health care system, many healthy and potentially productive members of our society will suffer genetic discrimination.

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Individual, Family, and Societal Dimensions of Genetic Discrimination: A Case Study Analysis

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Keywords: genetic discrimination, genetic testing, Medical Information Bureau, state insurance commissions.

ABSTRACT

Background. As the development and use of genetic tests have increased, so have concerns regarding the uses of genetic information. Genetic discrimination, the differential treatment of individuals based on real or perceived differences in their genomes, is a recently described form of discrimination. The range and significance of experiences associated with this form of discrimination are not yet well known and are investigated in this study.

Methods. Individuals at-risk to develop a genetic condition and parents of children with specific genetic conditions were surveyed by questionnaire for reports of genetic discrimination. A total of 27,790 questionnaires were sent out by mail. Of 917 responses received, 206 were followed up with telephone interviews. The responses were analyzed regarding circumstances of the alleged discrimination, the institutions involved, issues relating to the redress of grievances, and strategies to avoid discrimination.

Results. A number of institutions were reported to have engaged in genetic discrimination including health and life insurance companies, health care providers, blood banks, adoption agencies, the military, and schools. The alleged instances of discrimination were against individuals who were asymptomatic and sometimes impacted on other

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asymptomatic relatives. Few surveyed respondents knew of the existence of institutions such as state insurance commissions or the Medical Information Bureau, Inc., which may play roles in redress of grievances or correction of misinformation.

Conclusions. Genetic discrimination is variable in form and cause and can have marked consequences for individuals experiencing discrimination and their relatives. The presence of abnormal genes in all individuals makes each person a potential victim of this type of discrimination. The increasing development and utilization of genetic tests will likely result in increased genetic discrimination in the absence of contravening measures.

The growing knowledge of human genetics, stimulated in part by the Human Genome Project, has engendered a societal need to understand potential hazards as well as benefits of this knowledge. With the increased ability to identify genetic differences, it is important to elucidate appropriate uses of genetic information from the perspective of both individuals and the public. At the same time, safeguards must be developed to minimize inappropriate uses of this information¹⁻⁴. One area of concern is genetic discrimination.

The term "genetic discrimination" has been used to describe the differential treatment of individuals or their relatives based on actual or presumed genetic differences as opposed to discrimination based on phenotype⁵⁻⁷. Sources of genetic information that enable such discrimination include genetic and sometimes non-genetic medical tests, family histories, and information obtained from clinical examinations. Each of these sources has important limitations in terms of its reliability in predicting whether a particular genetic condition will occur and, if so, the clinical course of the associated disease. These limitations include but are not restricted to the sensitivity and specificity of genetic tests, the intrinsic clinical variability of many hereditary conditions, and the importance of environmental factors.

A pilot study of genetic discrimination showed this problem to involve more disorders than was previously revealed by isolated reports. The reported incidents involved a variety of social institutions such as life and health insurance organizations, and suggested that genetic discrimination may become a significant social policy problem⁵. Based on this work and reports of genetic discrimination by others, there is now serious concern regarding the importance of this problem^{3-5, 8-11}. This concern is intensified by the proliferation and increasing utilization of genetic tests made possible in part by technological advances made by the Human Genome Project and by the application of these technologies by commercial interests¹²⁻¹⁴.

Previous reports of genetic discrimination involved studies of relatively small numbers of individuals and, consequently, would not be expected to reveal a full range of the discriminatory experiences faced by affected individuals and their families. Here we report on case studies obtained in 1992-1993 of individuals living throughout the United States at-risk for or related to people at-risk for the following disorders: hemochromatosis, phenylketonuria (PKU), mucopolysaccharidoses (MPS), and Huntington disease. We describe the spectrum of discrimination reported by some of these individuals and discuss its implications through analysis of selected informative cases.

METHODS

Study Design

Individuals at-risk for genetic discrimination were sent questionnaires during 1992-1993. The definition of genetic discrimination has been previously presented and distinguishes genetic discrimination from discrimination based on phenotype¹⁵. Only those cases in which individuals appeared to have no symptoms (i.e. no apparent phenotype at the time of the reported discrimination) were included in this study.

Questionnaires and Interviews

Two questionnaires and an interview instrument were developed based on findings from our pilot study. They were reviewed by consultants experienced in qualitative research methods and questionnaire design and were approved by the Shriver Center's Institutional Review Board. The two questionnaires were distributed through the mailing lists of genetic disease organizations selected according to the criteria described below, and were accompanied by a letter describing the research group and the goals of the study. One questionnaire was directed at individuals who had or were at-risk to develop a genetic condition. The other questionnaire was directed at parents who had a child with a genetic condition. Both were designed to acquire information about whether an individual believed that she/he or a relative had experienced discrimination because of a genetic diagnosis or assumption of genetic predisposition to the disorder, as well as a brief description of the discriminatory event(s) (Table 1, see p. 85). The descriptions of the alleged discriminatory events were used to screen the returned questionnaires for cases that appeared to fit the definition of genetic discrimination used in this study. That is, the person alleging discrimination was not symptomatic for a genetic disorder (or any other disease which might confound the claim of discrimination) nor did the complaint appear to involve legitimate actions by companies or individuals that were construed as "unfair" by the individual claiming discrimination. Some cases were included where an apparent conflict between the perception of the individual and the apparent point of view of an institution illustrate areas of ambiguity concerning the "fairness" of the situation.

Telephone interviews were conducted with individuals who, from their responses to the questionnaire, appeared to have experienced genetic discrimination as described above. An attempt was made to interview all individuals whose questionnaire answers met the above criteria and who indicated in their returned questionnaires that they were willing to be contacted by telephone. The script (available by request) used to conduct the interviews was designed to obtain more detailed information about the perceived discriminatory event(s) (Table 2, see p. 86-87). For example, in cases of alleged discrimination by an insurance company, the interviewee was asked about how the company found out about his/her genetic status, what correspondence and conversations were held with individuals at the company, what reasons were given by the company for actions taken, whether outside support or counsel was sought, whether the case was reconsidered after communication with the company, length of time of response, who within the company handled the case, whether alternative policies were offered, etc. In addition, general information was elicited regarding the economic and educational status of the individual, as well as pertinent medical

information including whether the individual was asymptomatic. We also sought to determine the origin of the genetic diagnosis or supposed genetic risk factor, perceived ability to redress a grievance, the extent to which an individual challenged adverse decisions, and the impact of personal genetic information on the individual and her/his family. The Medical Information Bureau, Inc. (MIB) and state insurance commissions are institutions which may be useful to individuals seeking redress regarding insurance. After conducting a number of interviews, it became apparent that the respondents differed widely in their knowledge concerning avenues for seeking redress for complaints involving insurance. Consequently, questions concerning whether the individual knew of the MIB and state insurance commissions were added to subsequent interviews. Consenting interviewees were anonymously tape recorded to aid in the transcription of information. In addition, documentation of discriminatory events was sought, including such items as letters from insurance companies and medical and personal notes.

Study Groups

The specific disorders targeted for this study were Huntington disease, phenylketonuria (PKU), hemochromatosis, and mucopolysaccharidoses (MPS). They were chosen because they met the following criteria: (1) the genetic basis of the condition is known and unambiguous; (2) discrimination directed against individuals with these conditions would most likely be due to the genetic bases of these conditions, rather than due to physical symptoms; and (3) support groups for persons with these conditions exist so that individuals could be contacted easily. These conditions were also selected because they cover a spectrum of situations including dominant and recessive disorders, treatable and untreatable disorders, relatively common disorders for which screening programs exist, and rare disorders for which screening programs are not indicated. Note that the individuals with recessive disorders included both those with the genotype for the disorder and those who are simply carriers.

Huntington disease is a fatal, untreatable, autosomal dominant disorder whose symptoms generally appear in middle age. There is currently a molecular genetic test available to diagnose this condition. Hemochromatosis is an iron storage disorder with a variable phenotype, some individuals being completely asymptomatic. This autosomal recessive disorder is treatable by phlebotomy (drawing blood). PKU is an autosomal recessive disorder for which all newborns in the United States are tested. If left untreated the disorder results in mental retardation. However, PKU is successfully treated by placing the child on a special diet. MPS disorders are usually associated with mental retardation and organomegaly.

A total of 27,790 questionnaires were mailed by the following groups to their members: The Huntington Disease Society of America, The Hemochromatosis Research Foundation, The National M.P.S. Foundation, and the PKU Clinic at Children's Hospital, Boston, MA. These mailing lists included donors and interested individuals who were not appropriate respondents to the questionnaire, a factor which was expected to result in a low response rate. However, contacting individuals through a national organization was more likely to get a broader response than that obtained by contacting individuals through a few clinics. Respondents to the questionnaire

included individuals who were at-risk to develop a genetic disorder but who were not informed of their genotype, individuals who were presymptomatic for a specific genetic condition, and individuals who were asymptomatic because of receipt of a therapy or were heterozygotes and thus only "carriers" for an autosomal recessive condition.

RESULTS

Of the 917 returned questionnaires, 455 respondents asserted that they had experienced genetic discrimination and 437 that they had not. The remainder gave ambiguous answers that could not be specifically classified. Some respondents reported experiencing genetic discrimination in more than one setting. After screening the questionnaires for cases that appeared to fit the definition of genetic discrimination used in this study, we were able to contact for interviews 206 individuals who reported that they experienced genetic discrimination. Detailed breakdowns of the responses of all respondents to the questionnaire, categorized by disease group, are given in Table 3 (see p. 88).

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A variety of different institutions allegedly discriminated against the respondents. The majority of cases involved discrimination by health and life insurance companies but there were a number of cases involved employers, adoption services, and blood banks. The cases reported below are grouped according to agents/institutions allegedly engaged in the genetic discrimination, followed by results of the impact of genetic discrimination on individuals and family members, responses and counter-measures taken to mitigate the effects of genetic discrimination, and information pertinent to the underlying bases of this phenomenon.

Agents/institutions engaging in discriminatory practices

• Health and Life Insurance Corporations

Four aspects of discrimination are illustrated by the cases involving health and life insurance: (1) discrimination against individuals who were asymptomatic; (2) differential treatment of asymptomatic individuals or families once a genetic diagnosis was established, thus treating the genetic diagnosis as a preexisting condition; (3) the failure of some group insurance plans to provide coverage for qualified individuals with a genetic diagnosis; and (4) the loss of insurability suffered by relatives of an individual with a presumed genetic disease. These issues are illustrated by the following cases.

Case a: A health maintenance organization (HMO) had covered the medical expenses of a child since birth but refused to pay for occupational therapy after she was diagnosed with MPS-I, claiming that the condition was pre-existing. All bills relevant to the condition had been paid up to the time of diagnosis. The occupational therapy had been pre-approved by the managed care corporation. The situation was remedied after the family complained to a customer service representative of the HMO.

Case b: A private insurer in Colorado notified the parents of a three year old who had been recently diagnosed with an MPS syndrome that the child's policy was terminated

although the family had been on the policy for nine months before the diagnosis. After an extended negotiation that included retention of a lawyer and the threat of a lawsuit, the insurance policy was reinstated. However, a rider was added to the policy excluding coverage for two common MPS-related complications.

Case c: A 24 year old woman was denied life insurance due to her "strong family history of Huntington's Chorea" and the fact that she has never been tested to determine if she is "currently a carrier." The rejection letter stated that if she "should be tested and if found to be negative," the company would issue a standard contract.

Case d: A mother submitted applications for employment-based life insurance policies simultaneously for her two children, one of whom had Hurler syndrome, a form of MPS. Both were rejected. The rejection letter indicated that the child with Hurler syndrome was denied a policy because the condition is fatal; no reason was given for the denial of a policy to the other child. She was later able to obtain coverage for the healthy child through a different employer.

- Clinical Professionals

In several cases medical professionals reportedly pressured patients or clients at-risk for having children with serious genetic conditions, to undergo prenatal diagnostic testing or to forsake having children.

Case e: A PKU gene carrier reported that during a routine pediatric visit, her child's doctor advised her that it would be unwise to have more children and that she should consult a genetic counselor to understand "the implications of PKU".

Case f: A couple in which one member was at-risk for Huntington disease reported that physicians tried to compel them to undergo prenatal genetic testing and reportedly coerced them to sign a document agreeing to abort an affected pregnancy. They also reported being required by a health care provider to undergo genetic counseling despite their belief that they had comprehensive knowledge about the genetic risks and their decision to continue any pregnancy irrespective of Huntington disease status.

- Adoption

Three issues are illustrated by the cases of alleged discrimination by adoption agencies. They are: (1) a misunderstanding of the nature of the presumed genetic condition with consequent unfair treatment of the prospective parents; (2) the requirement that individuals "pass" a genetic test before being allowed to adopt a child; and (3) the assumption that individuals with genetic diagnoses should adopt only children at-risk of having a disability.

Case g: One respondent, a carrier for MPS, was required by an adoption agency to repeat the blood and urine tests routinely required of prospective parents. It was reported that agency personnel found it "inexplicable" that the original test results were normal in someone who was a carrier for a genetic disorder.

Case h: A married woman learned that she was at-risk for Huntington disease when she was 25 years old. A year later she and her husband decided to adopt a child on the advice of her physician. The latter told her it would be better for her not to have her own children and that she could easily adopt. She therefore underwent a tubal ligation and the couple began the adoption process. The adoption agency application asked why the couple was not able to have children biologically, inquired about the presence of hereditary disorders, and required certification from a doctor that the couple was sterile. Shortly after filing the application, the couple received a letter from the adoption agency refusing them the opportunity to adopt based on the woman's risk of Huntington disease.

Case i: A birth mother with Huntington disease was refused the opportunity to place her child up for adoption through a state adoption agency but the child was accepted by a private agency. A couple with one member at-risk for Huntington disease had been unsuccessful in trying to adopt a child who was assumed to be genotypically normal. However, that at-risk couple was permitted to adopt the at-risk infant.

- **Armed Services**

The case described below involving the armed services shows that even institutions as structured as the military may not have a consistent policy with regard to people at-risk for genetic conditions.

Case j: An individual enlisted in the Air Force and revealed his (approximately 50%) risk status for developing Huntington disease. When applying for reenlistment, he was discharged due to his risk status, although he was asymptomatic. The brother of this individual served in the Marines (who were aware of his risk status) until he became symptomatic for Huntington disease at which time he received a medical discharge and treatment at a V.A. hospital.

- **Employers**

In many of the cases involving employment, individuals believed that they were not hired or were fired because they were at-risk for genetic conditions. In other cases, individuals who were employed reported that they were reluctant to seek either a more desirable job or a job in a different location because they feared that they would be unable to obtain health insurance in their new position.

Case k: A 24 year old woman was fired from her job as a social worker shortly after her employers learned that she was at-risk to develop Huntington disease. In the eight-month period prior to her termination she received three promotions and outstanding performance reviews. However, while conducting an in-service training on admitting and caring for Huntington disease patients, she revealed that she had a family member with Huntington disease. Shortly afterwards, she was given a poor performance review. Her employers declined to give examples of poor performance. She was soon fired and told by a co-worker that the employer was concerned about her risk to develop Huntington disease.

Case l: A 53 year old man was interviewed for a job with an insurance company. During his first interview he revealed that he had hemochromatosis but was asymptomatic. During the second interview, the company representative told him that the company would be interested in hiring him but would not be able to offer him health insurance because of his hemochromatosis. He agreed to this condition. During the third interview he was told that although they would like to hire him, they were unable to do so because of his hemochromatosis.

- Educational institutions

Our study elicited a few reports of genetic discrimination occurring in educational institutions. As is the case for the examples described above, the examples of genetic discrimination by educational institutions involved the denial of opportunities to apparently qualified individuals because of a perceived genetic abnormality in those individuals.

Case m: In a small town, two healthy children attended the same school as their disabled brother. That brother had MPS II and attended a special education class. When in second grade, one of the healthy children was judged to have poor penmanship. A teacher decided that this indicated the onset of MPS II and sent the child back to first grade without consulting the parents or a physician. The parents protested and the child was placed in the appropriate grade.

- Blood Banks

Twenty-two respondents with hemochromatosis reported that they were not able to donate blood. The American Red Cross has a policy of rejecting blood donations from all individuals with hemochromatosis arguing that the donations are treatments, not gifts¹⁶. A significant number of respondents stated that they donated blood because their health insurance would not pay for phlebotomy treatments. In some cases, blood banks were willing to perform phlebotomies as treatment for a fee. Several of these cases have previously been discussed¹¹. In the case below, prejudice or ignorance of a medical condition apparently played a role in inappropriately denying a potential donor the opportunity to give blood.

Case n: A man who had regularly donated blood for a number of years was refused phlebotomy when the nurse responsible for scheduling at the local blood bank learned that he had been diagnosed with Huntington disease. Donating blood was important to this man as a way of making a contribution to society. His neurologic findings, if any, were apparently not an issue since the director of the blood bank invited him to resume his blood donations once that particular nurse retired.

Personal reactions among people at-risk for genetic discrimination

Not unexpectedly, experiencing one or more episodes of genetic discrimination engenders a gamut of personal/psychological reactions for both the affected individual and, often, for other family members. These involve loss of self-esteem, alienation from family members and others, and alterations in family dynamics. For example, some individuals reported that they felt stigmatized and unworthy of marriage or that

they should only marry disabled individuals. Others behaved as if they had a genetic condition even though they had not been diagnosed. In some instances, family members blamed each other for problems caused by a genetic diagnosis. The cases excerpted below illustrate some of these complex feelings and interpersonal dynamics.

Case o: A woman at-risk for Huntington disease chose her profession (school teacher) because it provided good benefits, especially disability benefits. Although some of her friends know of her risk she has kept the information from her employers as she "doesn't trust" them.

Case p: A woman at-risk for Huntington disease reported that friends and co-workers continually pressure her to undergo genetic testing arguing that if information can be obtained, she should not remain ignorant. As a result, she feels less free to discuss her risk status with friends and is resentful of the intrusion.

Case q: A woman learned of her risk of Huntington disease in adolescence. She reported feeling that she could not have a "normal" life, had no interest in marriage or children, and chose a career which had good disability benefits available. Later, genetic testing revealed that she was at low risk to develop Huntington disease and since then has actively pursued changes in career and marriage.

Strategies to avoid genetic discrimination

Many individuals utilized strategies to avoid experiencing genetic discrimination. These strategies included purchasing insurance policies prior to genetic testing, being tested anonymously, paying out-of-pocket for tests so that insurance companies would not obtain the results, providing partial disclosure of relevant information and, sometimes, providing incorrect information (see also reference 17).

Individuals reported avoidance of genetic testing or situations where genetic information could be used against them. For example, several of these respondents reported that they had never been rejected for insurance because they had not applied for insurance: they stated the belief (true in the case of Huntington disease) that their applications would necessarily be denied.

Case r: The parent of an individual died of Huntington disease. Fearing adverse consequences at work if the diagnosis became known, the individual arranged for the diagnosis "asphyxiation" to be reported as the cause of death so as to avoid mention of the disease in an obituary.

Case s: An 18 year old man, at-risk for Huntington disease, wished to enlist in the Marines in order to serve in the Persian Gulf War. He believed it unlikely that he would become symptomatic during his tour of duty but that his risk status would disqualify him from service. He therefore answered "no" to questions regarding hereditary disorders on his application and did not include Huntington disease in his family medical history.

Knowledge of the Medical Information Bureau

The Medical Information Bureau, Inc. (MIB) is a private, non-profit corporation which provides insurance companies with medical and certain non-medical information about potential insurees¹⁸. Member organizations (mostly insurance companies) have access to its computerized data bank of information about individuals. Thus, genetic information provided by the MIB could result in inappropriate discrimination in obtaining health or life insurance, particularly if recorded data are incorrect or misleading^{18,19}. The MIB does offer the opportunity for individuals to examine their MIB records and request corrections. However, doing so requires knowledge of the existence and function of the MIB. We therefore questioned individuals concerning their awareness of the MIB. Only 10 of the 55 (18%) respondents asked about the MIB knew of its existence and none had asked for access to their MIB records.

Knowledge and use of state insurance commissions

State insurance commissions are charged with regulating the insurance industries in their states. Consequently, appeal to a state insurance commission is one mechanism for challenging a perceived discriminatory decision regarding procurement of insurance. Therefore, study participants were sampled regarding their knowledge of state insurance commissions. Nineteen out of 58 respondents (33%) knew of the existence of state insurance commissions. Many of the nineteen thought the purview of these commissions was limited to automobile insurance.

DISCUSSION

The purpose of this study was to determine the varieties and impact of genetic discrimination using the case studies of individuals having, or who were at-risk for, abnormal genotypes. The results extend the findings of the pilot study: genetic discrimination exists and has significant impacts on individuals and their families. The pilot study provided evidence of genetic discrimination in several social arenas: health insurance, life insurance, and adoption. This study confirmed these findings, providing many more instances of genetic discrimination in each of these areas and in identifying additional institutions engaged in discrimination.

This study also revealed several additional ramifications of the phenomenon of genetic discrimination. First, some cases were reported in which clinical professionals appeared to give judgmental and possibly coercive counsel to persons who were at-risk for abnormal genotypes and/or who were at-risk of having offspring with abnormal genotypes. While all individuals, professional or otherwise, have personal positions on ethical issues, the imposition of a clinician's values with respect to reproductive decisions is regarded as inappropriate²⁰. It is not possible to determine whether the alleged incidents occurred exactly as they were related to us. Nonetheless, even if some of these cases reflect a misunderstanding between the health care provider and patient, they suggest that poor communication can give rise to the perception of discrimination.

Second, while this study was not designed to evaluate the psychological effects of genetic discrimination, the pervasiveness and importance of these effects became

apparent. The cases that we have summarized illustrate typical feelings expressed by respondents. Individuals reported stigmatization by relatives, friends, and other members of their communities. Some respondents reported that a genetic diagnosis resulted in feeling a loss of self-worth; others reported feeling powerless to challenge adverse decisions. These responses were common and indicate the significant impact that discrimination can have on people's lives.

Third, many of the individuals reported that they had not experienced genetic discrimination. However, comments on their questionnaires revealed that many of them had adopted strategies to ensure that others would not learn of their genetic backgrounds. Apparently they perceived a possibility of genetic discrimination and took action to avoid it.

Fourth, most respondents lacked either the information or the inclination to deal with the discrimination they encountered. For example, although it might appear that state insurance commissions are the appropriate avenues for redress of grievances against insurance companies, our study found that only 19 out of 58 individuals who reported experiencing discrimination knew of the existence of state insurance commissions. A recently published study found that state insurance commissioners were unaware of incidents of genetic discrimination²¹. Our results indicate that this lack of awareness arises not because genetic discrimination does not exist as was suggested by that study, but rather because consumers did not appeal to the insurance commissions.

Even individuals who were aware of regulatory agencies often did not avail themselves of opportunities to redress their grievances. Some of these individuals felt they had little hope of successfully challenging discrimination. In one case, an attorney who had been denied health insurance because of diagnosis of (asymptomatic) hemochromatosis, did not reapply for insurance or fight the denial because she "didn't want to be reminded" that she had a genetic diagnosis.

In order to increase our understanding of the causes of genetic discrimination and thus suggest strategies for minimizing discriminatory incidents, we attempted to determine whether instances of genetic discrimination occurred as a result of ignorance on the part of the discriminating institution or as a result of institutional policy. The study revealed instances of both causes of discrimination and, in some instances, the causes were difficult to distinguish. For example, in several reported cases involving a few specific companies, life insurance agents in a branch office were unaware of company policy that individuals with asymptomatic hemochromatosis should not be denied an insurance policy. This pattern suggests not only ignorance on the part of the agents, but may suggest that the company is not troubled by and may even condone such ignorance.

Other cases provide additional examples of ignorance giving rise to genetic discrimination. Representatives of an adoption agency reportedly thought that parents of children with MPS could not be healthy. Some teachers and school officials apparently believed that unaffected siblings of affected relatives must themselves be at-risk for becoming ill and in need of special treatment.

The single clear example of genetic discrimination due to institutional policy is that of the American Red Cross's refusal to accept blood donations from people with hemochromatosis. Because the standard treatment for hemochromatosis, phlebotomy,

is not covered by many health insurers, some individuals donate blood as an alternative to treatment. There is apparently no medical reason to restrict the use of this blood; the policy of the American Red Cross to refuse blood from hemochromatosis patients is based on the non-altruistic nature of the donation¹⁶.

The instances of differential treatment based on genetic tests described in this paper raise questions concerning the legality of this treatment. Under what circumstances is it legal to limit an individual's opportunities for employment, insurance, education, or for adopting a child on the basis of genetic information? Existing federal legislation such as the Rehabilitation Act of 1973 and the Americans with Disabilities Act of 1990 (ADA) provide that much differential treatment against those with disabilities is unlawful. The question of whether the ADA provides protective coverage for individuals who have abnormal genes but are asymptomatic or presymptomatic has been the subject of considerable debate²²⁻²³. Recent guidelines from the Equal Employment Opportunity Commission (EEOC) regarding the definition of "disability" under the ADA specifically address the issue of genotypically abnormal individuals who are asymptomatic or presymptomatic and states that those individuals are covered under the definition of disability in the ADA if they are regarded as disabled²⁴.

Although the ADA provides broad legal protections against genetic discrimination, it is apparent that, for the most part, its provisions do not apply to insurance underwriting^{6,7,18,24}. There is great concern that insurance companies are abandoning community rating, in which all people in a given geographical area pay the same premiums, in favor of underwriting, that is, setting premiums on the basis of an individual's risk. In principle, there is no need for underwriting since the rates set in community rating reflect the known incidence of morbidity and mortality in that geographical area. However, insurance companies are concerned that other companies will skim, i.e., insure only those individuals with the lowest risks. In addition, they fear that individuals who know that they are at risk for increased morbidity or mortality will buy an excessive amount of insurance¹⁸. Our study showed that this possibility is real; several individuals who were at-risk for Huntington disease reportedly attempted to buy life insurance or increase the amount of their coverage when they first learned of their at-risk status or when symptoms of the disease appeared. Given these pressures on insurance companies to resort to underwriting, it is likely that there will be increasing use by insurance companies of genetic as well as other sophisticated medical tests.

Since federal regulation of insurance companies is extremely limited, any meaningful restrictions of the use of genetic tests by insurance companies will need to arise from state legislation. States vary widely in their regulation of genetic testing by insurance companies^{18,25}. Some states have already enacted legislation prohibiting or limiting the use of genetic information in insurance underwriting. Although at this time there is little prospect of a universal health care policy in the United States, any health care policy will have to deal with the increasing use of genetic tests in medicine and by insurance companies.

This study of genetic discrimination is limited in several respects. The respondents in this study are members of genetic disease support groups who do not represent the entire population of affected individuals. They are also a self-selected subgroup of the

membership of those groups. In addition, documentation of discrimination was difficult to obtain in many instances. Some types of discrimination such as employment discrimination cannot be documented easily. In other instances, records of discrimination were not kept, especially if the individuals believed that there was no recourse for appeal of an adverse decision. Thus, it is difficult to determine to what extent reports of genetic discrimination are of actual rather than perceived discrimination.

The low response rate to the questionnaires is due to several factors besides the actual incidence of genetic discrimination. No follow-up mailings of the questionnaire were done, a practice which greatly increases the response rate. In addition, the mailing lists of the disease organizations include many individuals who are not personally affected by the disorders and so would not respond to the questionnaire. This is reflected in the fact that some questionnaires were returned by people who noted that they were not appropriate respondents. It is also possible that the response rate reflects a real result; that while genetic discrimination exists at this time it is not a widespread phenomenon.

Finally, we emphasize that this study is not a survey, but rather an attempt to collect case studies in order to examine the varieties of genetic discrimination. Consequently any statistical analysis of the cases would be both inappropriate and unnecessary.

This first extensive study of genetic discrimination extends and confirms the results of earlier ones. The cases from this study are consistent with the interpretation that although not systematic, genetic discrimination does occur in a wide variety of contexts and can cause hardship to affected individuals and their families. Many instances of genetic discrimination described in this study are similar to other types of discrimination. However, the distinctive nature of this type of discrimination lies in its effect on individuals who are asymptomatic and may never become symptomatic. Because the number and use of genetic tests is expanding rapidly and will continue to increase, it is vital that standards be developed in the near future to ensure that genetic information be used fairly. As our society struggles to be more equitable in its treatment of people regardless of race, age, or gender, it cannot ignore or justify inequities based on genotype.

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Table 1. Questionnaire

1. What is your genetic diagnosis?
- 2a. Do you think that you may have been refused social benefits or denied opportunities because of your diagnosed condition?
- 2b. If no, do you have any concerns for the future, or other comments?
3. In what year did the event/s occur?
4. Around what issue did you experience difficulties? (Health insurance, life insurance, adopting children, military, social services, church/synagogue, community/neighbors, other (please specify).
5. Please describe your experiences, explaining why these are discriminatory.
6. Are there any other comments that you would like to make?
7. Please note which institution sent you this questionnaire.
8. May we contact you for more information? (name, address, telephone)

The above questions were distributed in a questionnaire to individuals associated with genetic disease support groups who were likely to have a genetic diagnosis (see Methods, pp. 73-75). In addition to the questions listed above, the questionnaire had a brief definition of genetic discrimination and an assurance of confidentiality. A nearly identical questionnaire was distributed to individuals likely to be carriers for a genetic disorder.

Table 2. Partial List of Questions from the Telephone Guide Used for Interviews on Genetic Discrimination

Number of children/family members affected by the condition
Pedigree
Presence or absence of confounding disabilities
Context of the occurrence or the event
(Insurance, employment, public entities & accommodations/housing, education, government, community, other)

Insurance
Type? (Health, life, disability, automobile, home/mortgage, commercial loan)
Obtaining, renewing, or switching insurance?
Company name?

Employment
(Hiring, promotion, transfer, job responsibilities, compensation, eligibility for benefits, provision for disability, association with someone disabled, other)
Company name?
Employer/title?
Type of job and relevance of the condition to job performance
Was physical accessibility to an activity curtailed in any way?
Were reasonable accommodations requested? If so, were they provided?
After the incident, where did you work?
If you changed jobs, why?
Describe educational background and qualifications for job
Jobs held before and after (title/duties/length of time/ why left)

Public entities and accommodation
(Adoption agency, public housing, obtaining a loan, professional licensing, other licensing, transportation services, place of education, day care center, recreational facility, other)

Education
(Admission, activity restriction, termination, health service, other)

Government
(Military, benefits, social security entitlement, federal, state, local, other)

Military
(Entrance, transfer, job responsibilities, activity restrictions, termination, promotion, other)
Army, Navy, Air Force, Marines, National Guard, other

Benefits
(type: eligibility criteria and dates)

Community
(Neighborhood, religious community, recreational facilities)

Table 2 continued: Partial List of Questions from the Telephone Guide Used for Interviews on Genetic Discrimination

Details of Discrimination

- How was information about your condition revealed?
- Did the institution get information from the Medical Information Bureau? What information did they get? How did you find out?
- What did you request and why? (describe all events, contacts/correspondence which precede the institution's denial)
- Who did you first contact? (include job title)
- Type of correspondence (letters, phone, person)
- What, if any, additional medical information were you required to disclose? (May we receive a copy of their request?)
- Were you given a reason why this information was required?
- Did you voluntarily submit additional information or medical letters of support? (May we receive documentation?)
- Did you seek help from an outside source, such as personnel, other people you know, or a disease support group, etc.?
- What was the nature of their reply?
(refusal to consider case, request for additional information—if so, what was requested?)
- Were you or your physician requested to submit information? Other?
- Did the person making the decision explain to you what they thought and why? How was this communicated to you?
- How long did the institution take to respond to your initial inquiry?
- Was the response made in person? By phone? By letter? (Personalized form or letter?)
May we obtain a copy? If not, why?)
- Who replied? (job title)
- Did this person continue to handle your case, or was it referred to a supervisor? If so, how high up within the organization did consideration of your case go? Did you ever request that a supervisor take charge?

Table 3. Tabulation of questionnaires				
	Questionnaires (returned / sent)	Reporting genetic discrimination		
		yes	no	ambiguous
Huntington disease	623 / 25,924	276	329	18
Hemochromatosis	138 / 1,250	53	85	0
MPS	57 / 420	44	10	3
Phenylketonuria	22 / 200	12	8	2
Other*	77 / none	70	5	2
Total	917 / 27,790	455@	437	25

* This category includes questionnaires with information regarding disorders other than those listed above and those where it was not possible to ascertain the disorder the individual had. Questionnaires returned blank are not included.

@ The 206 interviewees were from the 385 individuals in this category reporting an association with Huntington disease, hemochromatosis, MPS, or phenylketonuria. See Methods (pp. 73-75) for a more detailed description of the selection of individuals interviewed.

Genetic Discrimination and Screening for Hemochromatosis

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INTRODUCTION

THE development of biochemical and molecular biological tests have enabled clinicians to diagnose various genetic diseases before symptoms appear. These diagnoses are based on redefinitions of the diseases. In these redefinitions, genetic diseases are characterized primarily by their underlying genotypes rather than, as is traditional, by the phenotypic manifestations of those genotypes and by other clinical symptoms and signs.

The ability to diagnose a disease in an asymptomatic or oligosymptomatic stage has several benefits. The treatment of a disease in its early stages may result in a more favorable prognosis and in reduced medical costs. If the disease is relatively common and potentially serious, the existence of a simple diagnostic test and an effective treatment may justify a general screening program to identify presymptomatic individuals.

Unfortunately, there are potential problems associated with both the redefinition of the disease and implementation of a screening program. One such problem is the increased likelihood of discrimination against an asymptomatic individual who is found to have an abnormal genotype and, as a result, considered to be affected according to the redefinition of the disease. This type of discrimination against an individual or members of the individual's family, that occurs as a consequence of knowledge about the genetic constitution of that individual, has been termed "genetic discrimination" (1).

In this paper we discuss genetic discrimination in the context of screening programs for hereditary hemochromatosis. We have chosen

to examine this condition for two reasons. First, in a recent paper appearing in *The New England Journal of Medicine*, Edwards and Kushner presented a persuasive argument for the implementation of screening programs for hemochromatosis (2). Second, a study we have been conducting of instances of genetic discrimination experienced by people with various genetic conditions has revealed a significant number of cases of discrimination against people with asymptomatic hemochromatosis.

We describe here 6 cases drawn from this study. These cases illustrate unjustifiable discrimination against individuals with asymptomatic hemochromatosis or against their relatives. We expect that the prevalence and importance of genetic discrimination associated with this condition will increase if mass screening or testing programs for hemochromatosis, which will identify a large number of people with asymptomatic hemochromatosis, are introduced without appropriate protections. Therefore, any proposal for a screening program for hemochromatosis should include an assessment of the impact of genetic discrimination and a protocol designed to mitigate its effects. As diagnostic tests for other genetic conditions become available, similar considerations should inform decisions concerning the introduction of screening programs for these conditions.

BACKGROUND

Hemochromatosis is a relatively common autosomal recessive iron storage disorder (3). It is estimated that the frequency of homozygosity (the presence of two, possibly different, disease associated alleles) in the U.S. is approximately 5 per 1000 (2). The gene responsible for the disease has recently been located on chromosome 6 (4). Some individuals with the genotype for hemochromatosis never develop clinical symptoms of the disease. Clinical symptoms, when present, are usually the result of excessive iron deposition in the liver, pancreatic islets, heart, and/or joints, and are manifest as various degrees of hepatic disease, diabetes mellitus, heart failure, and arthropathy. If untreated, the disease can be lethal. Despite the serious nature of the condition, hemochromatosis can be effectively controlled by a regimen of phlebotomies.

In recent years, there have been significant advances in the ability to detect the homozygous genotype for hemochromatosis in asymptomatic individuals (2). The result of a simple and inexpensive test,

transferrin saturation of more than 62 percent, is able to predict the homozygous genotype with an accuracy of better than 90 percent (5,6). In view of this test, Edwards and Kushner suggest that hemochromatosis be defined by the presence of homozygosity for the mutant allele(s) rather than by clinical symptoms or signs of the disease (2). Using this definition, hemochromatosis can be diagnosed by the saturated transferrin test before a patient has any clinical symptoms or signs.

Hemochromatosis fulfills the standard criteria outlined above for the introduction of a general screening program. It is common, potentially serious, a simple inexpensive diagnostic test is available, it has a long asymptomatic phase, and can be effectively treated. Screening for presymptomatic individuals who are homozygous for hemochromatosis should result in decreased medical costs and increased years of healthy life for those individuals who develop clinical hemochromatosis and are treated at an early stage of the disease.

Edwards and Kushner advocate including the test for hemochromatosis with blood tests already performed as a part of routine physical examinations (2). Because many people do not avail themselves of regular physical examinations, Edwards and Kushner believe that such testing for hemochromatosis should be supplemented with mass screening programs. They foresee that clinically normal homozygotes who have been tested or screened may encounter problems when preparing insurance applications. Nevertheless, they argue that since the genetic definition of hemochromatosis does not imply the presence of disease, healthy homozygotes will not be disqualified from health or other insurance. Edwards and Kushner include in the category of healthy homozygotes individuals whose hemochromatosis is controlled by phlebotomies since these individuals who have neither cirrhosis nor diabetes have no morbidity due to their condition and have normal life expectancies (2,7). We refer to these individuals as having asymptomatic or controlled hemochromatosis.

We believe that Edwards and Kushner have underestimated the risks of genetic discrimination associated with screening programs for hemochromatosis (8). In a pilot study of genetic discrimination using the case study approach, we found a significant number of cases showing unjustifiable discrimination against individuals and relatives of these individuals who are asymptomatic but are labeled with various genetic diseases (1). Our ongoing study has confirmed our pre-

vious results by uncovering many more cases of genetic discrimination. A detailed and complete discussion of the methodology and results of this study will be published separately (L. N. Geller et al., in preparation).

METHODS

Using a retrospective analysis of case histories, we examined discriminatory practices directed against individuals who have a genetic diagnosis and/or relatives of these individuals. Hemochromatosis was one of several conditions chosen for study because its genetic basis is known and unambiguous, discrimination directed against an individual with controlled hemochromatosis is likely to be due to the genetic basis of the condition rather than to its clinical manifestations, and because individuals with this condition could be contacted with the cooperation of the Hemochromatosis Research Foundation.

Questionnaires designed to determine whether affected individuals or members of their families experienced discrimination were sent to 1250 members of the Foundation. The completed questionnaires were evaluated and telephone interviews conducted with those people reporting incidents of genetic discrimination. The purposes of the interviews were to gain more information about the incident, to assess the validity of the report, and to obtain, when possible, documentation of the incident. The 6 cases reported here are a representative sample of the 38 individuals diagnosed with hemochromatosis who reported discrimination and were subsequently interviewed.

CASES OF GENETIC DISCRIMINATION

Case #1: Life Insurance

A 52 year old man with no health problems was diagnosed with hemochromatosis in 1986. For a period of two years he underwent routine phlebotomies and regular monitoring of his ferritin level. In 1988, after completing the regimen of phlebotomies, he sought to increase his life insurance policy from \$10,000 to \$20,000. The insurance company rejected his application, citing information in his medical records. In 1990, he reapplied to the same company and was rejected again for the same reason. On the advice of his insurance agent, who knew of his condition but believed that he should not have trouble obtaining life insurance, he applied to two other com-

panies. Both companies refused to insure him because of his medical history. Although none of the insurance companies would state the precise reason for the denials, the fact that he had no other risk factors suggests that the most likely reason was his diagnosis of hemochromatosis.

Case #2: Individual Health Insurance

An asymptomatic 25 year old man was diagnosed with hemochromatosis by means of blood tests after a family member developed the condition. He suffered no symptoms of organ damage as a result of the disorder. After his diagnosis he initially underwent weekly phlebotomies and was subsequently phlebotomized bi-monthly. After he became ineligible to continue receiving health insurance through his parents' health insurance policy, he applied for an individual policy since his place of employment did not offer health insurance. His application was rejected because of his hemochromatosis. He subsequently applied to other companies but was unable to obtain health insurance.

Case #3: Group Health Insurance

A 48 year old man was diagnosed in 1988 with hemochromatosis. He underwent weekly phlebotomies for six weeks and then bi-monthly phlebotomies for a period of approximately two years. Since completing this regimen, he has not required treatments. His wife was employed by a small company (five employees). During the Spring of 1993, the company attempted to obtain a group health insurance policy, engaging in negotiations with two large companies. Both insurance companies refused to offer a group policy for employees and their families which included his wife because he had been diagnosed with hemochromatosis.

Case #4: Disability Insurance

A 42 year old man was diagnosed with hemochromatosis during a routine physical examination. He underwent phlebotomies weekly for 40 weeks, then three or four times a year for one year. Since early 1992 he has not required phlebotomies. Currently, his hemochromatosis is controlled; he is asymptomatic and has suffered no organ damage. When a partner in his law firm suffered a brain tumor, the firm reevaluated its disability insurance policy and decided to in-

crease the coverage for its members. The insurance company refused to offer increased coverage to the attorney with hemochromatosis.

Case #5: Reimbursement for Treatment; Blood Donation Center

A 36 year old man was diagnosed with hemochromatosis and was treated by a regular schedule of phlebotomies. He remained asymptomatic while following this regimen. His health insurance company refused to pay for the phlebotomies despite the fact that his diagnosis occurred after he had been insured, arguing that his hemochromatosis was a preexisting condition. In order to obtain phlebotomies at no cost, he attempted to donate blood at a Red Cross blood bank. The Red Cross refused to allow him to donate blood because he was donating blood as a treatment and not as a gift.

Case #6: Employment

An asymptomatic 53 year old man had been diagnosed with hemochromatosis. His treatment consisted of phlebotomies at three month intervals. He applied for a position as an insurance agent and was accepted into the company's agent training program. In the course of an interview, he mentioned that he was being treated for hemochromatosis and agreed to submit a copy of his medical report based on a recent physical examination. A manager of the company told him that his illness might result in the company's inability to offer him medical benefits. However, the applicant was permitted to enter the training program with the expectation of a position when he completed it. After approximately five or six weeks of training, he was told that the home office of the company would not hire him because of his diagnosis of hemochromatosis. He was not paid for the weeks which he spent in training.

DISCUSSION

These six cases of genetic discrimination are representative of the types of discrimination reported by those with pre- or asymptomatic hemochromatosis. The majority of cases of genetic discrimination reported to us involve insurance. Most of the remaining cases, such as Case #6 presented here, involve employment. In most of the insurance cases we have obtained, as well as in the cases reported here, individuals whose hemochromatosis was controlled by phlebotomies and individuals who had previously undergone phlebotomies but no

longer required the treatments were denied life, health, and disability insurance (Cases #1, #2, and #4).

Case #3 is an example of discrimination involving group insurance. This case illustrates several issues. First, although it is commonly believed that every member of the group will be automatically insured, we found several cases like this one in which people experienced difficulty in obtaining and in retaining their health insurance when the number of people in their workplace was relatively small. Second, the individual experiencing discrimination might be the spouse (or, in other cases, a relative) of the affected individual, rather than the affected individual himself. Third, employees covered by a group policy who themselves or members of their families subsequently develop a genetic condition may lose their insurance. Given these realities, some individuals reported being unwilling to leave jobs that provide adequate and stable health benefits because they would not be able to obtain comparable benefits at the new place of employment.

Case #5 illustrates another common problem: health insurance companies have refused to pay for the phlebotomies required to treat hemochromatosis even though the condition was diagnosed after the person had obtained the insurance policy. These insurance companies seem to take the position that since hemochromatosis is genetic, the individual's condition was "preexisting."

There is essentially complete unanimity in the medical and actuarial literature that a person with hemochromatosis controlled by regular phlebotomy therapy who has neither iron-induced cirrhosis nor diabetes should enjoy a normal life expectancy. This view has been expressed in two recent review articles in medical journals (2,3) as well as in articles appearing in *The Journal of Insurance Medicine*, a journal read by medical directors of insurance companies who make decisions involving the actuarial risks of medical conditions (7,9,10). Nevertheless, our study clearly shows that discrimination against individuals with a diagnosis of hemochromatosis and against their relatives continues to be a significant problem.

We believe that in many of the cases involving insurance, the denial of insurance is based on company policy rather than on individual agents' ignorance of the medical literature. In the course of our study, one insurance company was cited repeatedly and several other companies were mentioned by at least two respondents. Although agents and individual underwriters may occasionally make decisions at vari-

ance with company policy, it seems unlikely that this situation would arise repeatedly.

Our data suggest that Edwards and Kushner's belief that people with controlled hemochromatosis will not encounter discrimination is unjustifiably optimistic. It is especially ironic that they state that screening for hemochromatosis will identify a large pool of healthy blood donors since we found nearly 20 people with controlled hemochromatosis who, like the man described in Case #5, were unable to donate blood at Red Cross centers. The Red Cross justifies its discrimination against people with hemochromatosis by arguing that "donors motivated by other than altruistic reasons might more readily misrepresent, even unconsciously, important historical data" (11).

The designation by some insurance companies of hemochromatosis as a "preexisting condition" even if it is diagnosed long after the insured individual purchased the policy indicates that hemochromatosis has already been redefined on the basis of its genetic etiology. For nongenetic diseases, preexisting has always meant that the disease was manifest, i.e., current symptoms and signs of the disease are documented in the medical record prior to the application for an insurance policy. It seems unlikely that a nongenetic cancer or a degenerative disease like arthritis would be labeled a preexisting condition even though the processes leading to the symptoms of the disease also begin long before a clinical diagnosis of these diseases can be made.

The introduction of mass screening or large scale testing for hemochromatosis based on the identification of the genotype underlying the disease will have the effect of placing a huge number of asymptomatic individuals at risk for discrimination. We have introduced the term "asymptomatic ill" to describe people who are clinically normal but, as the result of medical tests, have been labeled diseased (1). The number of such "asymptomatic ill" individuals can be estimated from the sum of the number of individuals that have the genotype but do not become ill and the number of individuals who do not have the genotype at all but nevertheless show a positive saturated transferrin test result.

The estimation proceeds as follows: First, approximately 3.4% of those individuals who are heterozygous for hemochromatosis will have a positive saturated transferrin test (false positive) (5). As a result of this high frequency of false positives, all individuals with a positive test will undergo additional testing which may even include

instituting a regimen of phlebotomies while monitoring serum ferritin concentration (2). Consequently, the medical records of individuals who do not even have the genotype for hemochromatosis may document treatment for the disease.

Second, hemochromatosis is variable in expressivity, i.e., not all those individuals with the genotype will develop clinical manifestations of the disease. It has been estimated that the prevalence of homozygosity for hemochromatosis in the white population in the United States is approximately 5 per 1000, while the estimated frequency of phenotypic hemochromatosis ranges from 0.5 to 3.7 per 1000 (2). Thus, at least 25% of those who are homozygous for hemochromatosis do not develop symptoms of the disease.

To estimate the number of people who will have a positive saturated transferrin test but will not develop the disease, we add the number of people with false positive test results and the number of people with true positive test results but who will never develop symptoms of the disease. If the actual frequency of homozygosity for hemochromatosis is 0.005, then, assuming Hardy-Weinberg equilibrium, the frequency of heterozygotes will be 131 per thousand. Of these, 3.4 per cent, or approximately 4500 per million will test positive but will not have the genotype for hemochromatosis. In addition, 1250 per million (25% of the 5000 per million) who are actually homozygous for hemochromatosis will never develop clinical manifestations of the disease. Since the test is 92% sensitive (5), approximately 1100 per million of these people will have positive test results. Thus, 5600 (4500+1100) people out of every million people tested will have the record of the positive saturated transferrin test in their medical files but will not develop clinical hemochromatosis. In a general screening program of 100 million people (less than one-half of the U.S. population), even if only 5% of these people encounter discrimination, this amounts to approximately 28,000 people. Given the practices of insurers and employers demonstrated by our study, it seems likely that this is not an overestimate of the number of people who will never develop phenotypic hemochromatosis either because of a false positive test result or because of the variable expressivity of the disease but who, nevertheless, will experience genetic discrimination. As more tests for genetic conditions are developed to diagnose the "asymptomatic ill," the number of people at risk for genetic discrimination can only increase.

It must be emphasized that the concerns described here regarding mass screening or testing for hemochromatosis apply to numerous other genetic conditions for which presymptomatic testing exists. Further, given the rapid advances in the development of genetic tests and the pressures on insurance companies and employers to utilize them, an increasing number of people are in jeopardy of suffering some form of genetic discrimination. Fragile X syndrome, cystic fibrosis, and Gaucher disease are but three examples of conditions for which there is significant interest in implementing mass screening programs (12). Programs for each of these conditions will subject a group of genetically abnormal people who are clinically well to the risk of genetic discrimination.

The "asymptomatic ill" are regarded as being disabled by those who discriminate against them. Consequently, we believe that in many cases these people can challenge the discrimination using the Americans with Disabilities Act of 1990 (ADA) (13). The ADA prohibits most forms of discrimination against people with disabilities and specifically prohibits discrimination against people who, although they have no actual disability, are regarded as being disabled. Thus, in our opinion, the ADA prohibits most institutions from discriminating against asymptomatic and presymptomatic people because of a genetic diagnosis (14,15). It should be pointed out, however, that the Equal Employment Opportunity Commission (EEOC) and the Department of Justice have yet to issue specific guidelines resolving this issue.

For the purpose of analyzing the legality of the majority of cases of genetic discrimination other than those involving insurance, the relevant provisions are found in Titles I, II, and III of the ADA. Title I prohibits discrimination in employment, including employer-provided benefits; Title II prohibits discrimination by state and local governments and their agencies; Title III prohibits discrimination in public accommodations, including commercial establishments and service providers. The ADA does allow discrimination if the individual poses a "direct threat to the health and safety" of other individuals [Sections 103(b) and 302(b)(3)]. However, in our opinion, this standard is not met in either Cases #5 or #6. Thus, we believe that the discrimination by blood donation centers described in Case #5 is prohibited by Title III and that the discrimination encountered in employment of the type exhibited in Case #6 is prohibited by Title I of the ADA.

Although the ADA provides far more limited protection in the area of discrimination by insurers, it does prohibit discrimination inconsistent with state law (Title V, Section 501(c)(1)) or not "based on sound actuarial data" (56 Fed. Reg. 35563 (July 26, 1991)). An insurer's denial of life insurance or medical insurance to asymptomatic individuals with hemochromatosis, as exemplified in Cases #1-4, might well violate provisions of the ADA.

CONCLUSIONS

There is no rational reason or justification for discriminating against individuals with controlled hemochromatosis. But, on purely rational grounds, there should be no discrimination on the basis of race or sex. Because we do not live in an ideal world, proponents of genetic screening and testing programs should not only consider the health and economic benefits of the program but should also take into account the potential harm resulting from prejudice and ignorance.

Our studies have shown that people with asymptomatic hemochromatosis are at risk for discrimination. The redefinition of the disease on the basis of genotype coupled with the institution of mass screening or large scale testing programs will create an increasingly large number of "asymptomatic ill" individuals, i.e., individuals who are healthy but who have been diagnosed with hemochromatosis and, consequently, will be at risk for discrimination.

Every person labeled genetically ill risks discrimination in his or her interactions with insurance companies, employers, or any other institution of society that has access to medical records. A health insurance company will know that a homozygous individual is at increased risk for the disease. The company might either exclude coverage for treatment when the disease becomes manifest by calling it a preexisting condition or deny the individual a policy, fearing that the person who develops hemochromatosis might not comply with the treatment regimen and suffer a general deterioration of health. Employers may not be willing to hire an applicant who is at risk for developing a genetic disease in view of potential health care costs and the possibility of diminished productivity. Some individuals will be stigmatized by relatives, friends, and members of their community for having an abnormal genotype, even when asymptomatic. These and other forms of discrimination against people diagnosed with hemochromatosis seem especially unjustifiable since, as noted above,

many of the individuals at risk for hemochromatosis will never develop the disease and those who do develop the disease but are treated are not expected to suffer any ill effects.

We are not categorically opposed to genetic screening or testing programs, including screening or testing for hemochromatosis (16). Indeed, programs that are well designed and carefully implemented can provide significant benefits to individuals and society with minimal adverse consequences. However, before instituting any screening program, we believe that it is essential to evaluate the likelihood and effect of harmful discrimination that may result from that program. To mitigate the harm caused by such discrimination, the design of the program should be optimized to minimize the number of discriminatory incidents. Participation in any testing or screening program should be strictly voluntary and informed consent should be required before testing. Genetic counseling should be provided to explain the implications of the test results. In addition, it is crucial that the results or even the existence of the test results be divulged only to the patient or to parties designated by the patient. On a more fundamental level, in our opinion, the most important single measure that could be taken to eliminate genetic discrimination in health insurance would be the implementation of an equitable universal health care program.

Even if the greatest care is exercised in the design of a screening program, occurrences and the resulting effects of discrimination probably cannot be completely avoided. Thus, it is especially important that everyone associated with screening programs, including the clients, clinicians, and administrators of the program, be made aware of the possibility of discrimination, of applicable anti-discrimination legislation, and of the limitations of such legislation.

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ABSTRACT

Recent advances in tests for the genotype for hemochromatosis and suggestions that the tests be used in mass screening programs for the disease raise the possibility of a large increase in the incidence of discrimination against

people who are found to be homozygous for hemochromatosis. This paper presents cases of genetic discrimination drawn from a study of discrimination against people with a variety of genetic conditions. The cases discussed here involve employment and several types of insurance discrimination against people diagnosed with hemochromatosis who either are currently asymptomatic or whose condition is controlled by means of phlebotomies. There is no justification for these types of discrimination since people with controlled hemochromatosis suffer no excess mortality or morbidity. Our study suggests that genetic discrimination is already a serious problem and that any proposed screening program for hemochromatosis or other genetic condition must consider and attempt to mitigate its effects.

Genetic Discrimination: Perspectives of Consumers

E. Virginia Lapham,* Chahira Kozma, Joan O. Weiss

In a study of the perceptions of 332 members of genetic support groups with one or more of 101 different genetic disorders in the family, it was found that as a result of a genetic disorder 25 percent of the respondents or affected family members believed they were refused life insurance, 22 percent believed they were refused health insurance, and 13 percent believed they were denied or let go from a job. Fear of genetic discrimination resulted in 9 percent of respondents or family members refusing to be tested for genetic conditions, 18 percent not revealing genetic information to insurers, and 17 percent not revealing information to employers. The level of perceived discrimination points to the need for more information to determine the extent and scope of the problem.

The rapid advances in human genetics, largely fueled by the Human Genome Project (HGP), have resulted in the expansion of the number and range of genetic tests (1). These tests are capable of providing carrier and presymptomatic information including risk of future disease, disability, and early death. In addition, these tests may reveal genetic information not only about the health of the individual, but also about his or her family members (2).

Concern about access to genetic information by health insurers has historical support (3, 4). In the early 1970s, several insurance companies discriminated against individuals who were carriers of sickle cell anemia, even though they were quite healthy (5). The use of genetic information to deny life insurance to individuals leaves their dependents more vulnerable to economic consequences than is the case with the 70% of adults who are covered (6). The use of genetic screening to identify workers who may be particularly sensitive to noxious environments has been the principal focus of concern about workplace genetic testing even when done for benevolent reasons (7). Issues of genetic discrimination in employment and insurance have become more urgent as a result of the genome project (8).

Overall concerns about privacy and confidentiality have led the Ethical, Legal, and Social Issues (ELSI) Branch of the National Center for Human Genome Research to identify this issue as a top priority with the goal of proposing legislation specifically designed to protect people against genetic discrimination (9). Additionally, several working groups and scholars are focusing on

this issue and have developed background papers and policy recommendations about the use of genetic information in health insurance as well as other areas such as life insurance and employment (10, 11). Despite these concerns about potential genetic discrimination and documentation of individual cases, there is little information about the incidence and range of the problem (12).

This report provides information on the experiences of 332 individuals with one or more family members with a genetic disorder who are affiliated with genetic support groups. The study was part of the Human Genome Education Model (HuGEM) Project of the Georgetown University Child Development Center and the Alliance of Genetic Support Groups. It was the first phase of the HuGEM Project with the aim of getting input from 300 consumers in order to develop, implement, and evaluate a collaborative education model for consumers and health care providers.

Participants were recruited primarily through the national, regional, and local genetic support groups affiliated with the Alliance of Genetic Support Groups. Notices were put in two issues of the monthly *Alliance Alert* and letters were sent to the directors of 101 genetic support groups (representing an estimated 585,800 members). The notices contained information about the study and requested volunteers that were at least 18 years old and with one or more persons in the family with a genetic disorder who would be willing to participate in a 30-min telephone interview to provide opinions on the ethical, legal, and social issues of the HGP as well as priority topics for education. Volunteers were assured confidentiality of their responses. Random sampling was considered and ruled out because of time, cost, and the primarily educational focus of the project. Thus, the findings are applicable only to this group. Support group leaders were requested to distribute the let-

ter requesting volunteers at meetings and in newsletters. Persons interested in participating were to complete a form at the bottom of the letter or call a 1-800 number for more information.

As a result of information provided by the support groups to the members or through the *Alliance Alert*, a group of 483 persons (from 73 different groups) contacted the Alliance of Genetic Support Groups about the study. They were sent information about the study and about the Human Genome Project. Of these, 336 (70%) returned consent forms (13). From this group, four persons decided not to participate after the interviews started, 306 persons completed telephone interviews, and 26 requested and completed the questionnaire by mail, for a total return of 332 respondents from 44 states and the District of Columbia (14).

Respondents were primarily female, highly educated, married, and Caucasian (15)—characteristics believed to be typical of genetic support groups (16). Age categories ranged from the twenties through seventies with a median age in the forties. A range of religious preferences was reported (17). There was an average of 2.1 affected family members per respondent with a range of 1 to 12 affected members reported.

The study questionnaire was composed primarily of questions with multiple choice responses. Telephone interviews were conducted by four social workers, a genetic counselor, and a consumer administrator (18) and lasted an average of 40 min with a range of 29 to 90 min. The content covered five areas: demographic information; knowledge of the Human Genome Project (61% had heard about the HGP before volunteering to participate in the study, 74% considered the HGP very important to their families, and 81% considered it very important to society); personal and family experience in areas related to genetic testing and research; opinions on a range of ethical, legal, and social issues; and priority topics for education. The education priorities were used to develop and implement educational forums in the mid-Atlantic and Pacific Northwest regions and will be described elsewhere.

Respondents were asked whether they or other family members had encountered problems with health insurance, life insurance, and employment (19). The term "genetic discrimination" was not used in the survey. It is used in this report to describe prejudicial actions as perceived by the respondents that resulted from insurers' or employers' knowledge of an individual's genetic condition, carrier status, or presumed carrier status, based on observation, family history, genetic testing, or other means of gathering genetic information (20).

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Respondents reported 101 different primary genetic disorders. The 18% of families with two or more disorders were asked to select one for purposes of the study. Of the primary disorders 68% were single-gene disorders, 10% were chromosome disorders, 11% were multifactorial disorders, 11% were major malformation syndromes, and less than 1% were mitochondrial and endocrine diseases.

Data analysis included frequency responses and comparison of responses to the questions on genetic discrimination by education, religious preference, and health of respondent and they showed no statistically significant differences (Pearson value of $P < 0.05$ was considered significant). Gender and ethnicity showed no significant differences when controlled for sample size.

Consumer experiences with health insurers were deemed important because the availability of affordable health insurance

often determines who does and who does not have access to health care (4). For many people with genetic disorders, health insurance may mean the difference between life and death (21).

Although considerable genetic information may already be available to insurers in medical records, 40% of the respondents recalled being specifically asked about genetic diseases or disabilities on their applications for health insurance (Table 1). It cannot be assumed that the remaining 60% had not been asked questions about genetic diseases and disabilities. Many of them volunteered the information that they had never applied for health insurance. Some were able to maintain the coverage they had prior to diagnosis of a genetic disorder. Others had not applied because they assumed the genetic condition in the family would result in being turned down. Whether or not this information was then used to

deny insurance to these people based on their genetic condition is not known.

Twenty-two percent of the respondents (Table 1) said that they or a family member were refused health insurance as a result of the genetic condition in the family. Since insurers do not need to provide reasons for turning down applications, it might be argued that respondents may have subjectively assumed that the denials were made because of the genetic condition. In this study, however, 83% of those who were refused health insurance had also been asked about genetic diseases or disabilities on their applications. Looked at in another way, nearly half (47%) of those who were asked about genetic diseases or disabilities on an application for health insurance were subsequently turned down. As health and life insurers are primarily regulated by states and most states are just beginning to address genetic issues in legislation (22), it is not known how many insurers actually ask genetic questions on applications.

The 31% of respondents with health insurance coverage who were denied reimbursement for some service or treatment indicated reasons such as the treatments were considered experimental, and services such as physical or occupational therapy were not considered a medical necessity. Time limits for submitting claims were also an issue, with insurers not paying claims that were more than a year old even when they had been submitted within the year and returned for more information. In several instances, payment was denied even though preapproval for a treatment or service had been given.

The large majority (83%) of respondents (Table 2) said they would not want their insurers to know if they were tested and found to be at high risk for a genetic disorder. The rate decreased to 78% when a similar question was asked that added the condition, "if the insurer pays for the tests." Some of the respondents noted that they would pay for genetic tests themselves or not be tested if they wanted to keep their genetic information confidential. The fear of genetic discrimination, as shown in Table 3, resulted in 9% of the respondents or a family member refusing to be tested for a genetic condition. This fear eliminates the opportunities of individuals to learn that they are not at increased risk for the genetic disorder in the family or to make life-style changes to reduce the risks or seriousness of the condition. It may also affect the number of people willing to participate in scientific research (10). Fear also prevented 18% of the respondents from revealing genetic information to an insurance company.

Approximately 70% of adults in the United States have some form of life insurance

Table 1. Questions and responses about experiences of consumers in areas of health insurance, life insurance, and employment. The total number of respondents is 332.

Question	Responses (%)		
	Yes	No	Don't know
As a result of the genetic condition in your family, have you or a member of your family been—			
Asked questions about genetic diseases or disabilities on an application for health insurance?	40	55	5
Refused health insurance?	22	76	2
Refused insurance coverage of some service or treatment?	31	67	2
Refused life insurance?	25	70	6
Asked questions about genetic diseases or disabilities on a job application?	15	83	2
Denied a job or let go from a job?	13	85	2

Table 2. Questions and responses to opinions about genetic information in insurance and employment.

Question	Responses (%)		
	Strongly agree or agree	Disagree or strongly disagree	Not sure or don't know
Genetic testing should be part of pre-employment physical exams.	4	94	2
Health insurers should be able to get genetic information if they pay for the tests.	16	78	6
If you were tested and found to be at high risk for a genetic disorder with serious complications, which of the following would you want to know the results of the test?	Yes	No	Not sure
a. Your employers?	6	87	7
b. Your insurance company?	11	83	6

ance (23). It is widely available, and only 3% of those who apply for coverage are declined. Of the 97% accepted, 5% are required to pay higher than standard premiums (24). This may be compared to the respondents in this study in which 25% (Table 1) of the respondents or affected family member have been refused life insurance (25).

Two questions were asked about the employment experiences of the study population. As noted in Table 1, 15% of the respondents said that they or affected family members had been asked questions about genetic diseases or disabilities on job applications. This increased to 20% of affected respondents ($P = 0.006$). It is not clear how often this information was used to subsequently deny the job to the applicants but the possibility exists and was of concern to respondents. In this study, 87% of respondents (Table 2) would not want their employers to know if they were tested and found to be at high risk for a genetic disorder with serious complications.

Thirteen percent of all respondents (Table 1) reported that they or another family member had been denied a job or let go from a job because of the genetic condition in the family. This was true for 21% of affected respondents and 4% of unaffected respondents ($P = 0.00001$). The percent was reduced to 9% ($P = 0.006$) for those with an affected child, even though a higher proportion of these respondents were in the workforce than the total population.

During the course of the analysis, a question was raised as to whether the perceived problems encountered in job application or denial or dismissal emanated from an employer's perception of a visible disability. To approach this question, analysis was done for the 77 unaffected respondents whose only affected family member was a child of less than 16 years of age. It was found that 7% of this population was asked about genetic diseases or disabilities on a job application and 3% were denied or let go from a job. These numbers should only be used as

a starting point for future analyses.

For the affected respondents, some specific examples highlight the kinds of problems experienced. A man with a sex chromosome disorder reported that he had been denied a job following a pre-employment physical exam after the doctor wrote the name of the possible disorder on his medical report. The employer, in this case, knew it was illegal to use the diagnosis in the hiring decision and told the applicant that he would deny the conversation in the future if asked. A woman with a skeletal disorder reported that she was given termination notice the day after she informed her employer of a genetic diagnosis. The notice was withdrawn after she sought legal counsel. Examples provided by other respondents focused on effects of the genetic condition that could come under the protection of the 1995 interpretations of the Americans with Disabilities Act (26). The dilemma for persons with genetic disorders is that they must show not only that they have a genetic defect but also that they were regarded as "disabled" by an employer and discriminated against because of that perception. This raises concerns about the privacy and confidentiality of genetic information in the workplace.

A total of 17% have not revealed genetic information to their employers (Table 3) for fear of losing their jobs or insurance coverage. This increased to 25% of affected respondents ($P = 0.00001$). Overall, 43% of the respondents reported that they or members of their family have experienced genetic discrimination in one or more of the three areas. This included health insurance only (9%), life insurance only (11%), employment only (6%), and more than one category (17%).

Additional studies of persons with genetic disorders are indicated to confirm or deny the perceptions of the consumers in this study. It is possible that members of genetic support groups who have experienced genetic discrimination may have been more motivated to volunteer for this

study. On the other hand, persons with these resources of higher education and membership in support groups traditionally have the skills and means to work with and influence social systems and may have experienced less discrimination than other groups. With adequate funding, a random sampling of respondents from support group or clinic populations could be selected with probability methods and objective as well as subjective information could be gathered.

Another goal would be to design more detailed questions to elicit information on genetic discrimination from respondents. Distinctions between the implications of overt genetic disease and conditions on each person and the effects on unaffected family members, or persons who are carriers or do not overtly express the consequences of the genetic condition will require further study. Consumers may be willing to participate if confidentiality is assured and trust is established. In this study, it was also found important for the interviewers to have clinical as well as technical skills in interviewing to facilitate the comfort level of discussing sensitive issues. This would also be recommended for future studies.

Although the goal of the HGP (and other genetic testing and research) is to help people, it could also cause harm if the level of perceived discrimination is in fact true. Neither the authors nor the respondents (as indicated in earlier responses) are suggesting that the HGP should not continue. On the contrary, there is strong support to continue research and to find ways to deal with genetic discrimination including federal or state legislation, guidelines, and standards among insurers, employers, researchers, and health professionals, and citizen advocacy to establish protections.

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Table 3. Percentage of respondents withholding information or refusing to be tested for a genetic condition as result of fear.

Question	Responses (%)		
	Yes	No	Don't know
As a result of a genetic condition, have you or a member of your family—			
Refused to be tested for a genetic condition for fear of your insurance coverage being dropped.	9	89	2
Not revealed genetic information to an insurance company.	18	79	3
Not revealed genetic information to an employer.	17	81	2

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11. *Genetic Information and Health Insurance: Report of the Task Force on Genetic Information and Insurance* (National Institutes of Health—Department of Energy Working Group on the Ethical, Legal, and Social Implications of Human Genome Research, 10 May 1993).
12. See, for example, P. R. Billings *et al.*, *Am. J. Hum. Genet.* 50, 476 (1992).
13. As the interviews progressed and it was apparent that few members of ethnic or racial minorities were volunteering, additional telephone outreach was made to support groups to increase awareness of the project and opportunities for volunteering. This was only partially successful as seen in the demographic data.
14. There were no statistically significant differences in the responses on the telephone and written interviews so they are reported here together.
15. Of the 332 respondents, 80% were female, 57% have at least a bachelors degree, and 90% were Caucasian. Other characteristics were: 75% were married and living with their spouses, 76% have children, and 63% work outside the home. Family relationship, whether or not the respondent or family members were affected, age of diagnosis, and current age were also recorded.
16. Precise data on the demographics of genetic support groups are not available. Impressions are from staff of the Alliance of Genetic Support Groups based on their conversations and communications with the member organizations and attendance at national, regional, and local meetings.
17. Religious preferences were Roman Catholic, 26%; Protestant, 41%; Christian-other, 9%; Jewish, 11%; other, 2%; and none, 12%.
18. The interviewers were trained in interview techniques by the principal investigator and participated in pre-testing the questionnaire.
19. The questions on possible genetic discrimination were taken from a questionnaire developed by Dr. Dorothy C. Wertz, The Shriver Center, Waltham, MA, entitled, *Ethical Issues in Genetics, Part 1*, p. 33, No. 34, and used with permission of Dr. Wertz (letter of 16 December 1993).
20. This definition of genetic discrimination differs from the one used by Billings *et al.* (12) as they did not include actions against persons who were symptomatic or visibly affected by their genetic disorders. The design of our questionnaire does not permit analysis according to the definition of Billings *et al.* (1993). Because the questions on discrimination ask about all family members at once, the questions do not distinguish among: (i) the direct consequences of ongoing genetic disease or conditions, (ii) the effect of genetic disease on other family members, and (iii) the consequences of genetic information gained through testing.
21. M. J. Ellis Kahn, in a video by the HuGEM Project, *An Overview of the Human Genome Project and Its Ethical, Legal, and Social Issues* (Georgetown University Medical Center, Washington, DC, 1995).
22. K. H. Rothenberg, *J. Law Med. Ethics* 23, 313 (1995).
23. The Ad Hoc Committee on Genetic Testing/Insurance Issues, *Am. J. Hum. Genet.* 56, 328 (1995).
24. "Report of the ACLI-HIAA Task Force on Genetic Testing," The American Council of Life Insurance and The Health Insurance Association of America (1991).
25. Many respondents said they had never applied for life insurance because they assumed they would be turned down.
26. Since 1990, the Americans with Disabilities Act (ADA) has provided protection for persons with disabilities in the workplace. The ADA prevents employers from openly denying employment or firing an individual solely on the basis of a "disability" if there are "reasonable accommodations" that can be made in the work setting to allow the person to perform his or her job. In April 1995, the ADA was interpreted by the U.S. Equal Employment Opportunity Commission to include healthy people who are carriers of genetic disorders. Implementation in general relies on employers and employees knowing and

similarly interpreting the law as well as having good faith efforts to comply.

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Kap104p: A Karyopherin Involved in the Nuclear Transport of Messenger RNA Binding Proteins

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A cytosolic yeast karyopherin, Kap104p, was isolated and shown to function in the nuclear import of a specific class of proteins. The protein bound directly to repeat-containing nucleoporins and to a cytosolic pool of two nuclear messenger RNA (mRNA) binding proteins, Nab2p and Nab4p. Depletion of Kap104p resulted in a rapid shift of Nab2p from the nucleus to the cytoplasm without affecting the localization of other nuclear proteins tested. This finding suggests that the major function of Kap104p lies in returning mRNA binding proteins to the nucleus after mRNA export.

Transport across the nuclear envelope occurs through nuclear pore complexes (NPCs) and is governed by the interaction of soluble transport proteins (karyopherins) with the transport substrate and the NPC (1–12). Most of our understanding of the mechanism of translocation comes from studying protein import in semipermeabilized cells (1) of model karyophilic proteins that carry a nuclear localization signal (NLS) from either the SV40 large T antigen or nucleoplasmin (2). These classical NLSs are recognized by karyopherin α in a dimeric cytosolic complex with karyopherin β (3–8). The complex docks at the NPC through its interaction with nucleoporins that contain characteristic repeated peptide motifs (6–11). The small guanosine triphosphatase, Ran, and p10 are required for the subsequent translocation of the substrate (and karyopherin α) through the NPC (11, 12).

Distinct saturable and noncompeting pathways for the import of different karyophilic proteins have been uncovered through the use of microinjection studies in oocytes (13–15). Similarly, saturable noncompeting pathways exist for the export of macromolecules from the nucleus (14, 16, 17). The signals that mediate many of these processes are different from classical NLSs (14, 15, 17–19) and thus may use recognition factors other than karyopherin α and karyopherin β for nuclear transport. Here we characterize the first such factor, which we

term Kap104p and which is required for the import of at least two yeast nuclear mRNA binding proteins.

The *Saccharomyces cerevisiae* proteins Kap60p and Kap95p are homologs of mammalian karyopherin α and karyopherin β (20). Sequence comparisons of Kap95p with the complete yeast genome database uncovered three additional proteins that are structurally similar to Kap95p; two of these, which we term Kap123p and Kap104p, have not been previously characterized (21), and the third, Pse1p, was identified as a multicopy enhancer of protein secretion (22). The sequence alignment of Kap104p with Kap95p is shown (Fig. 1A). The proteins bear substantial similarity over their entire lengths, and secondary structural predictions suggest that Kap95p and Kap104p share the same overall domain structure of HEAT motifs (23).

Deletion of *KAP104* resulted in a severe growth defect and temperature sensitivity (24). Immunofluorescence microscopy (25) with antibodies specific for Kap104p (in wild-type cells) showed that Kap104p was mainly cytosolic and was apparently absent from the nucleus (Fig. 1B). However, in *nup120 Δ* cells, which cluster their NPCs to a region of the nuclear envelope opposite the nucleolus (26), Kap104p colocalized with the nucleoporin Nsp1p (27) (Fig. 1C). The ability to detect coincident staining of the nucleoporins and Kap104p under these conditions likely was due to an interaction of Kap104p with NPCs.

Subcellular fractionation (28) was consistent with the distribution of Kap104p detected by immunofluorescence. Kap104p was present mainly in the cytosolic fraction,

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