DEPARTMENT OF HEALTH AND HUMAN SERVICES

SECRETARY'S ADVISORY COMMITTEE
ON GENETICS, HEALTH, AND SOCIETY

Second Meeting

Thursday,
October 23, 2003

Monet Rooms 1 and 2
Loews L'Enfant Plaza Hotel
480 L'Enfant Plaza East, S.W.
Washington, D.C.

IN ATTENDANCE:

Chair

Edward R.B. McCabe, M.D., Ph.D.
Professor and Executive Chair
Department of Pediatrics
David Geffen School of Medicine at UCLA
Physician-in-Chief
Mattel Children's Hospital at UCLA
10833 Le Conte Avenue, 22-412 MDCC
Los Angeles, CA 90095

Members

Cynthia E. Berry, J.D.
Partner
Powell Goldstein Frazer & Murphy
1001 Pennsylvania Avenue, N.W., 6th Floor
Washington, D.C. 20004-2582

Barbara Willis Harrison, M.S.
Certified Genetic Counselor and Instructor
Division of Medical Genetics
Department of Pediatrics
Howard University College of Medicine
520 W Street, N.W.
Washington, D.C. 20059

C. Christopher Hook, M.D.
Director of Ethics Education
Mayo Graduate School of Medicine
Assistant Professor of Medicine
Mayo Medical School
200 First Street, S.W.
Rochester, MN 55905

Debra G.B. Leonard, M.D., Ph.D.
Associate Professor of Pathology and Laboratory Medicine
Director, Molecular Pathology Laboratory
Hospital of the University of Pennsylvania
3400 Spruce Street
Philadelphia, PA 19104

IN ATTENDANCE:

Brad Margus
Co-Founder and Volunteer President
A-T Children's Project
Co-Founder and CEO
Perlegen Sciences, Inc.
2021 Stierlin Court
Mountain View, CA 94043

Reed V. Tuckson, M.D.
Senior Vice President
Consumer Health and Medical Care Advancement
UnitedHealth Group
9900 Bren Road East
Minnetonka, MN 55343

Huntington F. Willard, Ph.D.
Director
Institute of Genome Sciences and Policy
Vice Chancellor for Genome Sciences
Duke University Medical Center
Genome Sciences Research Building II
103 Research Drive, Room 4011
Durham, NC 27710

Emily S. Winn-Deen, Ph.D.
Senior Director for Genomics Business
Roche Molecular Systems
4300 Hacienda Drive
Pleasanton, CA 94588

Kimberly S. Zellmer, J.D.
2525 Tomahawk Road
Mission Hills, KS 66208

IN ATTENDANCE:

Ex Officio Members

DEPARTMENT OF COMMERCE

Arden L. Bement, Jr., Ph.D.
Director
National Institute of Standards and Technology
100 Bureau Drive, MS 1000
Gaithersburg, MD 20889

DEPARTMENT OF DEFENSE

Colonel Martha Turner, USAF NC, Ph.D.
USAF Surgeon General's Consultant for Medical Ethics
U.S. Department of Defense
Preventive Medicine and Biometrics
International Health Specialist Program
Uniformed Services University
4301 Jones Bridge Road
Bethesda, MD 20814

DEPARTMENT OF ENERGY

Lisa Devon Streit
Senior Advisor to the Director
Office of Science
U.S. Department of Energy
1000 Independence Avenue, S.W.
Washington, D.C. 20585

DEPARTMENT OF HEALTH AND HUMAN SERVICES

Administration for Children and Families

Martin Dannenfelser
Deputy Assistant Secretary for Policy and External Affairs
Administration for Children and Families
370 L'Enfant Promenade, S.W., Suite 600-W
Washington, D.C. 20447

Agency for Healthcare Research and Quality

Kaytura Felix-Aaron, M.D.
Senior Advisor, Minority Health
Office of Priority Populations Research
Agency for Healthcare Research and Quality
2101 East Jefferson Street, Suite 602
Rockville, MD 20852

IN ATTENDANCE:

Centers for Disease Control and Prevention

Muin J. Khoury, M.D., Ph.D.
Director
Office of Genomics and Disease Prevention
Centers for Disease Control and Prevention
1800 Clifton Road, MS E-82
Atlanta, GA 30339

Centers for Medicare and Medicaid Services

Judith A. Yost, M.A., M.T.
Director
Division of Laboratories and Acute Care
Centers for Medicare and Medicaid Services
7500 Security Boulevard
Baltimore, MD 21244-1850

Food and Drug Administration

David W. Feigal, Jr., M.D., M.P.H.
Director
Center for Devices and Radiological Health
Food and Drug Administration
9200 Corporate Boulevard, MSC HFZ-1
Rockville, MD 20850

Health Resources and Services Administration

Sam Shekar, M.D., M.P.H.
Assistant Surgeon General
Associate Administrator
Bureau of Primary Care
Health Resources and Services Administration
4350 East-West Highway, 11th Floor
Bethesda, MD 20814

National Institutes of Health

Francis S. Collins, M.D., Ph.D.
Director
National Human Genome Research Institute
National Institutes of Health
Building 31, Room 4B09
31 Center Drive, MSC 2152
Bethesda, MD 20982

IN ATTENDANCE:

Office for Civil Rights

Robinsue Frohboese, J.D., Ph.D.
Principal Deputy Director
Office for Civil Rights
200 Independence Avenue, S.W., Room 515F
Washington, D.C. 20201

Office for Human Research Protections

Michael A. Carome, M.D.
Associate Director for Regulatory Affairs
Office for Human Research Protections
1101 Wootton Parkway, Suite 200
Rockville, MD 20852

DEPARTMENT OF LABOR

Amy Turner
Employee Benefits Security Administration
200 Constitution Avenue, N.W., S-2524
Washington, D.C. 20210

EQUAL EMPLOYMENT OPPORTUNITY COMMISSION

Matthew Bradley
U.S. Equal Employment Opportunity Commission
1801 L Street, N.W.
Washington, D.C. 20507

Executive Secretary

Sarah Carr
Office of Biotechnology Activities
National Institutes of Health
6705 Rockledge Drive, Suite 750
Bethesda, MD 20892

C O N T E N T S

C O N T E N T S

C O N T E N T S

P R O C E E D I N G S (8:38 a.m.)

DR. McCABE: Well, good morning, everyone. I hope everybody had a restful evening last night.

We're very pleased this morning to be able to welcome two honored guests from the United Kingdom and Australia. We're delighted that Philip Webb, a member of the U.K. Human Genetics Commission, and David Weisbrot, president of the Australian Law Reform Commission, have journeyed so far to be with us today to tell us about some of the work that their commissions have produced and relevant policy developments in their countries.

We recognize the global nature of the issues we are addressing and how important it is to build bridges between our country and other nations who are grappling with similar questions. These ties can be mutually beneficial, and I'm certain that they will be of great benefit to us to learn from their experiences in an effort to enhance our own endeavors.

After the presentations, we will have a roundtable discussion with our guests which Chris Hook will lead us in.

First, let me introduce Mr. Philip Webb. Mr. Webb is a member of the U.K.'s Human Genetics Commission and the Board of Trustees of the Genetic Interest Group. Mr. Webb has also served as a member of the former U.K. Advisory Committee on Genetic Testing, the ACGT, where he worked on issues associated with genetic tests sold directly to the public, paternity testing, prenatal diagnosis, and preimplantation genetic diagnosis.

In addition to his public service, Mr. Webb currently is a management consultant in the health care industry. His previous professional positions were with AstraZeneca Diagnostics, Glaxo, and Imperial Chemical Industries, where he helped to establish a new genetics diagnostic business.

Mr. Webb, we really do appreciate your being here. Welcome and thank you so much for traveling so far to be here. Please.

MR. WEBB: Well, good morning, everybody, and thank you, Dr. McCabe, for that introduction and your kind invitation to talk to you this morning.

You're absolutely right. There are many similarities between our two committees or commissions, and I'm sure you'll see that in the presentation this morning.

What I'm going to do this morning is to give you an overview of the Human Genetics Commission came about, what it has done so far, how it operates, what its future work plan is, so that you get a good feel for the overall position, and then you'll also see some of the details.

So the Human Genetics Commission was formed in 1999, following on from the Advisory Committee on Genetic Testing, which was the predecessor body which was there for three and a half years, and I was a member of that before the Human Genetics Commission. We've been operating now for three and a half years and I guess we must have done a reasonable job because we got our mandate renewed for a further three-three term.

We're there to provide the U.K. government with advice on the big picture of human genetics and the advances that are coming down the track and what the implications may be for society. Our brief is particularly to look at the ethical, social, legal, and financial implications as well as the technical.

All of our members are appointed by open advertisement. Ads are placed in the major U.K. newspapers and interviews are conducted with a three-person panel: somebody from the Department of Health, somebody from the Department of Science, and an independent observer. People are chosen on their ability and the fact that they want a balanced commission with different people from different aspects of life, as you've got here on your committee.

One of the first decisions the Human Genetics Commission made was to work in public. We didn't want to be seen to be making decisions behind closed doors. Everything we do is either in public or all of our newspapers and agendas and minutes are all put on our website.

So there's a fine picture of genetic diversity. That's the Human Genetics Commission outside the city hall in Cardiff last month, where we held our last major commission meeting.

The commission is chaired by Baroness Helena Kennedy, who is a member of the House of Lords, and she also is a barrister and a human rights lawyer.

Our vice chair is an interesting person, Sandy McCall Smith. You may have seen recently some books in the bestseller list, one called "Tears of the Giraffe, and another, "The No. 1 Ladies Detective Agency." Alexander McCall Smith is a bestselling author. He's also a professor of medical law at Edinburgh University.

The rest of our commission is made up of a mixture of scientists, clinicians, researchers, and we have somebody from GlaxoSmithKline sitting on our commission to give us the commercial perspective. We also have lay people, consumer issues, lawyers, ethics, and we also have representatives from the chief medical officers of the four countries of the U.K. So they all have input there, what you would call ex officio members, and we have the chair of the Human Fertilization and Embryology Authority that regulates all assisted reproduction technologies in the U.K.

So our remit is to advise health and science ministers on issues of human genetics and the advances, and how those advances may impact society and health care in the future.

Part of our remit is to engage with the public and we think that's very important. We like to get the public views. It's not just a matter of us educating the public. The public can educate us and we very much want to hear their views.

We do also have a remit to look ahead because government doesn't want to be caught on the back foot as it was over GM crops. That caught U.K. government ministers in a difficult position. They weren't prepared for it, and so they want us to ensure that we're thinking ahead of the game and we're giving them good advice in good time.

So what have we done so far? Well, the first thing we've done is to produce a report called "Inside Information," and that report is on the storage, protection, and use of personal genetic data. I was going to bring six copies with me, but when I realized how heavy it was, I just decided to bring the one, which I will leave with you. It runs to 182 pages. If you can't manage that, we've put a small version together which is 32 pages, half-size, but in that there are something like 38 recommendations, and I'll tell you a little bit more about that in just a second.

The second one, "Genes Direct," is about the offering of genetic testing services direct to the public. That's a review that I chaired, and I'll tell you more about that as well in a few minutes.

The MORI survey. We felt it very important early on to get a feel for what the public were concerned about and what their thoughts were about the new genetics. So had a survey done of 2,000 members of the public and we had some very interesting feedback from that which helped us greatly to understand the concerns of the U.K. public.

The way we work is that we agree a work plan with government ministers. So one of the first things we did was put together our ideas of what we thought was important to give ministers advice on. So we have a rolling work plan. We just agreed a new work plan with ministers so we're clear on what it is we're going to be working on over the next two years, but we also have to have some capacity left in case we need to work in a reactive manner as well.

But we work through subgroups because with a commission of 18 members -- I'm sure you're aware of this yourselves -- it's very difficult to get actually to conclusions and get works done. So

the Genetic Services Subgroup is the largest subgroup and it's the one that I chair. It looks at everything to do with genetic services in the U.K.

The consultative panel that we have is made up of people affected directly or indirectly by a genetic disorder, and we value that group because they have firsthand experience and can help us to really understand better what the issues are for them.

The sort of meetings we have are twofold. We have information gathering sessions, which are perhaps similar to what we saw here yesterday, where people come and make presentations. Then we have our public, what we would plenary, meetings, where the business side of the commission's work takes place in public and where we make decisions and carry on with work.

We are also charged with advising the government on whether its regulatory frameworks are right. So we could decide that the Human Genetics Commission is no longer required, but I think we've got plenty of work to be going on with for the time being, so we won't be doing that just yet.

This dialogue that I mentioned earlier with the public, the picture you see there is one of our members, Professor John Burn, engaging with a group of 17-year-olds who are all studying science before going on to university. This was at one of our meetings in Birmingham, and we are keen to get the views of young people because they're not necessarily going to be easy to get their views if we do surveys and ask questions. They not necessarily very forthcoming. So we wanted to get a group of them together and really tease out what they think about genetics and what they think are the important issues.

We are keen to ensure that the public engages with us and as often as we can, around our meetings, we want to get the feel for what the people want us to be looking at, and that helps to form our work plan as well, but we recognize that that takes time and money to get it right.

Our consultative panel. We now have over 100 people who are volunteers who have decided that they would like to help us with our work, and we value their experience because they either are personally suffering from a genetic condition or a member of their family is, and they can give us real input about what the issues are, particularly about genetic services and how the health care system is looking after them.

So we ask them to comment on our draft reports, on our work plan, and they work mainly by correspondence, but when they first of all came on board, we had an introductory meeting which many of them attended, but we are cognizant of the fact that many of these people do find it difficult to travel, because of their disabilities in some cases, and therefore we work largely by email and correspondence. But we do value those people and their comments and that's very useful to us.

One of the issues that came out of our MORI survey was that the public were very concerned about the use by insurance companies of genetic testing information. Now, when we reviewed this in detail, we put a recommendation to the government that there ought to be a moratorium on the use of genetic testing while we look at it in more detail and see how it should be handled in the future. So there is currently a five-year moratorium in place where insurance companies have agreed that they won't look for genetic information on any policy less than 500,000 pounds in value.

We're talking here about life insurance. I know over here health insurance is a much bigger issue, but because of our National Health Service, we don't have that as an issue, but believe me. We have life insurance as an issue because many of these policies are linked to house purchase and mortgages.

So what we've agreed to do is during that moratorium period, do further work and research to advise government on what we think ought to be in place after that moratorium.

One of the things we want to do is to look at family history because family history, one could argue, is genetic information. That's why they want it. They ask you questions like "Have either

your mother or father died of any of the following diseases and at what age?" That is very often genetic information. So that's what we're doing on genetics and insurance.

Now, the report that I showed you, "Inside Information," that was a piece of work that took us about 15 months to produce. It's a report that was sent to ministers in May, 2002. There was a lot of interest in it. We carried out a big consultation exercise. We had a document called "Whose Hands on Your Genes?," which caught people's imagination, and we had a tremendous response from groups and members of the public who wanted to give us their opinions, and they were very useful to us.

We had a number of public meetings and this was the first time we'd used our consultative panel to make sure that the things that we were thinking about were the things that were important to them and they were very helpful in guiding us in that.

Now, we came up with a set of general principles, and we were trying to get a balance between respect for persons and privacy of the individual against the common interest that we all have in advancing genetic science and health care. So we drew up a set of principles in that report based on the overarching idea that there should be respect for individuals and those individuals should be entitled to genetic privacy. We don't think anybody should have their genetic information obtained without their consent.

I remember when your ex-President Bill Clinton came over to one of our quaint little English pubs and drank some of our wonderful warm beer, but after he finished, one of the big guys in black coats came and took the glass and put it under his coat. He didn't want anybody testing Bill Clinton's DNA. I can't imagine why.

(Laughter.)

MR. WEBB: But that's something we think is important, though, that people shouldn't take others' DNA and have it tested without their consent, and we have seen examples of that.

Obviously, personal genetic information can be extremely sensitive, and therefore should be treated with the utmost confidentiality, and we, like we heard from you yesterday, think that discrimination should not be based on people's genomes.

So our key recommendations -- there were 38, but I'm not going to bore you will all 38 of those now. The key one was that there should be a new criminal offense to prevent people deceitfully obtaining your DNA and testing it and publishing results of it.

That came to light in the case of Steve Bing, who had allegedly had an affair with Liz Hurley and produced a child, and somebody rummaged in his trash can and got his dental floss and tested his DNA and showed that he was the father of Liz Hurley's child. So that is the sort of thing that we're trying to avoid in the future.

We think that there may be a need for separate U.K. legislation to prevent genetic discrimination. We heard here yesterday about what's happening here in the U.S. and that was very interesting to hear, and we have recommended to the government that they should be looking at something along those lines in the U.K. We've also recommended that we need to be careful of that balance between the individual's rights and the general interest of society.

Now, genetic discrimination can come in all sorts of areas, in employment, in insurance, in education, and in health care, which is a particular issue here. What our public poll showed was that genetics and insurance provoked a really strong negative reaction with a large majority of people disagreeing with the insurance companies using our genetic information to raise their premiums, but we did note that it was a major concern among the public, and hence our moratorium.

Now, very recently, the U.K. government has produced a white paper on genetics. Now, a white paper in government terms is a statement of policy of what it intends to do, and this policy

statement is about realizing the value of genetic advances through our National Health Service. Additionally, 50 million pounds additional money has been put into clinical labs, training of staff, getting more people up to speed with genetics and understanding the importance of it in medicine and health care.

This white paper was produced by the government. The forward was by our Prime Minister, Tony Blair, and he recognized the importance of the Human Genetics Commission in ensuring public debate and advice to the government.

So the government's response to our first report, "Inside Information." The government has committed to developing the new offense that we recommended. That's fairly important for us, for our credibility and for the public seeing that government does take notice of what we do, and not only are they going to produce a new statute in that respect, they are going to consider the evidence for legislation on unfair discrimination and how to deal with that. They welcome our input to the debate and in developing policies for the insurance industry and they have committed to publishing a revised code of practice on patient confidentiality, which is another of the recommendations we put forward.

For those of you not familiar with the U.K. Biobank, that is a project being funded by the U.K. Department of Health, the Wellcome Trust, and the U.K. Medical Research Council to take 500,000 DNA samples from individuals between the ages of 45 and 69 to try and work out what links there are between various genes and certain health disorders.

Now, we suggested that that U.K. Biobank needed to be completely separate from anything that the police wanted to use for the DNA database, and the government has accepted that the police would only ever have access to any research database in very exceptional circumstances.

Now, the "Genes Direct" report, this was the review that I chaired looking at over-the-counter, as we would call it, direct genetic testing without the involvement of medical practitioners. We think that that's likely to increase. As I think you said in your debate yesterday, with faster, cheaper technologies and more and more genetic tests becoming available, it's likely to become a big issue.

We already know that a lot of testing is being offered over the Internet and we've even seen it in the U.K. in the High Street, people offering genetic tests to test your genes to tell you what your personal diet ought to be. Well, I'm not aware of the links between genes and diet and the interactions between those two as being well-proven science at this stage.

However, as you said yesterday, people increasingly want to take control of their own health care and not necessarily always to go through physicians and the traditional health care system. So we're trying to balance those two things, but we did find that in our consultation a great number of people were concerned that vulnerable people should have some protection from overzealous commercial organizations trying to sell them whatever genetic tests have come out this week.

So our key recommendations were that we didn't think there ought to be a ban on direct-to-consumer genetic testing, but we think there ought to be stricter controls on it.

Predictive genetic tests, where it comes to taking samples at home or even testing samples at home, we think there could be a serious problem there, and that comes back to this taking DNA and testing it without consent because if it is done at home, how does the lab know where that DNA has come from and whether or not there is informed consent? It's very easy to tick an Internet box, but it doesn't really tell you whether that person is, A, informed, or B, has consented.

So we think that most genetic tests should still be carried out in the context of the medical referral system and there should, in the U.K., be a well-resourced National Health Service Genetics Service that can properly manage and allow access to the latest tests and technologies. The white paper has confirmed that that will be the case.

How would that be carried out in practice? Well, the MHRA is the Medicines and

Healthcare Products Regulatory Agency. It's the equivalent of your FDA. It was only formed in April of this year by the coming together of two former bodies, the Medicines Control Agency that controlled pharmaceutical products and the Medical Devices Agency that controlled devices and diagnostics. So those two have now come together and we think that the formation of that body is an opportunity to further develop the regulatory system for genetic tests, and we have asked them to do that.

There's been a lot of debate in the U.K. about human tissues and the storage of organs taken without people's consent. You know, they've found huge numbers of organs stored in hospitals that were taken 20 and 30 years without people's consent, and that's become a big issue. So there is going to be an organization called the Human Tissue Authority that regulates the taking and storage of human tissues in the future. That may have a role to play in the taking and storage of DNA which, after all, is a prime human tissue.

We're also concerned about the Internet and we want to ensure that people are educated or at least they know where there is a reliable source of information, and we think our website, that I'll give you details of later, could be a reliable, independent source where people can find out information about genetics and genetic tests and what they ought to be aware of and what they ought to be looking for before going ahead and having a test.

So what about our future work? Well, our work plan covers genetics and discrimination. We need to have, in good time, advice to government on what ought to be the next steps when the moratorium on genetics and insurance runs out.

The main piece of work in 2004 is going to be on genetics and reproductive choice. Now, that is sure to be a hot topic conversation, and we know already that there are strong views on one side and strong views on the other and a big gap in the middle. It's going to be quite a challenge to produce a report and recommendations that will be signed off by the commission as a whole, but it's going to be an interesting piece of work.

I'm chairing a review into paternity testing and that's even going to cover genealogy testing as well. It's starting off as a short review, but I have a horrible feeling that it will take a little longer than some people think. But we're just in the process of producing a consultation document which will go out to groups and individuals who we know have an interest in those subjects and we will get some public debate going as to what the issues are. We have concerns about men testing children without the mother's consent or even knowledge, and we also, on the reverse side, have concerns about mothers testing other men who aren't necessarily the perceived father of the child without his consent. So there are a whole number of issues that we need to look at there and come up with some recommendations.

One of the other things that we've been asked to do in that genetics white paper by the government is to work with the National Screening Committee, which is another body that controls all national screening programs in the U.K. We've been asked to work with them to look at is there a case for offering genetic profiling of every child at birth? So we will be starting that piece of work in the next few months.

Then we have the ongoing of the monitoring of all of the things that we are keeping a close eye on, such as discrimination, gene patents, and those sort of things, and engaging the public.

Just before I finish, the genetics and reproductive choice, it has been identified as a key piece of work that we ought to do. Ministers do want advice on it. Every week in the newspapers, they use the words "eugenics" and "designer babies," and this does create a lot of concern for the public.

So we do want to come up with some advice in this area to how things ought to be carried out because there are new technologies, there are commercial interests driving more screening, and we all know about the gene chips, where it may soon be possible to test a child at birth for every gene that's

known.

We need to work with others, such as our Human Fertilization and Embryology Authority and the National Screening Committee, but you'll see from that pie chart there that when the question was asked what people think about the use of genetics to decide whether or not parents ought to make decisions about the birth of children, 57 percent agreed that that was a reasonable thing to do, 26 percent thought it was unreasonable, and 16 percent were neither strongly for or strongly against. But it just shows how the opinion is split.

So then in summary, the Human Genetics Commission is a well-established, well-respected body that has produced some key reports that government has acted on. Our role is to advise government and we are doing this by producing those reports, which are getting attention. We are promoting and participating regularly, weekly, in public debate. We are working in a very open, transparent manner, and we hope you will all take the trouble to have a look at our website and I'm sure you'll find some interesting things on there. We want to work with others to seek socially sustainable policies.

Thank you for your attention.

(Applause.)

DR. McCABE: Well, thank you very much, Mr. Webb.

I think we'll forgo with any discussion at this time and leave that to the roundtable.

I'm now extremely pleased to introduce Professor David Weisbrot, who is president of the Australian Law Reform Commission, where he is currently involved in an examination of gene patenting and human health issues and the protection of human genetic information.

Professor Weisbrot's past professional experiences included dean of law at the University of Sydney, foundation pro vice chancellor of the College of Humanities and Social Sciences, a commissioner of the New South Wales Law Reform Commission, and a member of the law faculties of the University of New South Wales and the University of Papua and New Guinea.

Professor Weisbrot is admitted to practice law in California and New South Wales and earned a law degree from the University of California, Los Angeles.

So it's a pleasure to welcome another Bruin to the podium today. Thank you very much for traveling so far.

MR. WEISBROT: Thank you. It's a pleasure to have been invited to speak. We've been admirers of the work of your predecessor committee and very pleased that the committee has been reestablished in this forum to do ongoing work.

I apologize for not being here yesterday. I really would have liked to have been, but I was sitting actually in Ed McCabe's chair at a similar sort of forum in Australia, and then had to make a made dash to the airport. I said something I never thought I would say in my life to a cab driver, which is "Do you think you could drive a little bit faster?"

(Laughter.)

MR. WEISBROT: And fortunately, have made it here.

As the introduction said, we completed a major review of the protection of human genetic information in Australia just over a two-year period. It was a joint reference, one to the Australian Law Reform Commission, of which I'm president. It's essentially the federal government's legal think tank on big picture issues. We get project work from the Attorney General and there are things that require detailed research and a high degree of public consultation and public engagement, and then we provide recommendations to the government afterwards. We've been in existence for about 28 years now.

We did this in partnership with -- and as it happened, it was a very personally and

professionally productive partnership -- the Australian Health Ethics Committee, which is a principal committee of our NHMRC, the National Health and Medical Research Council, which is more or less the equivalent of your NIH.

The inquiry said "The Protection of Human Genetic Information" in lawyer, bureaucrat, ability. It had three pages, I think, of terms of reference. They really boil down to three very simple propositions.

That is, in relation to human genetic information and the samples from which that information is derived, how in Australia do we best protect privacy and how do we protect against unfair discrimination. I've got that italicized because it is a tautological concept. We do allow many distinctions lawfully to be drawn among individuals and there are others we decide are invidious ones.

Then finally, how do we ensure the highest possible ethical standards, and although to a lawyer ethics essentially means the things that we impose on doctors and researchers, it was useful to have the Australian Health Ethics Committee remind us that that ethical dimension had to carry into all of the other areas we looked at as well, including law enforcement, insurance, and a whole range of other activities.

When we started, I don't think we realized the breadth and scope of the inquiry. We did have to go back to the government and say we needed a little bit more time. Well, partly that was because of the very high level of public engagement and we wanted to give people a meaningful opportunity to participate.

But going down the left side, you can see the things that were in the medical/health camp, and those are the things in blue I'll predominately be talking about this morning that I've been asked to speak about.

So we looked at issues of oversight of medical research, and that's primarily the Health Ethics Committee's role. We looked at issues of clinical genetic practice, at systemic health care issues, and at the emerging large numbers of genetic databases, tissue banks, and registers, because as we came to see, almost every collection of medical things, of tissue, blood, and almost anything else, can be systematized into a genetic database.

One thing, for example, is that every child in Australia who has been born since 1960 or so has been subject to a Guthrie test. I think you probably have the same thing here -- that's not real blood -- and these are all over, literally, Australia. They're stuffed in filing cabinets in pretty much every hospital where children are born. It's only now that we realize that there is in fact an unsystematized or an unorganized national genetic database and some real issues about how that's to be treated. Of course, issues about genetic privacy in relation to cancer blocks, blood and pathology labs, and whole range of related issues. I'll talk a little bit more about that in a moment.

Going down the other side are things I won't spend much time on here. You probably will talk about them at other parts of this conference or in the future, but we looked at issues of employment discrimination, where genetic testing can be an issue. We don't have major problems in this area at the moment, but that's not to say that there won't be significant problems in the near future.

It seems that every Australian has seen the film "Gattaca," in which we have a highly geneticized future, children are tested at birth, streamed into different occupations, and streamed out of others. We actually had to show the film to our staff because they kept hearing about it at every meeting that they went to. That's the sort of future, of course, we want to desperately avoid.

We did also look and spend quite a bit of time on the area of insurance. As in the U.K., this is primarily in relation to life insurance. We don't have a problem in relation to health insurance, essentially, because we also have a comprehensive medical system that covers almost all the things that

people want. You can also get private top-up insurance, but even that is community-rated in Australia, so you just choose from a menu of services whether you want to include dental, whether you want to include visits to the gym and shiatsu or whatever, and you get your price. It is not individual risk-rated.

So the key issue for us was simply life insurance, and we don't in Australia tie mortgages to life insurance either. The property in Australia is secured by the title to the property. So it's a lesser-order issue, but nevertheless quite an emotional one, and so we did spend quite a deal of time on that.

We took different approaches in relation to employment and insurance, I should say. In the employment area, our recommendations are highly interventionist. Essentially, we said genetic testing should not be a feature of employment decisionmaking, with some rare exceptions, and those are exceptions where you can show that there's a very clear occupational health and safety reason -- if you're running a beryllium mine -- and there won't be many of those examples because everybody keeps using the beryllium mine example. So there's not likely to be many, many others.

Or if it's a circumstance in which you're putting lives at risk. So it may be that for professional pilots, you have slightly more of a reason to look at genetic testing if there's a rational scientific basis to do so, but basically we said don't do it in that context.

In the insurance area, where it's essentially based around the interchange of information and actuarial practice requires accurate and material data, we were a little bit more open to the idea that it should continue to be used. We also had some empirical studies done of the insurance industry which they were cooperative in and found that over the last two years, there were only about 100 applications a year that contained genetic test information. So it is still not a major feature. In fact, it's hardly a feature at all in actuarial decisionmaking.

However, family medical history, of course, is a feature in almost every application. So we actually focused a bit more of our attention on trying to get the industry to deal intelligently with family medical history, not to overreact to labels, not to overreact to information, to try to get actuaries that are trained in genetics, so that they're assimilating the latest medical and scientific knowledge, rather than relying on what they might have studied many years before in first-year biology before they did their actuarial studies.

We also spent quite a lot of time looking at law enforcement because, of course, now genetic testing is a standard feature of all metropolitan police forces. That was driven home, I think, to the Australian public recently in relation to the Bali terrorist disaster in which nearly 100 Australians died. Most of those people were identified positively by genetic tests because there was no other effective means of identification, similar to the World Trade Center disaster here.

There are real problems in law enforcement in Australia in terms of harmonization of laws. We have nine different police forces, six state and two territory, and federal. They all have different rules and regulations relating to genetic testing. They essentially operate in two different planes.

There are the ones that rely on recommendations that came out of a major task force in Australia, and those only collect DNA in serious crimes. They only store it as necessary. If someone is acquitted or charges are dropped, their DNA profile is destroyed. So it's, you might say, the civil libertarian model of collection.

On the other side, we have the states in the Deep North, which is the equivalent of your Deep South, as you'll appreciate from the geography, and those places want to collect everybody's DNA for every reason and keep it in perpetuity. The only exception they make are for other police officers --

(Laughter.)

MR. WEISBROT: -- because there have been some industrial issues. One of the few

employment disputes we've had has been in relation to police officers who have been asked to provide a sample because they're investigators and there's a possibility of their DNA being intermixed in crime scene samples. There have been a couple of threats of industrial action and the authorities in the various jurisdictions have backed down from asking the police to provide compulsory samples.

We also look at issues of kinship and identity, a broad grab bag of some very different but complex issues. The extent to which genetic testing might be used in immigration, for example. It is used now sometimes for identity. So our immigration authorities tell us, using their soft voice, that sometimes there are people who come from countries where there aren't good records and they want to show that they are related to an Australian who's sponsoring their application for immigration, and this will give them an opportunity affirmatively to establish that they are the parent, son, or daughter of the person who holds Australian citizenship and can sponsor them.

They don't do health testing, predictive health testing, at the moment. That could come in the future. At the moment, they tell us it's tough enough just to do sort of TB testing around the world on people who are applying. But it's something we've asked them to look at in much more detail and to come up with protocols on.

Parentage testing, paternity testing, is something we didn't think would be a big issue in the inquiry. It turned out to be a very big issue. There are a lot of very angry dads in Australia, it seems, or putative dads. So I would say the bulk of the submissions we received were in that area. They were all very angry ones. They usually started off with a denunciation of the Australian Law Reform Commission for no particular reason other than we were government or people who had some authority. Then it proceeded into stories about how this an area in which men are again being disadvantaged by the state and by our family court.

There's a lot of advertising going on around that now. That's something I'll talk a little bit more about in a moment, but almost any doctor's or surgery office has these kind of things in it and they're all over the place. "Are You My Daddy?" In fact, our experience is little children never ask that question, but angry dads certainly do.

We also made recommendations that were disappointing to those groups because we said we didn't think that surreptitious testing should be allowed in those circumstances, non-consensual testing, without an order of the relevant court.

We looked at issues of Aboriginality with the most sensitive concerns. It was an area where, when we did our original brainstorming, we said what would happen if somebody denied or challenged someone else's right to describe themselves as an indigenous person of Australia, an Aboriginal person? Would genetic test come up? We thought, well, fortunately, we don't have to deal with that question.

About mid-inquiry, there was an election of ATSIC, the Aboriginal and Torres Strait Islander Commission, which is our vehicle for self-determination of indigenous peoples, and one activist in Tasmania challenged the ability of 800 people in Tasmania to be on the roll of electors. He said, "I know you. You're not Aboriginal."

A lot of those people came forward with genetic tests that said, "Here's a document from the 1800s that shows this is an Aboriginal person. Here's the chain. Here's how I fit into that family."

Very, very difficult issues, but real-life ones at the moment. In the end, we didn't make positive recommendations in that, other than to say it was primarily a matter for Aboriginal people to determine as a matter of self-determination and we provided a lot of history and comparative data, and also some of the guidelines on how the law currently operates, but we didn't feel we had done the proper

consultation with Aboriginal communities to make positive recommendations in that area.

Then we looked at a grab bag of other services and entitlements. We had some people come forward to us to say that they were not allowed into government job retraining schemes because they had predictive health information that indicated that they would not be good candidates for that in the views of the government. People from Huntington's families, for example. We had people raise with us the issue of whether the education system might ever debar students from coming into schools or certain schools because they shared a predisposition to ADHD. People were concerned about nursing homes using testing for early-onset Alzheimer's.

We had issues already in sport. The State of Victoria's Boxing Council has been talking about the idea of having professional boxers genetically tested before they would issue a license to see if they had the marker for punch-drunk syndrome. It's not clear whether you would need it or not have it to get the license.

(Laughter.)

MR. WEISBROT: But they're starting to talk about those issues, and you may have read in the paper that the Australian Institute of Sport, which is a very high tech operation in Canberra, has now identified the genetic markers that are found in 95 percent of the world's elite sprinters. It's fast-twitch muscle fiber.

So we have talked to them about whether there is any ethical dimension in there or whether they're talking about gene "therapy" as the next doping issue in world sport, or whether they will do the "Gattaca" thing and say to the promising young sprinter, you're winning all your age groups, you're the best young sprinter in Australia, but your genetic marker is just not showing it, so why should we invest hundreds of thousands of dollars in your development?

Sport is like religion in Australia. In fact, much more important, and so this is an issue that we looked at in some detail. I have to say that I'm pleased that the Australian Institute of Sport, which hadn't been looking at the ethical dimension overmuch, is now starting to take that very seriously.

The inquiry processes I think were very important in shaping how our ultimate recommendations were found. The commission itself consists of a few of us lawyers and judges and we have ultimate responsibility, legally and morally, for the report and the recommendations, but our standard modus operandi is to set up an expert advisory committee, a steering group, for every project that we do.

In this one, we included some of Australia's leading genetic researchers, people who have been talked about as potential Nobel Prize winners. We had people who headed up clinical genetic testing services, bioethicists, health consumers, indigenous health consultants, the head of state and territory public health systems, actuaries, privacy discrimination commissioners, and so on. Lawyers, human rights lawyers, forensic scientists. It was to make sure we were being steered in the right direction and we gave the proper weight to the different issues.

We engaged in the usual extensive literature review, which took us to the work that SACGT did previously, and also of course we were heavily reliant on some of the groundbreaking work that the U.K. Human Genetics Commission has done.

We produced two consultation documents which were in heavy demand -- those are just the acronyms for them -- and issues paper and a discussion paper, and that was to help the public work through these issues in stages and to help us work through in stages.

We conducted 15 major public forums around Australia. They were in all of the capital cities and most of the major regional centers. They were quite well attended. They got a lot of media coverage, which also helped the attendance at these things.

We received more than 300 written submissions, and these ranged from the big, well-

researched, computer-produced ones that you'd expect from the Australian Medical Association, the insurance lobby, the employers groups, the major cancer research labs, and so on, but most of them were individual heartfelt letters from people saying, "This is what we're going through. We have a child who has this genetic condition. We don't want other families to have to go through this same sort of thing in the future." They were very, very important. Many of them were very moving.

The 300 is probably a very conservative figure. We have some people who wrote to us every day for two years and they continue to do so. We counted those as one continuous submission.

(Laughter.)

MR. WEISBROT: We also had over 200 targeted meetings. These were with genetic support groups, clinical geneticists, GPs, rotary clubs, anybody who wanted to talk to us either to get information from us or to provide information to us about the issues. So it was a pretty thorough national process.

The final report that we lodged in this inquiry we called "Essentially Yours," and it is meant to be kind of a double entendre. On the one hand, we argue very strongly against ideas of genetic essentialism. That is, how people should be dealt with is simply by a reading of their genetic code. On the other hand, we talked about it being essentially up to the person to control their own genetic information and genetic destiny.

The report was launched by the Attorney General and the Minister for Health in our federal Parliament in May, 2000. It again received quite a bit of media coverage.

Given the breadth of subject areas I talked about earlier, you'll appreciate that we had 144 recommendations, but these were not all -- in fact, maybe not even primarily -- directed to government, and I think it's a complexity of the modern world that ensured that we had to make recommendations directed at all layers of government -- federal, state, and territory -- but we also made recommendations aimed at regulators; at educators because we found that our Australian doctors are not terrifically skilled up in genetic issues, and we heard that consistently from people in the public forums; we talked more generally about health professionals, genetic counselors, and others; and then recommendations directed at insurers, employers, law enforcement agencies, and all of those I talked about before.

I've been delighted to keep verbaling, as we say in Australia, Dr. Francis Collins, who, when he spoke at the International Genetics Congress in Melbourne just a month or two ago, talked about this report as "a truly phenomenal job, placing Australia ahead of what the rest of the world is doing." We thought that was very nice of him to say and we hope it's accurate.

Our recommendations are not self-executing, of course. We're an advisory body. However, we're fairly optimistic. We have a very good track record of having our advice turned into action, whether that's legislation or administrative or other action.

You can see there that about 58 percent of our recommendations are implemented. Most of them get in. There are another 22 percent that are partially implemented. The nil implementation block we hope will continue to narrow because that includes three or four of the most recent reports, including "Essentially Yours," that the government and others are still working through. So there's a only a small proportion of cases where government actually rejects our advice or puts it in the too hard basket.

In our public process, we found, not surprisingly, the same sort of things that American and European commentators have talked about, and that was a real ambivalence about genetics. On the one hand, there was tremendous optimism, maybe even overoptimism, about the medical breakthroughs. So there's not a day goes by where the Australian media now don't have some story about a remarkable so-called discovery of a so-called gene for something and that that's going to lead to tremendous clinical breakthroughs either in gene therapy or smart drugs or one of those sorts of things.

Almost all of those, when you track them back, of course, come from the pharmaceutical company or from the lab that's doing it, and there are good reasons for them to do that in terms of getting funding and so on, but it also maybe creates a little bit of overoptimism in the community, or at least not a sense of how far away we are still from some of these therapies. On the other hand, there are some remarkable breakthroughs happening every day.

On the other side, there is this still kind of a gut anxiety about loss of control. So we had people talked to us about is this more mad science? Is it possible for us to regulate? There are people in the indigenous community and other communities who have fears about eugenics.

I remember one meeting in which we talked about a Tay-Sachs screening program in the Jewish school system in Sydney and Melbourne, and a woman came up to me afterwards and said, "My grandchildren have been tested in one of these schools and I guess it's okay. I think it's probably important. I do have some serious reservations, though," and she paused a moment and pulled up her sleeve and showed her number from the concentration camp, and I didn't have to ask her why she had some lingering concerns about that.

There are interesting fears about commercialization, too. Again, there's a gut instinct. I notice that the latest John Le Carre novel, now that the old evil empire has fallen, is about big pharmaceuticals. You know, is this the new evil empire?

We found that, continuously in meetings, it was at almost every meeting we had that there would be somebody who would get up and make an impassioned plea and say, "I'm a good citizen. I'm happy to give my genetic material to research to help find cures for Parkinson's disease and diabetes and so on, but I don't want those big drug companies making a profit on it," and especially those big American drug companies." The Swiss and French and other ones seem to be getting off fairly lightly, but there was a real fear -- again, a gut fear -- and particularly of genetic material being taken out of Australian, taken offshore, where there might be lesser controls or at least an inability to track what was being done with it.

The data is interesting, though, in that all of our public opinion surveys show that Australians have not -- and we hope won't have to say not yet -- lost faith in the possibility of effective regulation of biotech in the public interest. If you look at some of the surveys in Europe, they're fairly dismal, and maybe for good reason. We haven't had in Australia Chernobyl and foot and mouth disease and mad cow disease and so on, and areas where public authorities have shown themselves singularly unable to cope or unable to show how they're coping. So Australians still are exhibiting some good will and openness in this area.

A threshold issue for the commission, of course, was is genetic information exceptional? Do we have to come up with whole new regimes, a genetic privacy law, a genetic discrimination law, and other kinds of qualitatively different protections? We went through the usual litany. In this group, I think I need not talk about all of those issues in any detail at all.

But our conclusion was we need to recognize the special features and challenges of genetic information, but not to embrace genetic exceptionalism as a guiding principle for public policymaking. I think we've moved through, in the world that's thinking about these issues, the cycle of first the future shock and being struck by the new genetics and the need to regulate heavily to deal with it and then more to an inclusivist approach which says, well, we've already developed all kinds of sound principle and practice in relation to other challenges to our public health system -- HIV and others -- and really we can just build upon that and then deal with the different features of genetic information.

This is something we use to explain to the public, I think, more about the process than I need to talk about here, but it's a scientist looking at the 3 billion piece puzzle and the one on the right

saying, "I think I found a corner piece." So this is trying to say don't rush to far ahead in thinking where we are with the science, although it is moving remarkably quickly.

I guess, partly in our esteem for the U.K. Human Genetics Commission and other similar bodies, our central recommendation in "Essentially Yours" was that we needed to something similar in Australia and we recommended the establishment of a Human Genetics Commission of Australia. Again, this is recognizing the rapid pace of change and the need for a continuing independent, authoritative voice that could talk to government, that could talk to the public, and so on. So in order to keep that level of public content up, so that people thought we're not being lied to, there are people who are giving us the cutting edge information, and it's open process, we needed to establish this sort of body.

We have had since quite a lot of groups come to us -- genetic support groups, scientists, and others -- and say there's almost a post-review depression in the community. When you were operating, when the Law Reform Commission inquiry was going for two years, we had somebody to come and talk to and share all these ideas and concerns, and now that you've stopped, we've got nobody to talk to. So I think it is important for our government to move quickly to establish a Human Genetics Commission to be able to capture the concerns, the ideas, and so on.

As with the U.K. commission, we recommended broad-based membership. So on the one hand, we need people who understand the cutting edge science. On the other hand, we need also the ELSI dimension, the people who understand the ethical, legal, and social implications, and of course, people from affected communities, and that idea about providing a national forum.

We assign some specific responsibilities to the HGCA. One is in the insurance and employment areas, to keep a watch and brief over that, and to provide high-level advice to those industries. The other I'll talk about more in a moment, which is looking at genetic tests in terms of access and equity.

I was asked to talk particularly about our approach to regulating access, and so the next bunch of slides deal with that. One role we assigned to the HGCA was to identify certain sensitive genetic tests that might require restricted access and counseling, and to advise our regulator, which is the Therapeutic Goods Administration, kind of the equivalent of the FDA, accordingly.

We used an analogy with the approach to HIV/AIDS testing. In Australia, this testing can only be done at certain public hospitals that do a lot of the testing, so they have the highest possible quality assurance and analytical regimes, and they are also very, very sensitive to issues of privacy and non-stigmatization, and they also are very good on counseling. So if you limit the number of suppliers of the testing, you can also ensure that the counseling regime is appropriate.

The technology exists, of course, to have an AIDS test kit in every doctor's office, but the Therapeutic Goods Administration, on advice from our public health system, has restricted access, and we've said there may be some genetic tests that are in that category, that people really shouldn't have them without the ability to have the appropriate counseling and other kinds of infrastructure in place.

On the other hand, there are going to be other genetic tests that are really just replacing other kinds of biochemical tests, and if it's just another generation, if it's a test that used, for example, for hemochromatosis, rather than a liver biopsy, well, then maybe you don't need to go through a whole lot of that process. But there are going to be some that do have that extra sensitivity.

We also ask the HGCA to look at request pathways. That is, in Australia, to say who can order a genetic test. Should general practitioners be able to order every one? Should some only be orderable by clinical genetic specialists? If they are ordered, should that trigger some sort of counseling thing on a computer? Should they be rebatable through Medicare, which is our comprehensive national health scheme similar to the NHS in the U.K.? So those are issues we asked the HGCA to look at.

We also looked at the lab side and said that any DNA testing for health purposes where

there's a reportable result should only be done by accredited labs that have been accredited specifically to do that sort of genetic testing. So wanted to raise both the quality assurance side there and also to make sure that those labs had the appropriate ethical regime and also were sensitive to the appropriate counseling needs.

On the regulating illicit testing side, from remarks that were made earlier, I take it that you looked at this yesterday, but we predicted the obvious, that as the technology increases and improves, that there will be increasing availability and decreased cost. There will be a lot of direct marketing. There are financial and other incentives for people to, or think they need to, have genetic testing done.

So we predicted increasing pressures for, for example, non-court, non-consensual paternity testing. That's already evident in Australia. As I showed you, there are leaflets available. There are ads on late night television in similar terms and they usually involve do-it-yourself buckle swabs, which you then put in a little plastic bag and send offshore because Australian labs, by and large, won't do their testing now on that basis.

We were concerned about surveillance testing of partners and children because there already are groups that market non-genetic tests in those areas. You know, we can tell you whether your children are taking drugs or are there going to be the equivalent of those genetic tests?

Then we were concerned about the illicit testing that Philip talked about earlier. Will journalists and private investigators try to get Nicole Kidman's DNA for a front page story? Why is Russell Crowe so angry? You know, he's got the gene for anger.

Then on the more serious side, will we have employers doing that kind of illicit testing that we saw in the case here, the Burlington Railway case handled by the Equal Employment Opportunity Commission.

We also followed England in recommending the creation of a new criminal offense, and I should say I spent 30 years in law reform trying to avoid the use of criminal law in regulating most areas, but we thought this was serious enough and compelling enough that we would create an offense in very similar terms. That is, knowingly or recklessly -- recklessly is adverting to the consequences, but not really caring about them -- submitting another person's genetic material for testing without their consent or without other lawful authority. So a court can order a paternity test, even if the person doesn't want it, the police have statutory authority to take DNA in certain circumstances, and so on.

There are also questions about regulating curiosity, in a way. We thought that there is probably going to be more and more of these "genetic health" test kits available. One of the most famous is Sciona's test, the Philip Smiley, and they were marketing these through the Body Shop until very recently. I guess they could look at your genes and tell you whether you should get the raspberry bath balm or the passion fruit shampoo or indeed both to improve your genetic health.

(Laughter.)

MR. WEISBROT: They were only 120 pounds, so widely accessible, and I'm sure a lot of people gave them as Christmas gifts. After some adverse publicity, the Body Shop has stopped distributing them and I know quite a few of the major pharmacies in Australia have refused to stock it as well.

But when the controversy started, the CEO of Sciona was asked what do people need this for? Why would they want to do it? I thought the quote is worth looking at in full. He said, "There's already a lot of information out there recommending, for example, a diet high in fruit, broccoli, and grains and low in char-grilled red meat, smoked and preserved foods, and alcohol. Consumers find this advice daunting, as they are not sure to what extent it pertains to them as individuals."

So I guess that means you could either have lots of fruits and vegetables and exercise

and eat grilled fish or you could lie on the couch and have a lot of hot fudge. If you're not sure, what you really need is a genetic test to make your mind up for you.

We thought, well, how do we regulate these areas? The best responses involve a high degree of consumer education, obviously, and we assigned a major, major role for the Human Genetics Commission in promoting public education about genetics. Also, providing some financial incentives and disincentives. In Australia, again, with our comprehensive health scheme, not putting a genetic test or genetic test advice on our schedule of things that are covered for free is a substantial financial disincentive for people to engage in it. It becomes expensive otherwise.

We looked at some areas of formal regulation. As I mentioned earlier, our Therapeutic Goods Administration could restrict access to certain sorts of test kits, and we recommended the introduction of criminal law in one area.

Then some extent it really is probably buyer beware. Those Sciona tests, it seems to me, are probably the genetic equivalent of mood rings. I'm not sure whether we want to use the full force of the state and criminal law to crack down on them. On the other hand, we hope that consumer education will be sufficient that people won't waste a whole lot of money and, even more, a whole lot of anxiety on them.

Some systemic health care issues. We wanted the system to start gearing up now for the time when, and this is very loosely, but all medicine will be genetic medicine. There will be a genetic component in most sorts of medical tests and medical advice. So we asked the Australian government to start looking at strategic planning issues, at cost issues, training needs, and so on.

We asked family doctors to start taking this more seriously because, as I said earlier, a consistent message we heard from affected families was "Our doctor didn't know anything," and of course, the more rare the condition, the less the doctor knew. Of course, the older and more experienced and better the doctor in many ways, the less they knew about genetics as well, by and large.

So we heard it over and over again about a lack of knowledge, about a lack of communication skills, and it's not easy to communicate ideas about probability and risk. The literature in that area is quite frightening, actually, and in a country where gambling is a passion -- gambling on sport is probably the national religion -- you understand very quickly that people have no idea about probability and risk when it comes to many areas of their life and they should have a better handle on it when it comes to important aspects of their own physical well-being.

So we talked about the need to develop an integrated program of medical professional education -- not just for doctors, but for other health professionals -- starting at the medical schools and working through.

A very close friend of mine is the dean of a leading medical school in Australia. He's also a fairly defensive person, and so when I approached him with all of this criticism, I sort of stood back and waited for the response. He thought about it for a moment and said, "Yes, that would be right."

(Laughter.)

MR. WEISBROT: So there is a recognition that we need much more in that way.

We also need more resources and backup for genetic support groups because, again, the lesson from the families was, yes, it was good talking to a doctor, but things really started to fit into place when they talked to other families, and sometimes that meant families around the world through good linkages on the Internet and they told us how they were coping. They often had very practical, down to earth, kind of day-to-day solutions for things, good advice, and so on.

So we need to support those people as well as medical professionals. It's a very, very cost-effective way of assisting the affected community, but one that doesn't seem to figure into government

budgeting as often as it should.

Genetic counselors were, in many ways, we thought the heroes of the revolution and our inquiry because, again, people said to us, here's what happened. There were some symptoms or some family history. We went to get a genetic test. We got a result. Our doctor didn't know much about it, so we went home and, in the way of the modern world, we typed it into Google and then we scared the wits out of ourselves when we looked at all of this information. We're all going to die. We're all going to die very soon.

But 99.9 percent of the time, they were greatly reassured after visiting the genetic counselor. Now, it may have been that they still had some serious issues to deal with, but at least they were given perspective, they were given an entre into social and psychological counseling, they were given some good concrete information, they were put in touch with genetic support groups, and so on.

So we thought this was really, really a key to the development in this area, and we made some recommendations that would try to facilitate this about more funding, about recognizing genetic counselors as a distinct profession in Australia, and this has financial implications because if you're a professional in the health area, you get a provider number and you can bill Medicare, which is again our national comprehensive system, and so there's some resistance to increasing the number of people who have the ability to bill the system. On the other hand, if you're going to limit it, I don't think it should be starting with genetic counselors.

Then we wanted to improve the articulation among the various clinical geneticists, GPs, public health officials, and counselors.

This is, from the New Yorker, I thought a perfect comment on issues of genetic counseling, which is the doctor is saying, "There's no easy way I can tell you this, so I'm sending you to someone who can."

One of the controversial issues that we dealt with, and which will continue to be one of the big issues, I think, is how health professionals deal with shared genetic information. It's the other side of that. When we first started the inquiry, our idea, well, this is really sensitive, personal information. So we're probably going to have to beef up our privacy laws to protect individuals.

Part of the way through, we started saying, well, in some areas for sure, but in other areas, the shared characteristic, the familial dimension of genetic information, is going to require moving away from some of that idea of individual privacy and protection. We heard that from genetic counselors, from family cancer registries, and so on, and increasingly an idea that there was a real problem around individual confidentiality, the individual doctor-patient relationship.

So many genetic counselors, many people at the cancer registries, clinical geneticists, said to us, you know, what I have all the time is somebody coming in and getting a positive test for breast cancer, colon cancer, FAP, one of those, and I'm not at all confident they're telling their genetic relatives that they maybe need some screening, that they're sharing that information, and what I live in fear of is the phone call from somebody who rings up and says, "You know, two years ago you tested my sister. She was positive for BRCA1. She never told me. We haven't spoken in many years since that terrible Christmas night some time ago. All you had to do is phone me and I would have gone and got my screening, but I didn't know anything, and now I've got an advanced cancer."

So they're saying what can we do in that area? Sometimes we're not confident from body language and sometimes we're not competent because the person says "I haven't spoken to my family for years," and sometimes they say -- you know, the perversity of it -- "Well, good. Here's my vengeance on all of those people who made my life miserable for all these years. I know something important and I'm not going to share it." What do people do in that circumstance?

It's a very tough one. What we did was recommend to our NIH equivalent -- which does set medical standards, doctor-patient standards, in many areas -- we ask you to develop a rule through more thorough consultation in this area that will allow health professionals to disclose information, even where the individual has not given that consent, where a patient's genetic relative would be at serious risk of harm. We know that there are very difficult and complex issues around that, especially in English-speaking countries, according to all of the cross-cultural data.

I know those of you who have seen Dorothy Wirtz' very impressive cross-cultural work, which is confirmed by our own international consultations, is that in the English-speaking countries, and it's consistent across Australia, New Zealand, U.K., U.S., and Canada, the individual doctor-patient relationship is the prime one. It's very difficult to get people to budge off that.

If you look across Asia -- I was just at a national bio symposium in Korea -- people had much less difficulty with the idea that this is shared familial information and that, of course, other family members should be an integral part of the decisionmaking and of the information flow.

I think it's in my next slide. Yes, this was one I was given by a Korean geneticist friend, and he said he thought that genetics was proof of Confucianism. So here's the idea that it is all shared. It's all family. It's not individual. These ideas of shared genetic information fit perfectly into his world view, into his personal and religious views.

Issues about managing genetic databases. I mentioned that earlier and Philip mentioned that. I won't spend much more time on that. We don't have in Australia an equivalent of BioBank or Iceland's DeCODE or the Estonian one, the name of which escapes me just this moment.

However, we have vast numbers of inchoate or unorganized databases; a national "collection" of Guthrie cards; of course, genetic information that could be taken from pathology labs and blood banks and tissue banks and familial cancer registries; and then thousands and thousands of research projects all over the universities, the biotech companies, and so on.

So we need to look more carefully at it. We asked our, again, NIH equivalent to provide a new chapter of our national statement on ethical conduct in research involving humans, to develop a chapter specifically on the ethics of how to manage these databases. These included issues relating to more effective oversight by ethics committees, human research ethics committees; better consent and disclosure protocols, and we provided some model prototypes in our report; and more thought given to issues of deidentification or use of gene trustees, independent mediaries. Then controversially, and it got our privacy commissioner upset, but that's okay, is extending the Privacy Act beyond data to cover samples, genetic samples from which genetic information can be routinely got. We said that if those things don't work, then we might move to a more heavier regulatory system involving actual licensing or registration of databases, but we didn't move that way yet.

We looked at issues of population genetics and screening. You know, are these sensible programs that we've got? There are a whole lot of them out in the community. Are we appropriately targeting at-risk populations or are we doing genetic testing to satisfy grant applications and researcher interests? We have the issues of the neonatal testing. There are school-based programs.

Then there's a program starting to emerge in the workplace. There's a HaemScreen initiative in Australia in which employers fund hemochromatosis screening. It's an entirely voluntary program, but it's done at the workplace level, and the employers have said and the insurance industry has said if people volunteer for this and they then do the right thing, which is give blood to the Red Cross and keep the hemochromatosis in check, then we won't discriminate against them either in the workplace or in terms of risk-rated insurance.

I don't know. It's an interesting collaboration. I'm glad those various sectors are

talking to each other, but I have some concerns about the program and also whether, just epidemiologically in Australia, that's the program in which we want to invest lots of money.

So we've asked for kind of better balanced decisionmaking there and also some thought about how we use these databases. Do we use the Guthrie cards in an epidemiological way, even if it's got no individual consequences? Would it be appropriate for the Australian government to say let's test all the cards from 1960 for diabetes markers to see if we need to have a better education program in that area? I mean, are those the sorts of things we would want to do?

I would mention just briefly that we've moved on to the next stage or a second inquiry, which is looking at the intellectual property aspects of genetic materials and technologies. Many people urged us to do this within the context of the previous inquiry, but it really wasn't possible.

But we have now moved into it and our terms of reference talk about the need to balance encouraging innovation and investment, on the one hand. In other words, support the current intellectual property registration regimes of granting of patents and, to some extent, copyright without harming further research or reasonable access to clinical genetic services.

In Australia, as here, a lot of these issues arose out of concern about access to breast cancer testing, for example, about the relationship between Myriad and our public health system. Myriad has since licensed an Australian company, GTG, to provide those services in Australia. So there's a very direct and lively public debate in Australia about those issues.

Of course, we are ultimately bound by our international obligations in these areas as well under the TRIPS agreement, although I guess part of what our inquiry is looking at is whether there is significant enough wiggle room in there for us to take some steps in Australia that will provide a better balance than may be done in some other countries.

Again, those are the issues that we're looking at, which I won't spend much time on now, but they're there.

It's probably worth saying there are some important differences from the U.S. situation. We have a comprehensive public health care system with subsidized community-rated private health insurance top-ups. Most of our genetic testing occurs within the public system now, although there will be an increasingly significant private element, including the company GTG is now moving to develop very high-tech labs that will provide much quicker turnaround than our current public testing labs do. So there will be more and more pressure to shift things from public to private.

We have a question about whether there is less active, less aggressive, enforcement of patent rights in Australia. It's just an empirical issue that we're looking at at the moment. We've done the usual establishing the advisory committee for that program, we're doing our usual consultation program, but I think significant in this area is that we're looking at collecting empirical material about these issues in Australia. So we've admired the work that Mildred Cho and others have done at Stanford on looking at the actual practical effect, day to day, in clinical genetic services and research labs, and we've got some mirror studies that are being done in Australia.

Some possible approaches to reform in that area, we're looking at how to use existing law and process more effectively and issues about compulsory licensing and Crown use. We're looking at our fairly vigorous competition laws, and there's a tension in the intellectual property area where the grant of a patent gives someone a monopoly right. On the other hand, we have very vigorous competition laws and a very active Competition Commission. So what's the interplay there?

Then in Australia, again more so than here, there's a key issue about government purchasing power. We effectively have a monopsony in this area. That is, that all the testing is done through public health systems and drugs are almost all purchased through our PBS system, Pharmaceutical

Benefit Scheme, which is subsidized by many billion dollars by the government and keeps the cost of drugs down. So our government has very strong purchasing power.

This has been a key issue in the free trade agreement discussions that are going on now between Australia and the U.S. in which U.S. authorities have said, well, those billions of dollars of subsidy are harming the interests of our pharmaceutical companies, and the Australians are saying, well, a lot of things can be on the table, but PBS and MBS, the medical side of it and the pharmaceutical side of it, are things that are very, very important. You'd be a very brave politician in Australia to talk about weakening either of those things.

Some other reform options. Possibilities of looking at statutory exceptions for medical research or clinical practice, and maybe some altered criteria for patentability, but I think we're not likely to go that way because of the locked-in structure of the international rules and practices.

Further information. Everything we do, we do in public, and all of our publications are on our website. The report that we did, which I shared a slide of, I can't carry because it's that thick, 1,200 pages. It does go nicely on a CD, however, and we're happy to provide those for people who contact us. All of our issues papers -- for example, the one now in the gene patenting area -- are available on our website.

I'd also just like to say that I hope this next little period won't be the end of the dialogue. So if you've got issues or concerns or want to find out more about what we're doing there or have suggestions about any of our papers or wish to make a submission, then please do contact us in any of those ways. Probably electronically is the most efficient way, given the time zone difference.

Thank you very much for your attention.

(Applause.)

DR. McCABE: Thank you very much, Professor Weisbrot.

While you're taking your seat, I'd like to acknowledge Ms. Lynn Mainland from Health Canada, who is in attendance, and wish you a warm welcome. I'd also mention that the SACGT heard and benefitted from the Genetics Commission of Ontario, and we appreciate your attending the meeting here.

I'll now turn it over to Dr. Hook, and we should go until 10:25 and then we'll just take a 10-minute break.

DR. HOOK: Well, I'd like to begin by thanking both of you gentlemen for outstanding presentations, and also, please take back to your various groups our acknowledgement and thanks for the tremendous work that you're doing. I hope that we can continue the dialogue.

I'll just begin with a question in that are there areas that either of you would see as important for all of our various committees or commissions to be directly collaborating on? I know we could learn a great deal from each other and we often have similar thinking in various areas, but are there areas that you would identify, perhaps the direct marketing or Internet sales of genetic testing, which we should be explicitly working together on?

MR. WEBB: Yes. I think that the Internet testing services that are offered are of concern and nobody has the answer individually as to how we can best deal with those matters. We do want to enter into dialogue with other nations about how best to use sites for education because we all recognize that education is probably the easiest answer, rather than legislation, and so we would certainly like to have dialogue about how to regulate, as far as possible, Internet testing.

DR. BEMENT: Arden Bement from the Department of Commerce. I also echo Dr. Hooks' comment about your lucid and very informative presentations.

My question has to do with measurement uncertainty. Microarrays and gene chips are still relatively new technologies and still have rather high error budgets. So questions of statistical

significance and validity enter in and also protocols to analytically resolve legal challenges, especially in forensic applications of genetic determination.

The other thing that is often misconceived is that accreditation equals conformity to standards, which it does not. So accreditation by itself is probably necessary, but not sufficient.

I'm curious to know whether your commissions have gotten into these issues and, if so, what you have concluded to date.

MR. WEISBROT: I can talk about some of those things. I think we tried to discuss the issue of accreditation in those terms. We certainly heard lots of criticism of the accreditation system as one in terms of pro formas and so on and occasional inspection, rather than routine analysis. There seemed to be a clear correlation, though, in Australia, and I'm sure elsewhere, between labs that do a vast number of tests in a particular area and ones that do it occasionally in terms of getting their quality assurance right and being able to provide all of the double-blinds and all the other tests you need to do to ensure quality. So that was another reason for our trying to centralize somewhat the sensitive tests, to say let's have these done in places where you have a lot of testing done and where you have people who are expert and consistent in analysis.

We also made quite a few recommendations to our two bodies that do accreditation -- there's a national accreditation body and there's also the Royal College of Pathologists -- about lifting their game in terms of accreditation. So we said we want to improve the processes. We want much more guidance in the protocols. We want much more attention paid to ethical concerns and counseling concerns, as well as quality assurance ones.

But on the other side of that, I think we probably simply need to resource the people who keep them in check, the consumer health groups, medical practitioners, public health authorities, and so on, the people who are consumers of the labs, to make sure that they're sufficiently up to date with the issues to provide the challenging environment for those people to do their job very well.

On the evidentiary side, we did discuss in some detail and there are several chapters and lots of recommendations relating to the use of genetic information and genetic test information in the courts. This has been a big issue in the U.S. and there has been some collaboration there across Australia and the U.S. and some other common law countries in terms of EINSHAC's, the Einstein Institute, initiative for educating judges. Just a few weeks ago, there was a major program in Australia that involved about 60 or 80 Australian judges and a number who had come from the U.S.

So there's a need in that area. We've identified a number of cases in Australia where certainly the science was very bad, the lawyering was pretty bad, and there's a need to lift the game in both those areas.

DR. BEMENT: Thank you.

DR. HOOK: Dr. Willard?

DR. WILLARD: Hunt Willard. That was terrific. Thank you both very much.

Just a point of information as I sort of balance the output that both your commissions have generated and try to see where we're going on this committee, can you give us some idea of the frequency of your meetings and also the number of full-time staff that were presumably behind you gentlemen so that you would have time for other pursuits?

MR. WEBB: Yes, certainly. That's a very important question because we can't work without adequate resources, and as I understand it, you are trying to do that.

The way we work is that we have three main meetings a year. We then have working groups who may meet on a monthly basis to produce specific reports with the backup of a secretary out of five full-time employees. So the review that I'm chairing at the moment into paternity testing, I have a

subgroup of six or seven people who are working with me. We meet once a month with Department of Health officials who act as the secretariat and at the end of each meeting they produce the minutes, they draft reports, and circulate it electronically to us. I believe our budget is somewhere around half a million pounds a year.

DR. WILLARD: And Australia?

MR. WEISBROT: The Australian Health Ethics Committee is similar in terms to your organization. It has 17 members. It's a formulaic composition. It has a lawyer familiar with health issues, a minister of religion or equivalent in the community, a scientist, a medical doctor, and so on.

I think one of the things that emerged through the process was that they were pretty amazed at what we could do by not being like that, by being a full-time commission with full-time staff and resources. So we really were the ones who drove the project, but with their very considerable informed input.

The Australian Law Reform Commission has a budget of about $3 million a year Australian, about $3.5 million, close to $3 million U.S. This was a major project for us. We had two commissioners working on it, full-time commissioners working on it full-time, myself and another, and we had about six to eight researchers and then a few support staff. So I'd estimate over the two years we would have spent about a year's equivalent budget.

We also got a grant of another half million dollars from the Department of Health to do the extensive public consultation program. So, of course, money to rent hotel rooms and travel around and talk to people and produce a lot of literature, which we did as well, like the HGC, at all different levels. So we have the big thick reports, but then it distills all the way down to one-page brochures and we even tried that thing where you give out the free postcards in coffee shops to tell people to contact us if they were interested in further information.

The experience of dealing with AHEC and the Law Reform Commission, the differently comprised and resourced ones, led to our recommendation that the Human Genetics Commission should be established on the same basis that the Law Reform Commission is.

I should say that the Health Ethics Commission unanimously endorsed that recommendation. So we said it should be a full-time body. It should contain permanent staff, although the commissioners would rotate over time, of course, and that it should be given an adequate budget. We even did some discussions of similar bodies, like the Human Rights Commission and the Law Reform Commission, to get a principal idea of what the budget would be, and we said it looked like about $8 million Australian a year would be appropriate.

DR. HOOK: I have, I think in our time remaining, Debra, Reed, and Emily. So, Debra?

DR. LEONARD: Debra Leonard. This is a very targeted question, which is, David, you say that DNA testing won't be done by accredited laboratories, and you include with QA ethics, consent, and counseling. So is it going to be, in your image of what should be done, the laboratory's responsibility to do the counseling in place of the physicians?

MR. WEISBROT: No, definitely not, but if they were going to on a fee-for-service basis do this, then they should have in place a regime that naturally referred people to genetic counselors. So it's not that they do it themselves, but that they worked in association with genetic counselors or with family doctors or whatever was relevant in the circumstances.

DR. LEONARD: Both in the pre-analytical consenting phase as well as in the post-analytical genetic -- I mean --

MR. WEISBROT: Probably. Again, it would depend upon the particular test, but by

and large, yes, pre- and post-test counseling. Again, we used HIV/AIDS as the model for that, and so we're also conceptually dealing with the most sensitive kinds of tests, but genetic tests, that would not be the case with things that were determined by the Human Genetics Commission to be more routine sort of tests that could be handled by a doctor or by the provision of literature, for example.

DR. LEONARD: One other. Have you determined how you would define "serious risk of harm" when you're allowing physicians to communicate results to -- or not results, but recommend certain types of genetic testing to relatives of a patient? How will you define "serious risk of harm"?

MR. WEISBROT: Well, there is some detailed discussion of that in the material, and of course, it's very difficult. One of the problems was in the concept of imminent risk, and so there are protocols around now that allow moving past the normal strictures when there's an imminent risk of harm. When you're dealing with genetic information, by and large, you're not dealing with that kind of imminence.

So that was one principal question. In general terms, it would be more or less what you would guess we said. We asked the NHMRC, the NIH equivalent, to do a more detailed study of this in association with doctors organizations and so on to try to get a handle on it. Our brief to them was to give them general terms of reference, saying this is what the community and the specialists are telling us needs to happen. We haven't done enough work to come up with a rule that we would recommend to government, but we think you should do that after you've done the appropriate level of consultation.

DR. HOOK: Reed?

DR. TUCKSON: First of all, David, again, it's amazing for you to get on that cab and get here and be this fresh, and we really appreciate it.

Philip, I was wondering, given how closely you're working with the government and all the various ministers, and you made that plural, what is your sense of how organized or how it is organized in the U.K. in terms of the Cabinet or the government leaders in terms of their interaction together around these issues and focus? Are you dealing with them one on one and by one or is there some central mechanism that helps the country to focus on priorities or are you sort of discovering those priorities on your own based on those?

Secondly, how soon do you think you'll have that website for consumer information and education available and can you share that with us as you develop it? Because I think that's something that we learned yesterday that we may want to model.

MR. WEBB: Yes, certainly. As regards our interactions with government ministers, the British Civil Service, as it is known, is well-established and we report both to health and science ministers. Now, it is very helpful to us that our secretariat is provided by the Department of Health. So the five full-time equivalents are actually employees of the Department of Health seconded to work for the Human Genetics Commission. So they have all the right contacts within the Department of Health for making our recommendations go to the right places and the people.

Secondly, they also have the right contacts in the Science Department. So the channels are already well-established, they're already existing, and the people who support us have the right contacts. So I think that's tremendously helpful.

As regards to website, we would like to think that that is going to be ready and set up and operational midway through next year, and we would be delighted to share that with you.

DR. TUCKSON: Is there one coordinator for those four different categories or do they, again, work independently or co-equally? I mean, is there any coordinating mechanisms for the government leaders?

MR. WEBB: Well, certainly, there are cross-government initiatives, and too often in the past there have been accusations of being in silos and not talking to one another. Now, since this

government has been in power, the concept of, as they call it, joined-up government is becoming much of a reality, with health and science ministers and even Department of Trade and Industry ministers regularly meeting to discuss issues of common interest.

DR. TUCKSON: Thank you.

DR. HOOK: Last question, Emily.

DR. McCABE: And if we could keep it brief, please, Emily.

DR. WINN-DEEN: Okay. I'll keep the question brief. I'm not making any comments on how long the answer is.

I want to know what the trick was to get the government to specifically target 50 million pounds towards improving the education of the health care workforce in genetics, so that they could be informed and intelligent about how genetic services are provided, because in the U.S. this is apparently mostly a grassroots effort and not a coordinated government initiative.

MR. WEBB: Well, this is something that we have been saying for the last five years. So it's not an instant turn out of 50 million pounds.

We've also worked through such groups as the Royal College of Physicians, Royal College of Surgeons, Royal College of General Practitioners, and the nursing organizations. They all recognize that they need better education and that genetics is infiltrating every part of medicine. So with their help -- and the Department of Health recognized that that is important, too.

If I put my cynical hat on, I would say that perhaps some government ministers think that genetics is a sexy subject and that if they are seen to engage with it, it may be good for their image and ensure their longevity in office.

DR. HOOK: Thank you very much, gentlemen.

DR. McCABE: Well, thank you very much. On behalf of the committee, I would to thank both of you for coming to visit with us and tell us about the exciting things that you're doing in both Australia and the U.K. I hope that we can use this as just the beginning and keep lines of communication open, share reports, and continue to have dialogue, and also, with our past history with Canada, I hope we can do the same. So we look forward to continuing to talk with you, and thank you again for coming to visit with us.

We're going to take a 10-minute break. Before we do that, though, I just would like to briefly meet here with Drs. Boughman, Shekar, Cooksey, and Willard, if I could, please.

Thank you.

(Recess.)

DR. McCABE: Well, in June, we talked about the importance of ensuring that our health professional workforce was well-prepared for genetic knowledge, services, and technologies, and there was concern expressed by our members about the role of genetics in health and disease, that as it increases and the technology advances, that the health professions may not be sufficiently well- prepared to integrate this information.

We decided it would be helpful to learn about federal and private sector activities in this area. HRSA, since they have a leading role within HHS in addressing workforce issues in the health professions, we asked Dr. Sam Shekar, HRSA's ex officio, to take the lead in compiling information on federal efforts.

Dr. Joann Boughman, executive vice president of the American Society of Human Genetics, who testified before our committee in June about the importance of preparing future genetics professionals, generously offered to take the lead in compiling information on various professional society activities. I should also note that Dr. Boughman served as chair of the SACGT Education Work Group,

and she will be sharing some of that information.

In addition, we will also have the benefit of presentations from Dr. Judith Cooksey, who is principal investigator of a federally-funded genetics workforce study, and then after lunch we will hear from Joe McInerney, director of NCHPEG, the National Coalition for Health Professional Education in Genetics, and Robin Bennett, past president of the National Society of Genetic Counselors.

This series has been organized to give us a better overview of public and private sector efforts in these areas, and I'll thank in advance Drs. Willard and Tuckson for serving as facilitators. Thank you.

So with that, Dr. Boughman.

DR. BOUGHMAN: Thank you.

In the next few minutes, what I would like to do is frame some of the issues and, in just a few minutes, save you many hours of reinventing some of the wheels or rehashing again some of the issues that have been talked about for a great deal of time.

The challenge we have is enormous. Genetic knowledge and technologies, as we know, are moving very, very rapidly. We also know that the integration of genetics into health care and public health practice is actually driven by three main forces. First of all, these advances. Secondly, consumer and patient demand, but we always have to include cost considerations and the new emphasis on prevention.

These forces are what are driving the health care professionals to need to in fact be so well-educated, and in the webcast session that I watched yesterday -- all of you were in my office as well as here, two places at once, at least -- there was a question of is it adequate? The answer is no? Is it outrageously inadequate? Becoming so if we don't catch up quickly.

In fact, I'm going to come back later this afternoon and say that one of the aha moments that we had in the SACGT Education Work Group was when one of the private practitioners who is a family practitioner said, "Give me all the curriculum you want, but what I need to know is what should I be doing in my office differently tomorrow."

It all comes down to that and until we can reach that bullet, then in fact we have more challenges before us because health professionals are the ultimate arbiters and we know that major gaps exist in health professionals' knowledge and education and training in genetics, and you'll see later today with some data that actually the age of the clinician is inversely correlated with the amount of genetic knowledge they have and use on a daily basis. There are gaps in knowledge, education, and training that will limit that integration into all aspects of health care as we know it.

We have several key stakeholders that we'll be hearing about in various aspects. I'll be talking about genetic specialists on the M.D. and Ph.D. side. We'll hear about genetic counselors. I will also be talking about other physicians, several of us will be mentioning other public health professionals, and then we will also hear about nurses and allied health professionals, always with the background of patients, consumers, and the general public as our ultimate stakeholders.

There have been several prior national efforts, and I'm sure you all have had materials given to you. I'm going to point out what I believe, after our years in SACGT and the Education Work Group, the real highlights were.

There was a National Academy of Sciences group in '75. The IOM had the "Assessing Genetic Risks" in '94. There was the Task Force on Genetic Testing in '97. IOM came back again and in fact had their issues on educating public health professionals in 2002, and then the SACGT Education Work Group in 2001 and 2002.

Even in 1975, the National Academy of Sciences group said medical school curricula

and continuing education should emphasize genetics, that physician knowledge of genetics should increase orientation toward preventive medicine, and that schools of medicine and public health and allied health should be provided support for programs to set these standards and train personnel.

The Institute of Medicine -- I'm just going to highlight some of these -- they again said we needed to train more professionals and several of these other things.

What I have done in the materials that you have on the slides is try and indicate to you some of the progress that has been made and some of the groups that have been working on these issues. We have genetic counseling programs. We have ABMG, the American Board of Medical Genetics, training programs, and you'll hear later today about some of the programmatic efforts to teach the teachers, if you will.

It was also said in 1994 that we needed to maintain genetic counseling programs and also to develop programs for single-gene disorder educators and counselors, and we believe the shift has changed from that focus.

It also suggested that consideration of genetic counseling and education should be a part of the standard of care. This, of course, would fall into the area of professional guidelines, as well as changes in curriculum that we will talk about later today, too. Also, that CME programs should be expanded.

They went on to say that we should simply reform education, which is why the breadth of our challenge is so huge. However, that's what we're doing because we're not only talking about the science. We are talking about looking at medicine from a genetic point of view, including the ethical, legal, and social implications. That is an educational reform across the board. Progress is being made in many of these areas, and I've listed several of the groups that are at least chipping away at this challenge, and we'll hear more about that later today as well.

We come again to CME programs for all professionals, not just the physicians. At that time, there was concern and there is continuing concern about recruiting more minorities, and there are federal programs that help us do that, and even in 1994, there was the admonition that we should develop and evaluate online systems of education, be they programs, full curricula, or simply websites themselves.

They went on to talk about genetics curricula that we'll talk more about later today, and to develop tools to enable professionals to acquire competence. We'll come back to this issue as well, but there are several organizations that in fact are providing some of those educational programs.

We need to strengthen genetics training programs in schools of nursing, social work, and public health, and in your handouts, for the members of the committee and others, in fact we believe that there are several of these groups that are working very hard toward this goal.

In 1997, that task force again talked about increasing genetics curricula in medical school and residency. There was a slight change here, though, to talk about enhancement of licensure and certification. This now involves the states, the boards, and professional licensure organizations, and we will in fact address that later today.

Also, they were very concerned about increasing the continuing medical education, both federally-supported programs and those based in the physician and other professional programs, and that it was incredibly important to develop the competence of physicians and laboratorians. We'll come back to this later, but this includes the maintenance of certification and in fact deeming what competence is in relationship to the individual professions as it relates to genetics. Of course, we also have CLIA and the accreditation of the laboratories and the laboratorians there.

The Institute of Medicine added that schools of public health should be teaching students to think genomically. Again, an admonition for reform in education to include the ethical, legal,

and social issues, and the Institute of Medicine was the first national body to in fact affirm the NCHPEG and the CDC core competencies that in fact could be used to guide curricular development.

The Education Work Group of SACGT continued on in looking for more innovative approaches to integrating genetics into the continuing care because at this point, from the 1997 to the 2001 time frame, we are now moving from genetics as a group of rare disorders into the mainstream of all medicine, and our focus then was on integrating across all the traditional departments and disciplines.

We thought that federal funding could be directed to the training of genetic specialists and general genetics education for all professionals in a variety of ways, and we'll talk about a few of those initial suggestions this afternoon, and I hope that that spurs many more suggestions from the group around the table.

Our ultimate goal, restated from the Education Work Group, was an educated public. At least, a public educated enough to receive the information that they would be receiving from their physicians and other health care professionals.

We also tried to put some emphasis on creative, interactive, and accessible programs. The Internet is not what it was in 1997 and we need to be utilizing the technologies that we have.

SACGT also emphasized collaborations in outreach, and we'll have a few of those examples this afternoon, plus the web-based kinds of things that we could use the expertise of multiple groups to come together to utilize the web as a resource for all of the groups, all of our stakeholders.

These are just a few of the highlights, but we've been at this for a very long time. There have been a lot of people thinking very hard about these things.

We are, though, in a new phase. We have the Internet, we have an awareness now that was not present five years ago, and we have an opportunity now to capitalize on that very basic information that we have. So this morning, we're going to have some discussions about workforce and some of the issues about workforce, and Dr. Shekar's going to share with us some of the many federal initiatives that there are out there. I think that by the end of the day, you will feel pleased that there are a lot of people working very hard, but you're also going to help us be able to find where the big gaps are and how we can muster more resources to help us do better.

With that, I think it's Dr. Cooksey that's next, correct?

DR. McCABE: No, it's Dr. Shekar.

DR. BOUGHMAN: I'm sorry. Dr. Shekar.

DR. McCABE: Thank you very much, Dr. Boughman.

Dr. Shekar is the ex officio representing HRSA and has brought together to report to us the activities of the various agencies that he will tell us about in that report.

Dr. Shekar, thank you.

DR. SHEKAR: Thank you very much, Dr. McCabe. I appreciate being here today. I was asked to speak on behalf of the ex officio federal agencies of this committee regarding the issue of genetics education and training professionals. I thank the committee for the opportunity.

I thank my colleagues in the agencies who took a lot of time to help focus and to help respond to the survey. It provides a unique opportunity look across 16 agencies participating as ex officio members of the committee.

A special thank you to the SACGHS staff for putting that survey together, compiling the data, and preparing the report, and a very special thank you to Dr. Suzanne Feetham for her continued support and guidance on our own HRSA genetics activities and helping to represent us here the many times that you've had these meetings and continue to.

As reported by the Institute of Medicine and others, there is at least a 10-year time lag

of translating relevant science from the laboratory to general application across the people. Some examples of the rapid advances in genomics with application for health were presented yesterday and by Dr. Collins in the first SACGHS meeting. With the exponential growth of genetic science and genomics, this lag time is not going to be very acceptable. In order to be positioned for 2010 and beyond, we must address this now, for there are over 11 million health care professionals with over 200 professional titles who must eventually serve as some of our principal partners in translating genetics into everyday practice, as Dr. Boughman had mentioned earlier with her apocryphal example of a family practitioner asking, "What does this mean now? What does this mean in my daily practice?"

At SACGHS' inaugural meeting in June, the committee identified genetics education and training of professionals and the adequacy of the genetics workforce as areas of interest and priority. You requested additional information about federal and private sector efforts to address that need.

It's important to note that the primary role and responsibility for education and training of professionals lies in the non-federal private sector, with the academies and with the professions, and we absolutely understand that and want to serve as partners with you to make this happen.

You'll be hearing about many of the genetics education efforts being undertaken by the private sector later today. On behalf of my federal colleagues, I'm pleased to present the information you requested on the federal activities in this area.

A copy of the survey instrument and three parts of the agency submissions are in Tab 6 of your briefing books. Around the meeting table are several copies of each agency's entire submissions for pleasure reading tonight, including the detailed descriptions of their individual projects.

The survey consisted of four parts, a Part 1 narrative summary; Part 2, which was lists of relevant activities; Part 3, detailed description of each activity, including target audiences, partnerships, and impact; and Part 4, funding of these relevant activities from fiscal year 2000 to fiscal year '03.

The intent in asking for funding levels was to obtain a general sense of the total investment across the agencies, not to make comparisons across the agencies, but to give a general sense of where we are for the federal government.

It is important to note that there may be some underreporting of activities because of a couple of reasons. First is that there are other federal agencies, as we all know, that are not necessarily ex officio members of this committee and obviously were not included in this survey, but could in fact be doing genetics activities. Second, the time provided was extremely short in order to meet the deadlines, less than a month, so therefore the breadth and depth of activities in genetics and genomics may not be quite complete through some of even the agencies surveyed.

As you know, agencies were asked to describe the mission-relatedness of the efforts, the decisionmaking criteria, specific needs that were being addressed, and results of any program evaluations.

Seven agencies reported having over 180 relevant activities, and you see some of them listed here, and this being Washington, we can only describe things in acronyms. So there you go.

Eight agencies reported having no relevant activities. It's important to note that the non-reporting agencies are nonetheless carrying out critical education and outreach activities in a broad range of areas which include, as part of it, some genetic components. For example, the Office for Human Research Protections , OHRP, is undertaking important work with respect to revising guidance for genetics research and other research, the Office of Civil Rights is conducting critical outreach and education around HIPAA privacy regulations which have some impact on genomics and genetics, and so forth and so on.

The fundamental need addressed by the federal efforts was simple and clearcut: facilitating the translation of genetics into practice.

As noted, the Human Genome Project and other genetics research has increased the pace and amount of genetics research and created higher expectations for the application of genetics knowledge and technologies in medical, health care, public health, legal, and other arenas. The federal government realizes that the science of genetics has advanced well beyond the genetics knowledge base of many health professionals and that professionals play and will play a greater critical role in integrating genomics into practice.

Questions were raised yesterday, as I understand, as to what is different about this genetics knowledge. The premise from the federal agencies is that genetics/genomics will become a central science for the practice of health professionals, increasing our knowledge of mechanisms of diseases and conditions for diagnosis, treatment, prevention, and health promotion, and providing a new lens for all health professionals, particularly those in primary care. As presented in the pharmacogenomic examples yesterday, there is opportunity through that to provide more focus for individualized or targeted care.

Agencies are thinking comprehensively across the system, choosing activities in a systematic way, and integrating those efforts across their agencies, doing this through partnerships with local, state, and federal organizations, developing expert recommendations, working with stakeholders, and assessing impacts of funds.

An indicator of the complementary roles that I've mentioned is that from a rough hand count of the responses from just three agencies alone -- CDC, NIH, and HRSA -- each reported partnerships with other federal agencies for about a fifth of the activities that they engaged in, and the partners most frequently cited were those agencies themselves, along with other agencies, such as the Department of Energy. Most activities are complementary, due to significant agency cooperation which will increase over time, and also target a broad range of audiences.

Clearly, health professionals are the primary target, but what's of interest is many other professionals are also targeted as we think about, as Dr. Boughman mentioned earlier, our ultimate stakeholders, the general public. So clergy, journalists, teachers, underserved populations, judges, lawyers, law enforcement personnel, all these individuals, all these professionals, are also targets of genomics and genetics education and training, and that will also increase over time.

Most agencies support a variety of activities in education and training, including dissemination, educational outreach, consensus building, and generalized training, either directly or online.

An example is the Genetic Interdisciplinary Faculty Training Program at Duke University funded by HRSA's Bureau of Health Professions, which, primarily through four approaches, provides greater understanding of genetics and genomics, focusing on curriculum revisions, presentations, publications, and web-based resources. We were able, through this activity funded at around three-quarters of a million dollars, to arrange for 22 regional, national, and international presentations and articles, 34 courses, graduate program lectures that were incorporated into faculty teachings by three interdisciplinary teams, three research studies, over 2,000 hits on an online module, 3,000 brochures distributed, et cetera, et cetera. It's important to note that the teams were interdisciplinary and included members from nursing, medical, and many other professional groups.

This grant included, as I said, 16 interdisciplinary teams from 13 states. It started off with six states in 2002 and expanded over time and now has gone beyond 10 teams in 10 states, as noted on the map. There was also an earlier activity called the Genetics in Primary Care Project, which was a partnership led by HRSA, but also with very strong partnership from NIH, CDC, and AHRQ, where 20 physician teams in 19 states provided that training in genomics, and now there's a website being developed to expand into other disciplines.

The Department of Energy's Genetics Adjudication Resource Project at the Einstein

Institute for Science, Health, and the Courts worked on a number of projects to help inform judges about the advances in genomics, and it funded 38 workshops to train 3,300 judges through a $1.5 million project to have them understand the implications of genomics for society and law. This was a model that was then replicated and advanced in other programs, such as the Judicial Conference for the upper New England states, as well as the Genetics Conference for Clergy organized by Catholic University focusing on the ethical, legal, societal, and personal challenges that may arise from genomics.

The total funds spent by those surveyed in the agencies were roughly over $100 million, $102 million, to be exact. HRSA carried out the greatest number of activities in health professional education and training in terms of funds spent. NIH had the highest figures in all four fiscal years. Again, this is a retrospective estimate of discretionary actions taken over a very short time period in terms of the survey. So again, there may be some underreporting going on, but federal agencies have generally not made specific resource allocations to genetics education activities, but have incorporated that among their other training and education missions.

The very good news is that this funding represents not only a significant amount across four years, but an ever-increasing amount, as you can see from this slide. A steady increase in the overall funding at federal levels for activities in education and training in genetics and workforce analysis reflects the emerging importance of genetics in the clinical sphere.

The federal government realizes that the science of genetics has advanced beyond the practitioner's general knowledge level in genetics and that health professionals play a critical role in integrating genomics into practice. In response to these developments, federal agencies, as you can see, are taking and will take an even greater leadership role in helping to advance the training and education of professionals in partnership with societies and private sector colleagues.

Of note is our own little Hawthorne effect through the survey. Participation in the survey is resulting in increased dialogue, even more increased dialogue, among some of the federal agencies that participated.

A few summary observations. Genetics education, training, and workforce analysis efforts are supported by discretionary funding. Discretionary funding is very flexible and is critical to allowing agencies to respond to new needs as they arise. However, as we know, when multiple needs arise simultaneously, that funding has a degree of support concerns that then needs to be looked at.

Under workforce analysis, it is very important to understand who is out there to provide that genomics education, but it is not the only factor to be thinking about. In fact, the pace of scientific discovery and the general applicability of genomic advances -- the family practitioner example that was quoted earlier -- to common diseases and conditions will also help determine the health workforce balance that's needed to provide the adequate support to serve the general population, and obviously, additional evaluation efforts at the project, and possibly agency, level may be needed.

It's clear that agencies are helping to look at the genetics evolution and revolution, and two major action items were identified in the survey by the agencies as helping to really push this whole focus forward: helping with our partners to identify the appropriate use of new genetics knowledge and technologies and also helping with the across-the-board integration into general practice across multiple professional groups and across multiple locations. Are the issues the same in urban areas as they are in rural areas? Are they the same for different populations?

In conclusion, the agencies look forward to working with SACGHS and the private sector as we all proceed with these great advances in science and, ultimately, practice to help improve the health of this nation.

Thank you.

DR. McCABE: Thank you very much, Dr. Shekar.

Now we'll hear from Dr. Cooksey, who is the PI of the Genetics Workforce Study funded by HRSA with support from NIH and CDC, and I thank you for participating in this meeting.

Please proceed.

DR. COOKSEY: Thank you. I'm delighted to be here. I'm presenting work that has been carried out and planned for for the last three years by a multidisciplinary group of researchers and consultants across universities in the country.

I'd like to give you a brief overview. I'll begin with a discussion of what is workforce research. I consider workforce research as one type of health services research, and we'll briefly describe what HSR, health services research, is.

As we've done this research, we have encountered problems in the field with some of the terminology that's used, and I think there may be time, as others have discussed, to really clarify some genetics-related concepts and address some of the issues that were discussed yesterday. I think Dr. Willard brought this up several times. What is special about genetics? Is genetics services considered special or is it becoming part of health care?

In that vein, I'll discuss an Institute of Medicine study on primary care. I'll present the type of workforce research that we have engaged in, which I would call functional workforce research, which looks across professional groups and pays less attention to categories of providers, but looks in the context of the health services being delivered. I'll describe the University of Maryland-Baltimore study and propose some priority areas for future research.

Traditional workforce analysis tends to be somewhat quantitative. It's done by professions. It is also done by medical geographers, medical sociologists, and health economists.

Some purposes are to assess whether we have a shortage situation. Is the supply greater or less than the demand? With shortages, supply is less. With surplus, supply is greater. That's an unstable state and is usually corrected by market forces.

Some of the most difficult workforce research to do, and usually the questions people would like answered, is forecast the supply and the demand by 2010. Will we have enough genetic counselors, geneticists, well-trained primary care providers, or others? I'll show you some examples of some of the best forecast work that's done and some of the limitations.

Most workforce research looks at a specific health professional group. I conducted this study at the behest of HRSA and particularly the Maternal and Child Health Bureau. They had a specific question. Should they support funding of expanding genetic counselor programs?

This was an easy one to do in some ways because the genetic counselor group, which you'll hear about with Robin Bennett's talk this afternoon, is an articulate, fairly homogenous group that has studied itself through survey research, has written about its professional activities and professional roles, and was very interested and has a strong sense of professional identity and plays a very important role as one of the genetic specialty groups providing services.

But the market had not been kind. The study was done in 1999 and published in 2000. Genetic counselors were underemployed in some market areas. Reimbursement from health insurers and managed care was uncertain, and they worked in academic health centers, which have continued in the last eight years or more to see financial crunch.

We expected demand to increase, but could not come out with a recommendation at that time to support expansion of programs when people coming out of programs faced underemployment, but that profession could not be studied in isolation and we recommended a further study. We also felt it extremely important to educate health insurers and managed care plan payers about this, and the SACGT

committee had some wonderful panels that brought groups together.

Another study that we conducted at HRSA's request, but really at Congress' request, was to look at whether there was a shortage of pharmacists in 2000. We confirmed that there was, that the demand outpaced supply, despite supply growing.

A useful measure of demand there was prescription volume. Sixty percent of pharmacists work in the outpatient area. There was data on prescription growth, which has been phenomenal. Prescription expenditures have been phenomenal.

When we began this genetics study, we thought it would be wonderful if we could find a measure that would be countable and would measure demand for genetic services, and we somewhat naively thought we would look at genetic testing volume, which, as mentioned yesterday, is not a number that's available, but I think it's still something worth looking into.

Other useful health workforce reports simply give counts and information about who is going into the profession and who is being trained. This is a wonderful resource that's been produced by HRSA by the New York Health Workforce Center that looks at for every state and for the District of Columbia information on about 25 professions, mostly on the supply question, and a new report has come out by the Washington Workforce and Rural Health Center that looks at state-level supplies in rural areas, chronically underserved areas for many professions.

The Bureau of Labor Statistics does the most complex forecast of workforce demand for all occupations of the nation. They collect state-level data through surveys of employers on hundreds of occupations. They organize it at the industry level. They use complex economic modeling to look at where growth is expected by industry and then by occupation. They present information that looks at the current employed workers and the projections for 10 years out. A primary user of this sort of information are guidance counselors in high schools and in other areas because they're projecting workforce need at 10 years.

We've met with them. They've been very involved and very helpful with some of the work that we do. They cannot use their data sources to predict shortages very well and they cannot zero in to very small professional groups, such as genetic counselors or geneticists.

Health services research, which I think health workforce research fits into, has been recognized as a field of research for about the last 15 years. The issues that are studied by health services research -- and the researchers are principally social scientists, although you're seeing clinicians get more involved -- is the way health care is organized, how it is financed, the different delivery models, and the staffing and the workforce.

Access to care is an important component of health services research, and outcomes and quality of care is an area. With the outcomes and quality work, AHRQ, the Agency for Healthcare Quality and Research, is one of the funders, although, as we looked broadly at the national meaning for health services research, their abstracts presented and their talks for about the last 10 years, very, very little coverage of genetics. Occasionally, there would be a paper or an abstract submitted on genetic counseling and a few papers on pharmacogenomics, but no real health services research in genetics. So there's an immense void in health services research in this area.

Switching to genetics now, I think in the field there's a lot of confusion. When we start thinking about talking to people, oncologists, nurses in oncology, many other groups don't consider what we call genetic tests as a genetic test. They think of it as a diagnostic test, a test to help evaluate.

Genetic conditions. Hemophilia, which is a clotting disorder that's X-linked in inheritance, sickle cell anemia, which has been recognized as an inherited disorder, are hematologic conditions to the world by categorization, by billing, by which specialty manages this condition, and yet

they're genetics conditions. I think that issue needs to be clarified in the work that we do with workforce and the work that's done with health services research and in thinking about how the greater world sees this integration that's happening. Similar issues will come up and have been discussed by this group.

In defining and describing genetic services now, I put on the table for your consideration an Institute of Medicine study that was commissioned and completed in 1996 after two years of work. This was on the topic of primary care. Primary care was a very hot policy issue around the Clinton health reform movement, and the market responded despite the lack of passage of health reform.

Barbara Starfield is a Johns Hopkins researcher, a pediatrician by training, who did some important pulling together in a book that she wrote about some of the concepts of primary care. The Institute of Medicine conducted a study that looked at primary care, and one of the handouts has some of the findings and recommendations from that study.

What I see as some of the issues that you've been discussing, in that summary you could substitute the word "genetic services" for the word "primary care services" and see that a lot of similar issues were addressed by this group that has the ability to conduct more comprehensive studies.

As was mentioned by Dr. Boughman, in 1994, IOM reported on an extremely helpful study that looked at assessing genetic risk. It focused heavily on genetic testing. It helped clarify definitions. It had a wonderful chapter on the genetic personnel. We've not been able to find evidence that IOM has done a study specifically focusing only on genetics since then, so it's sort of a decade later look.

The Institute of Medicine is charged by Congress to look at public health and medical issues, and they cover a broad array of issues and they do superb work. They tend to take a little while to get the projects done. That's one of the downsides, but they're excellent.

Now, shifting to clinical genetic services, what I'd like to cover very briefly is how we've looked at what are the services included, how are they organized, where are they provided, and who provides the services? The more difficult questions that we're touching on with our work are looking at emerging models of services, information generated, and we're not really addressing cost, but it's out there. I'll start with some preliminary findings and then sort of show you how we got to this place.

Clinical genetics is a very, very small specialty area of medicine and health. It has been extremely small, and that explains a lot of what we're seeing right now. Numbers of specialists are small. Training programs and trainees produced each year are small. Geographic distribution tends to be in urban areas. It's been a quaternary or tertiary care function, although it has had a strong public health component, and that's helped get services out to rural and less-served areas. There is limited diversity in racial/ethnic and just backgrounds among the professionals and the specialists.

What we've found and others have identified is genetic test availability is influencing demand for services. So if we had some ways to keep tracking that, and we're trying to get to that, that would help one get a little heads up.

Diffusion of medical innovation and technology. Much discovery and translation work occurs in academic medical centers, which train specialty physicians, fellows in cardiology, in GI, in neurology. They learn the new technology, whether it's lithotripsy or genetic testing for neurologic conditions. That's learned in academic medical centers and the group that learns that are the specialty groups.

So the diffusion of using this information technology will be in academic medical centers and out. So it will be with specialists. Principally medical specialists, but also nursing specialists, pharmacist specialists, and others. The diffusion out to primary care physicians takes longer and needs a lot of support and help.

The fourth concept that is extremely important is you can't beat the market. Market

factors are extremely important drivers of how health care is organized, where it's provided, who does what. Financial incentives or the lack of financial incentives have a lot to do with the providing of services. There's competition in the market and that affects it. Reimbursement design is critical, and our reimbursement design system is flawed for health care.

Settings for genetic services. This is a structural look that helps us organize our thinking. The number of ovals has been growing over the last year. Different places. Academic health centers are in the center because that's where you have the specialists of all sorts. That's where people come who have had rare conditions or they come to outreach clinics that are sponsored by state programs, by Medicaid often, that are located more.

So there's a lot of outreach work that's going on in genetics that's highly admirable, but you have commercial labs getting more into more full-service operation. You have freestanding diagnostic clinics happening, particularly in the neonatal area. You have market-driven -- i.e., financial incentive-driven -- change that's happening. You have policy change, particularly with state-sponsored and Medicaid conditions, and you have a lot of community-based services coming up, as you've recognized. Hospitals are a major source as well.

So anyhow, we've taken a bit more of a functional assessment of the workforce. Who's qualified? We're not judging that, but the professional associations do. What roles do various providers say? In other words, we're looking at what's happening. And do the roles differ by the categories of services? We've identified five broad categories of the local. Who else is there?

I'll go quickly through some of the categories of genetic services and just ask you with each of these to look at the provider and identify the profession. Is this a medical student? A pediatric neurologist? An ophthalmologist? A family physician, pediatrician, geneticist, genetic counselor, physician assistant, nurse practitioner? It could be any.

Genetic counseling as a skill set and a counseling service has been well-organized in defining and consideration by the genetic counselor community, but they and others recognize that genetic counseling is a service that many providers provide. Is this a nurse midwife? A nurse practitioner? An obstetrician? A genetic counselor? A family physician? You know, what is the profession of the person providing genetic counseling?

Genetic testing. The laboratory direction is extremely important. Pathologist, Ph.D. or M.D. geneticist, the staff that are involved. This is a little more defined.

Another important phase of genetic services right now is diagnosis and risk assessment, and then management plans. We don't have curative therapy for many genetic conditions, so management becomes important.

The people who make the diagnosis and help with the risk assessment again are not always physicians. There can be many different professions, and it's changing. We're seeing less focus on genetic conditions in children, such as Down's syndrome. You know, historically, Down's syndrome was an important condition to recognize and to offer treatment for. It didn't need a genetic test to confirm it. I mean, a clinician can identify it, but the management is extremely important and early intervention and looking for system involvement.

Family cancer risk assessment is an emerging area that many professional groups are taking up, and then we've discussed the rare and complex conditions where you need specialty team involvement.

Other services. Sadly, there are not yet a lot of specific therapeutic interventions. As those come available, the market will take them up and it will have an immediate effect on demand for services.

Care coordination. There's an important role for nursing and others. Also, the supportive and rehab therapists. So how far do you define genetic service providers? And then family-related genetics issues and follow-ups is extremely understudied and underprovided at this point, I believe.

What factors really determine who can provide services? What I'd like to say is we're used to thinking about the first and second factor, but I think all of the rest are extremely important. These are some of the areas that our research is looking at. Local market factors, setting and organizational structure. Reimbursement has certainly been discussed. Interprofessional collaboration. The political strength and vision of the profession and advocates and public interest. Advocates can change reimbursement policy.

Specifically, our study looks at the way genetic services are now delivered across the country. We want to describe models. We'd like to look at how it's organized, who staffs it, and how it differs, identify factors driving change, and establish a foundation for further services research to inform educators, policymakers, and professionals.

Some of our research studies specific professions as entities. We've conducted a study, and I'll show you some of the data, of the medical geneticists, which are M.D. and Ph.D. board-certified geneticists.

The National Society of Genetic Counselors conducts biennial surveys of their members, and Robin is going to present some of that information this afternoon, extremely important time/time data available.

Very little research done on nurses in genetics and, as was mentioned yesterday, this will be one of the key groups that we look at, but we'll look at a very tiny group of nurses. Nurses who are in ISONG, who are self-identified nurses, in the U.S. maybe number 300 who provide research, education, services, and policy service as well.

We will also look at primary physicians, and we've not been funded to look at medical specialists and subspecialists, although I think this is important, as are looking at other professional groups.

The first study, and I'll try to just go extremely briefly, looked at the medical geneticists. These are Ph.D. and M.D. certified physicians, certified specialists. The bottom line, very small numbers, somewhere between 900 and perhaps 1,000 active board-certified geneticists practicing in the U.S.

Five-hundred or so are Ph.D.s. Many of the M.D.s, a quarter of the M.D.s almost, in addition have a Ph.D. Fifty percent women. Mean age 52. Few members of minority groups. Highly educated. The M.D.s, the first level of training is pediatrics, much less internal medicine and OB and pathology. Then the laboratory genetics, these are the three subspecialties of laboratory genetics.

We looked at the aggregate time that this workforce indicates they spent with various efforts, and I think that one of the most telling numbers there is patient care. You see the M.D. geneticists, 43 percent of the time aggregate of the M.D. geneticists is in patient care. The rest is in other activities. That's their service function, their patient service, whereas Ph.D.s, the equivalent would tend to be clinical lab time. So the M.D.s spend about 50 percent of their time in clinical patient care or lab and the Ph.D.s also about 50 percent. The other 50 percent is spent in other activities.

Where do people work? This helps explain why we see the time spread the way it is. The M.D. geneticists are not M.D.s working in a private practice office, where 80 to 90 percent would be patient care. They tend to work in academic health centers, where they have many roles that they're required to play, or they work in lesser amounts in laboratories and practice in other areas.

When we look at just the geneticists who say that they provide patient care, we see their patient mix, which is the first three boxes, for all M.D. geneticists -- and we also looked at this 70+ percent

that are pediatrics-trained geneticists, and then the internal medicine and OB geneticists, because we thought there would be differences, and there are -- about 74 percent of the care that's provided in aggregate is with children and infants and 11 percent with reproductive. That's the history. The history of genetic services has been with children and reproductive areas. Adults is a fast-growing area.

Waiting time. If you're concerned about shortage right now, you look at issues of waiting time to see new patients. Fifty percent can see a new patient within a month. Several of that can see them within two weeks. I don't know how to interpret that. We can look at this further to tease it out by location, by setting, by other things.

The most striking number on this chart to me is the number of new patient visits per year, 240. Robin will give data on genetic counselor visits per year, which was about 350 and has gone up quite substantially. You can do the math to figure out about how many new patients are seen by geneticists. Some of those new patients are also the patients seen by genetic counselors, so you can't sort of add those together to get how many new patients.

So we have a professional group that is small and has limited patient care time. The majority are pediatrics-trained who provide direct, hands-on patient care services. Their productivity is very low and, from a workforce perspective, that's a concern.

They are time-intensive visits, 75 minutes for a new patient. You know, you hear the 10 minutes per patient, maybe 20 minutes to 30 minutes for a new patient for a family practitioner or an internist. So there is potential there for efficiency gains.

To compare numbers, these are board-certified. About two-thirds of the physicians in the U.S. have at least one board certification. All physicians, and this is AMA data, in 2001 numbered over 800,000 and 560,000 were board-certified. So you can see the numbers there, 97,000. Again, for geneticists, active board-certified, we're using maybe 1,000 M.D. geneticists.

The second part of our study, which we really feel is the core study, is looking at what is going on now in delivering genetic services in several areas. It doesn't matter who is providing the services. That's what's we're trying to find out. The methodology that we use is based on a really well-grounded research that also uses survey research and in-depth case study of representative communities in the U.S.

This project is called the Community Tracking Study, CTS. It is sponsored by the Center for Health Systems Change. This is a group that's based in Washington, D.C. and has been entirely funded by the Robert Wood Johnson Foundation, over $100 million in the last eight years, an extremely important group that grew out of the failed Clinton reform plan when leaders of health services research said we need timely information to look at how health care is changing across the country. We think it's changing in different ways in different communities because of the history of providing health care, the strengths of insurance companies or managed care, and other factors, and we'd like to study that.

So they study it with survey research in 60 communities and they select 12, three from each region of the country, where they do in-depth case studies with interviews of people principally from physician groups, hospitals, managed care, health insurance plans, public sector, sort of the safety net providers, state Medicaid programs, community health centers, and others.

They report. Their website is extremely useful. They make their survey data and this data publicly available. They report. They have done these in-depth case studies since 1995-96, so they're in their fourth series of interviewing originally 40 people, now 90 to 100 people, in each of these communities and writing up these interviews, so they can look across communities, they can look across time, and they can see what's changing and why it's changing, some of the underlying factors. We've modeled our research after that.

We think that genetic services is grounded in overall health care organization, finances, and policies. We think that genetic services is a specialty service that's going to infiltrate across, but the way it develops is going to be based on it is one of many types of health services.

What's special about it are some very significant issues. It crosses all population groups. The genetic technology advances and clinical applications are potentially paradigm changing in the way we think about health, disease, health promotion, managing conditions, diagnosing conditions, but professional practice and a varied adoption of the genetic framework is what is evident now. Consumer awareness and interest, as you've discussed, is varied, and we think local health market factors are extremely important to study. So that's what we've looked at.

We've conducted at this point about 100 interviews, and some of them have been national to get a perspective. The four communities that we are focusing on, based on sort of a random selection of the 12, are Cleveland, Miami, Orange County, California, and Boston. We've done most of our interview work so far in Cleveland and in Miami, and we see differences in the way genetic services are organized for a variety of reasons. Certainly, across all communities, new genetic tests affect things.

Individual leadership and institutional vision makes a huge difference. Sitting at the table is someone who gave that leadership and institutional vision to Cleveland, and he moved and there's a big hole there, and Duke and North Carolina are doing some exciting things.

We've seen that in other places. Someone has moved into Miami to bring sort of a new vision to genetic services. Genetic services in Miami have been underdeveloped, and I think Skip Elsis is going to make an impact there. It's still a little bit early.

Geneticist roles. To me, as a workforce researcher, it was a little weird to hear pediatricians counseling on adult breast risk assessment. These are pediatric geneticists, but it kind of goes against the conventional, very much silo-organized, that breast cancer assessment would be done by an oncologist of some sort or an internist or a family practitioner. A pediatrician was a bit of a stretch, but pediatric geneticists have stepped up to that and certainly genetic counselors have stepped up to that, the change that's happening.

But the other thing that we're beginning to see within the geneticist community is genetics is complex and they are subspecializing in specific conditions, and that's a very important issue. You lose boundaries to some extent. Are they the expert for the country, for the region, or for the condition? I think that's an interesting observation we'd like to learn more about.

Laboratory competition. Clinical genetics laboratories, to a great extent, started out in academic health centers and they fund some of the genetic counseling work and other provider work that's done. They have the potential and have been revenue generators for genetics programs in academic health centers.

That revenue is going to commercial genetics laboratories. It's a market factor. Large commercial, large national or regional genetics labs, can cost-compete with academic health centers, and particularly when there is managed care contracting involved, they can direct, and people literally overnight can lose half of their laboratory test volume if a contract happens that shifts things one way or another. So I think that's been an extremely important issue for academic health centers and has undermined some of the limited funding that was there for clinical genetic services sort of fund sharing.

The other thing is genetic services is an outpatient service to a great extent. There's work done in the newborn nursery and other things, but you sort of look at hospital services or outpatient, and it's heavily in the outpatient arena.

It's a cognitive service. It gathers information and shares information. It doesn't have high cost and high revenue-bringing-in procedures, except the testing, which is gone, which is leaving now.

So it gets low reimbursement, and that's a real issue.

Safety net funding and services, just from looking at a couple of states, are there. I think that needs to be looked at more, but I think some of the funding that's happened from a variety of different federal agencies have helped bring genetic services through regional programs that are organized within states to have geneticists actually go out and see patients, and there is some telehealth happening, but other things. That becomes very vulnerable when Medicaid programs and other state budget cuts happen, as are happening now. So I think that's going to be changing.

Lab tech and biotech strategy models are out there and interesting. We don't have enough to say about it.

We're struck with the interviews that we've done how genetic counselors play such an important role in providing services, how they're flexible and take on and create jobs, but the market challenges, particularly for reimbursement, are there. Some programs laid off genetic counselors eight years ago and have not hired them back yet.

Most services still relate to counseling, testing, and test interpretation. There are small numbers of genetic specialists. The people out in the field -- so far, very preliminary -- aren't saying that's a problem quite yet.

A mixed forecast from the people we've talked with about the timeline for therapeutics.

A restrictive market. We're seeing some interesting market responses. With procedures such as amniocentesis for prenatal diagnosis, money is to be made and there are freestanding diagnostic prenatal centers opening up.

It's my understanding that obstetrics and gynecology physicians tended to be one of the groups that went for genetics training. They compose about 10 percent of our sample now. What we're hearing is that maternal/fetal medicine or perinatology, the specialty of obstetrics that deals with high-risk pregnancies, has taken the genetics piece into their practice. So in many ways, it's a positive thing. It's been integrated into the subspecialty of obstetrics practice.

So those OBs are not getting genetics training now, which means some people would say there's less specialization and in-depth work, but the specialty has taken it and they have a procedure to do and they manage high risk. They have an important procedure to do that pays better. So there's a very dynamic happening now with OB and genetics.

We've not tended to find from the people that we've spoken to much of a big picture view or expectation so far. They're dealing with their issue, their practice, their institution.

Future research recommendations, kind of quick thinking to put a few things on the table.

At the policy level, the Institute of Medicine does studies when it is asked to do studies and when they're funded to do studies, and I think that they could be asked to help study the issue of genetic services broadly, along the lines of what they did with the primary care model. There are important questions that could be answered and would need in-depth study of the sort that they're able to do.

You've heard about the federal agencies, so I'll skip over that. Some are involved. Some are not.

Priority workforce-specific research, and I tend to shy away from specific research, but there is a void of information that I think we need to fill.

Some sort of assessment of supply and demand for clinical geneticists, M.D. geneticists, Ph.D. geneticists, and genetic counselors I think is needed.

As I think about the specialist professions in any area, we need specialists. This will be an important area for specialization and the market doesn't seem to be working right now. The number of

new applicants, physicians in training going into clinical genetics fellowships or residencies, is very small. The number that sat for the last board exam, which is only offered every three years for M.D.s, was 69. So that's 20 new M.D.s certified. Now, people don't have to sit for that, but that's a strikingly low number.

You have geneticists reaching retirement age. It's a little hard to assess the demand right now. We can't do it from the work that we've done, but my sense is that this is such a complex area -- like cardiology, intensive cardiology, tough, complex patients -- there's no doubt we need cardiologists. All internists, all family medicine physicians, do cardiology with what they do, but for tough cases for training the future professions, for many, many reasons we need a substantial number of cardiologists. The market has helped fill that because it's a very lucrative specialty, it's procedure-oriented, it's respected, hospitals like cardiologists and cardiac surgeons, whatever.

That has not been the case for geneticists, but this is complex stuff that I think we need a cadre of people going into and serving that specialist role. The same for genetic counselors and the same probably for nurses in genetics. I put that down as sort of a second level, but I think that's probably because we haven't studied it enough.

The fact that has been unrecognized that I've seen so far is that genetics is being taken up by medical specialists and medical subspecialists. Oncologists are the most notable example. That's part of their practice. When I was a medical oncologist, back then the black box was, 25 years ago, you have a strong family history of breast cancer. Wow, you have a really strong family history of breast cancer. We have to watch you carefully.

That was about all we could offer. Now, we're beginning to be able to offer a little bit more risk assessment and management thinking, at least, there.

The same with neurology. We're seeing talk about neurogeneticists within practices or within academic medical centers, people trained as neurologists who have an interest in looking at the genetics association. I think that area needs to be studied more.

Nursing, I think it's particularly that advanced practice or advanced training nurse who's a specialist in oncology. The Oncology Nursing Society has nurses who have a special interest in nursing and oncology, and that nursing group has 29,000 members. ISONG has about 300 members in the states. The ONS special interest group in genetics is 100 now, and we don't know about all the other subspecialties of nursing where this could be very important and has to be looked at.

I think we have to look a lot more. I talked with the people from the CDC yesterday about public health genetic services providers, genetic epidemiologists and others. There are therapists who help manage patients. Pharmacists will become more involved and are not yet greatly involved. So I think there's a lot of important workforce research to do, but I would frame it as a type of health services research, not solely workforce research.

These are my colleagues from universities across the country. Five of the six HRSA-funded Workforce Centers are very much involved with our project and HRSA just funded a very exciting project at the University of Texas Health Science Center in San Antonio to look at the access and providers of genetic services in the Texas-Mexico border area, which is an extremely important group.

Thank you very much.

DR. McCABE: Thank you very much, Dr. Cooksey.

Hunt, I'll let you take over for facilitator.

DR. WILLARD: Thank you to all three of you for framing what are not only critical issues, but ones that I suspect will generate a fair bit of interest and discussion around the table.

I'd like to start with two questions, which anyone can answer or not, that seem to me to be critical here. First of all, Joann, you argued or presented information that this has been on the agenda

for anywhere between 25 years and 10 years, depending on the lag there. So the question is is there any evidence in at least the last 10 years that the education level of the workforce regarding genetics has increased or not, and has that been measured and how will we measure it over the next 10 years? Because if we're going to be serious about this, then we'll have to have a way of measuring that and assessing that.

DR. BOUGHMAN: Measurement will be a very difficult issue, but I think, starting with some of the things that you've heard this morning and in the presentations this afternoon, you will also see advances that have been made in a variety of areas, and one of our big challenges I believe is differentiating between the adage "Make it and they will come" -- build the website and people will come and learn it -- versus actually taking it to the practitioner and getting them to incorporate it in their daily practice in a competent kind of way.

As Dr. Cooksey indicated, measuring in the practice situation is going to continue to be extremely difficult. It's a hard thing to do, although there are some ways, and we'll talk a little bit about that this afternoon on ways to try and get at that and not measure it very well, but in fact at least have a baseline.

DR. WILLARD: The other question, which may take more lively discussion, and I'll start with Dr. Cooksey, is in a perfect world, how do you define success? That's directed at you, but it's really something for the committee to evaluate.

So given the amount of resources being put into education, given the obvious need for education, given what you're measuring, in a perfect world, what's the model where we would say great, that's the model we're aiming towards, because until we have a model in which we know how we'd like it to be in 2010 or 2020 or whatever the logical endpoint is, it's very hard, it seems to me, to focus either educational resources or workforce planning if we don't know what the model is that we're aiming towards.

If I have a concern about the study that you've been funded to do, and I phrase it that way because I realize some things you're supposed to be doing and other things you're not supposed to be doing, it's that it's focusing at present on genetic specialists, and yet many of the data that you gave us, some people would conclude that the battle has already been lost. If there are only 1,000 clinical geneticists in a sea of 560,000 other specialists, each of whom, to one degree or another, feels that what they work on has a significant genetics component, it may be that studying who becomes a board-certified medical geneticist or a clinical geneticist and how they spend their time and where they're located is really just scraping a little bit of the ice shaves off the top of an iceberg when in fact we should be focusing our effort on what the iceberg is.

I'm always struck and have been repeatedly by the data that you didn't mention from the AAMC, which every year, as medical students graduate from medical school and they're asked what do you want to be when you grow up, and they all tick off a box on their chosen medical specialties, and medical genetics, over the five or six years I've tracked this number, is either 0 or 1 out of 17,000 medical students graduating every year. It's very clear.

This is at a time when we all go around, many of us, and say this is the most exciting specialty, this is a fantastic field, we quote Francis liberally, and the public is engaged and has bought into that, and yet somehow that never trickles down to the medical student who is trying to decide what he or she is going to do when they're out there in the real world.

So I'll throw that to you, and then there are hands popping up all over the place here. So I'll just have that as a point of some discussion.

DR. COOKSEY: Where to start? What I presented was only a portion of the work that we've been doing and thinking about. Let me take the last part of the question first, with the medical students.

The market really does work, and the market perception for becoming a geneticist has been modest at best. What our survey shows, and we're still analyzing it, is we asked the question would you advise a young person to go into genetics? And we actually got a little higher -- I can't remember. I think it was 75 percent said yes. We ask a lot of questions probing the esteem of the profession because we had heard anecdotally we're low paid, we don't see many patients, we're not recognized within our med school. We heard mixed pictures with geneticists.

That's not what we see in the survey. We see sort of a rather robust sense of self-esteem and a robust satisfaction except for reimbursement kinds of issues.

On the primary care situation, the AAMC, the Medical College group and many others, HRSA and others, have put strong focus that we need to build a strong primary care system to bring access to care to Americans, to help contain costs and improve quality and more holistic approaches.

What I heard last month at the International Medical Workforce Conference, which was very interesting and included a fairly large delegation of the U.S., including people from HRSA and others, in the U.S. there is real rethinking about the future of primary care providers in the country, that the concept that we have too few physicians, and particularly that we have too many physicians maybe, that was out there as a policy position and that we have too many specialists maybe, is being almost turned 180 degrees right now, that we don't have enough specialists perhaps.

Why? I don't know, but some of the contexts are the U.S. health care system, which is very different in some key ways than the Canadian or the U.K. health care system, is built on specialty care. Our reimbursement methods, the strengths of the profession, the professionalism that occurs in medical school and in residencies, is toward specialization, despite lots of efforts to get U.S. medical school graduates to go into primary care since the early '90s. The numbers went up and with the managed care and with the market looking like it was changing, with salaries for family physicians and general pediatricians going up and people going into particularly family practice, one marker profession went up and it's been going down.

It's also going down in Canada and in the U.K. despite efforts. In Canada and the U.K., they've recognized they're not producing enough homegrown medical students now and they are both increasing substantially the medical school capacity. In the U.S., the medical school capacity has been relatively stable, 15,000 or 16,000, except growth in osteopaths.

This is sort of more than you want to know, but why students aren't choosing genetics is a very important question, but I would say that they tend to be rather smart creatures. Physicians are rather smart and they respond to what they hear and incentives that are out there.

We have heard from training programs, from genetics programs. The loss of funding has been a significant issue, funding support.

DR. WILLARD: That's a hypothesis. The alternative hypothesis is they're really excited about genetics and they've decided that the way to do that is through cardiology or hematology or medical oncology, and the answer is not necessarily to go through medical genetics.

I'll throw that out, and I've got Debra and then Ed on the list.

DR. LEONARD: So this is kind of a follow-up to that. Thinking about genetics as a specialty area isn't consistent with the future model of genetic medicine in that every physician is going to have to practice genetics, and so are workforce studies looking at the amount of genetics being practiced by all the other specialty areas? And then it comes down to an education issue, not an issue of whether there are enough specialists.

One of the issues with having it be in everybody's practice is that genetics, because the public is not well-educated about genetics, it's very time consuming because you have to start back with

what's a gene and what's a mutation, not just explaining you have a mutation in such and such a gene that causes this disease.

A question. Are the current models and mechanisms for health care practice, with genetics being compartmentalized as a specialty area, and health care reimbursement issues, where you don't get paid for spending time talking to a patient, are they consistent with the anticipated future of genetic medicine?

DR. COOKSEY: Just to be clear, I fully believe that genetics will be integrated into our practice and understanding and thinking about health and medicine without question. When it will occur and how it will occur, it will occur in a jagged path probably. I have no disagreement with that.

I think when I've said that I think that it needs to be a strong specialty, is that every area that tends to be organ systems, we rely upon a cadre of specialists to help us practitioners with the more complex patients, with understanding the new and assisting with the diffusion out.

As far as, and I'm forgetting a little bit the nub of your question, is the education adequate or is the model adequate or are we studying the right things --

DR. LEONARD: Can the current health care system adapt to genetic medicine and what has to happen to make that happen?

DR. COOKSEY: We are looking. We're just starting this year to look and to talk to primary care physicians, internists, and family medicine physicians to see what they're doing now and to try to get some indication of things like what got you involved?

The data that's out there, and there is some data, says that primary care physicians become more involved with genetics when they are younger. They've had some course work in genetics. So they've been younger graduates, they're at academic medical centers, they have patients who ask them about it, and when there is something that they can do with the information.

Now, some of this research dates to the early '90s before BRCA testing and other testing was available, but again, I think that the primary care physicians are individuals that play an extremely important role and need to be educated, but they're targeted. Their look will be targeted. There have been editorials. The primary care physician can't do him or herself a full family history. They can do targeted. So they can construct pieces of the family history.

For instance, if the family history is a core part of genetic services, I think we have to look at how that's collected within the health care system and where that data is held and how you refine it so that you don't reinvent the wheel every single time. Certainly, genetic counselors and others are very well-trained at doing the complex three-generational family history. I don't think it's necessarily efficient to expect family medicine doctors, and they've sort of said we don't have the time and we can't do it. So we have to look at this as a systems problem that we need to address and I think that efficiency is extremely important in rethinking the systems of health care.

There are some interesting models out there. The Kaiser of Northern California, Ron Bachman has a model that has a sufficient number of genetic counselors for that population. I can't remember how many covered lives he had, but there are some ways you can look at some good models and sort of get the numbers that might be needed and see how they're used when you remove reimbursement constraints and whatever and when you have a defined population group and a care system that has responsibility and it takes responsibility for managing the patients.

So I think there are some models that are out there, but I think it requires new thinking, less focus on the silos of who does what, more recognition that the incentives will be for subspecialists and specialist physicians to take it up. They will have incentives to do that. For the primary care physicians, there are different incentives and different issues, and you've got to recognize the incentives and go with

those, whether those are professional or financial or whatever. That's what drives people.

DR. WILLARD: Thank you.

Ed?

DR. McCABE: Two things. One is that I firmly believe we've got to improve genetics education. Everyone has to be knowledgeable in genetics.

I'd also like to think there's going to be a role for the geneticist in the future, and the analogy is the radiologist. We all look at x-rays, but when we really want to know what's in that x-ray, we go to the radiologist, and I think that's the way we're going to dig in genetic information.

Having said that, though, I think there's a cultural problem in genetics. Medical students go to medical school because they want to make people feel better. Geneticists are diagnosticians. Neurology went through this a few years ago when they were losing all of the -- they couldn't do anything either, and they developed an area of interventional neurology, because otherwise that was going to be lost to the neurosurgeons and the others.

I think if we continue to focus on diagnostics in genetics, we are doomed to failure because it will be a very small part of the physician graduates who want to go out and never make anybody feel any better, and that's a problem. Outside of biochemical genetics, it was mentioned today, what we do is manage. We don't really make people feel better.

DR. WILLARD: Thank you.

I've have Reed, Francis, Barbara, Kimberly, Chris, and Brad in rapid order. Reed?

DR. TUCKSON: Rapidly, how would you assess, any of you, the adequacy of the genetic-based education in health professional schools today? Are you convinced that the leaders of medical schools and nursing schools have understood the inevitability that medicine is a genetic-based discipline now and that if you're going to learn heart disease, there is a genetic basis of heart disease? If you're going to learn cancer, there's a genetic basis of cancer. If you're going to learn liver disease -- there's a genetic basis of all these things. Has there been a revolution or are we still at the point where today's graduates, we will still be having to plan on continuing medical education catch-up, even for this class, or have they accepted this?

DR. BOUGHMAN: I will give you a little bit of data this afternoon, but the current students, at least in medical school, by the counts that we have available, are not only receiving in nearly every medical school the basics of genetics, they in fact are being tested on at least aspects of the basics of genetics, even in the national medical examinations by the national boards. That is a great improvement over five years ago.

The real trick, though, however, is not back to the definition of the gene or the basics in didactic lecture format. It is actually during the training and the practitioner aspects of this. How do I incorporate this? That immediately becomes both a curricular issue in the training program and a process that the professional guidelines that are out there that help determine what the services that a patient with Disease X should be able to receive in order to be cared for adequately.

DR. TUCKSON: Nursing schools?

DR. COOKSEY: I can comment on nursing. Nursing has studied this issue and their consensus is that general nursing education at the baccalaureate level or the associate degree level has had very limited inclusion of genetics in the curriculum, and they are trying to address that.

I think another issue that should be considered when we think about educational preparedness and competency for the workforce is to recognize that you have 2.5 million or so practicing nurses out there, and you need some level of awareness for all professionals within various areas.

But something else that has to be right up there as well is recognizing that within

professions there needs to be again this cadre of genetics specialists, I think both to help educate and to help move the profession along and set the vision and the pace. So whether it's pharmacists, nurses, other professionals, it's that higher level, whether it's master or at least trained, who are doing clinical practice that's in more sophisticated genetics to help do the training and education and keep the profession aware of the changes that are happening. So it's not just the body of everyone, like all med students. It's that specialty training.

DR. TUCKSON: I can't ask any more questions, but what I hear, though, and what I don't hear from these answers is I think I hear that it's coming. What I don't hear is we're on top of it.

DR. SHEKAR: Well, let me respond to that from our perspective that both the projects that I mentioned, the Genetics in Primary Care as well as the GIFT program, succeeded and are succeeding because of the commitment of the university administration over and above the faculty that are applying for those projects. So those are applied for by those universities, and the only way that those programs are getting those is obviously the quality of the applications, but also the support from people from within to provide that support to those projects. So it's very clear, Reed, that in order for genetics to move forward through those venues, there obviously needs to be support over and above the faculty investigative level.

DR. BOUGHMAN: The NCHPEG talk this afternoon will help answer that.

DR. WILLARD: I want to keep this going because my watch says we have 10 minutes.

Francis Collins?

DR. COLLINS: I'll try to be quick. I think, Hunt, you asked the right question at the beginning of this discussion about what is the model that we would like to see happen in the course of the next decade or so, as opposed to what path are we on right now, because I think the path we're on right now doesn't look very encouraging. The statistics quoted about medical students running away as fast as they can from the discipline of medical genetics are sobering indeed.

I think a major reason for that is the absence of effective role models in their vicinity. As medical students, we're all influenced by the people we see in our environment who sort of are the kinds of docs that we would like to become, and sadly, I think medical geneticists, most of them pediatricians, are seen as taking care of relatively rare conditions. They're seen, sadly, as practicing the genetics of yesterday, instead of the genetics of tomorrow. The genetics of tomorrow seems to have been grabbed up by other disciplines, BRCA1 and oncology as the most obvious example, but there are others. I think students are pretty clever to figure out sort of which way the wind is blowing.

So I think the model that you ask for, which most people I think would now endorse, is the sort of radiology example, where everybody who is a primary care provider, and that's not just M.D.s, need to acquire genetic skills, and when Joe McInerney talks about NCHPEG, I'm sure he'll go through some of the ways in which that organization is trying to push that agenda forward, but that there will have to be experts, specialists, who are in a position to sort out the complex circumstances that are going to happen very frequently.

I see two major problems with that model. One is reimbursement, and that would be a very appropriate topic for this committee to think about in terms of how are we going to take something which is time-intensive and procedure-limited and turn that into something that could be adequately reimbursed as an incentive both for the primary care provider and the expert to actually be compensated for what they do.

The other barrier, though, that I see is what appears to be at the moment a fairly strong level of resistance on the part of those who would become the experts to taking on that new role, to giving up the role of being the academic clinical geneticist taking care of the rare disease and becoming really, in a

much more different kind of model, the place to which a wide variety of complex conditions, most of them probably affecting adults, would get referred for sorting out. Also, as part of that, abandoning, sadly, the model where you can't see a patient in less than an hour and a half, which has to be abandoned if this whole thing is going to work.

So the question I have after my little speech here is to ask Judith, in your survey of medical geneticists, do you in any way get feedback from them about their receptivity to that kind of a very different kind of professional role in the future?

DR. COOKSEY: We did not probe those questions on the survey. I think the survey methodology would be very limited to that, but it could have been something -- I'd have to go back and look at the survey and see if we can get some hints. We certainly asked them attitudes, beliefs, their sense of the quality of care that's provided, the quality of genetics care that's provided. I'd have to think if we covered some of those others and I'd be happy to do it.

I think that one example to think about in this issue might be the infectious disease doctors and how they stepped up to the plate with HIV and how they're continuing to step up to the plate with emerging infections, with bioterrorism, with other things. You had a very relatively small specialty that, as another example to radiology, treats all ages, everybody can get infectious diseases, and they can be mild or they can be serious and life-threatening. They're treatable.

They stepped up to the plate and they helped train others, which geneticists are doing. There's a continuing need for infectious disease experts because of some challenges that we're facing, but leadership from that community, at a time that many physicians and other health professionals did not want to see HIV and AIDS patients because of personal risk or perceived personal risk, they came up. That was important and we've diffused the treatment of HIV out into fairly broad medical practice now with the specialty.

Francis, the question that I pose to you, I think it's a little premature to answer Hunt's question about what's the right model. We can think more about that. We're keenly aware that the current model isn't the best model. I happen to think there's value in studying it.

You haven't addressed the issue of the large group of specialist physicians who are taking on genetics and how geneticists really have an opportunity to work with that group because that's where things are going right now.

DR. WILLARD: Great. Thank you.

Barbara?

MS. HARRISON: One of my main concerns, as well as we've been talking a little bit about increasing the number of medical geneticists, but also increasing the number of genetic counselors, and I think the root of that problem is reimbursement, which we may talk about more later. I don't want to spend the time now.

But I one of the highlights when you were talking was this Bureau of Labor Statistics and how we are just not on there and how that's used by a lot of career counselors and guidance counselors, which, especially from the genetic counseling standpoint, are who we need to reach. I was just wondering if you had any ideas about how we can get on that list or kind of what the issue is there.

DR. COOKSEY: The Bureau of Labor Statistics is down the street a bit. The problem is that the data they collect, they fund every state, usually an Office of Employment Security, to do surveys of employers as to what categories of employees they employ by occupation, and then they fill in with Census data the self-employed.

We could certainly talk with them. I think the numbers are probably too low, and when they look at physicians and when they look at the 600,000 physicians or nearly 600,000, they look at

physicians, they look at pharmacists, they look at dentists. They don't look at the subspecialty level. So it's a very coarse measure that looks at every occupation in the country, including some small-level ones. You know, hair stylists, manufacturers of stainless steel. I mean, this is labor statistics information for our whole economy.

So we could, but I don't think that's quite the way to go to get on their radar. I just think that's not quite the -- it won't succeed, I think.

MS. HARRISON: Okay. Just real quick, would allied health professionals be on that list?

DR. COOKSEY: Specific allied health professions are. They have diagnostic professional groups and they have technicians and technologists and sort of more support level. Again, it needs to be a fairly large group, but there's about 30 different professional groups that they do collect data on, and I can share that and see if it looks like it makes sense.

DR. WILLARD: Thank you.

Kimberly?

MS. ZELLMER: I just had kind of a quick comment. I think that from the lay person's point of view, having been through amniocentesis and all that with my kids, it seems like there are certain areas that maybe there's a better job of getting genetic information out there and doing a much better job.

But I think in the case of rare diseases, I think that they are still way far behind, and I think that the primary care physicians, I think they're probably good to refer patients on to specialists, but I think even in rare diseases, the specialists aren't making the diagnosis of rare diseases. In our case, we went to three different pediatric neurologists at teaching hospitals before we got a diagnosis of a rare genetic disorder.

Then the other thing that I think is important for a primary care physician's education is not only knowing to refer patients and having that basic genetic information, but part of the problem that I know that we've had and also I know from other families with kids with genetic disorders is once you've been diagnosed with a genetic disorder, many primary care physicians don't know what to do with you.

We've been to three different pediatricians to find one who's comfortable giving us advice on whether or not our daughter should get immunizations or not or what types of medications it's okay for her to take, and the pediatricians like to defer to the pediatric neurologists, but the pediatric neurologists know nothing about more common medications or they know very little about immunizations. So it's hard to find someone who's going to make those basic decisions.

So I think you've got issues of not only finding people to recognize less common genetic disorders, but I think also teaching primary care physicians what their role is going to be once they're dealing with a patient who has a genetic disorder.

DR. McCABE: Thank you very much.

With that comment, I think we're going to wrap up. Thank you, Hunt, and thank you to our panelists. Dr. Boughman, we'll see you again after lunch.

We only have 45 minutes for lunch today. So that's going to be a challenge to everyone. We do need to be back here sharply at 1 o'clock, though, because that's when we have our public comment session. We have seven people in a 30-minute session this afternoon, so I would ask all of our public commentators to please try over lunch to think about how you can keep your comments to two to two and a half minutes so there is time for us to have discussion.

Thank you.

(Whereupon, at 12:15, the meeting was recessed for lunch, to reconvene at 1:00 p.m.)

AFTERNOON SESSION (1:00 p.m.)

DR. McCABE: We're going to begin our afternoon session. We have seven speakers, as I mentioned before the lunch break. I would encourage the speakers to please try and cut their presentations. If it stretches into a four- or five-minute presentation, there will be no time for questions, and I would encourage you to turn in your items in writing and permit the committee to have time to discuss them with you.

Our first speaker is Dr. Paul Billings, who is speaking for the Laboratory Corporation of America. Thank you.

DR. BILLINGS: Thank you, Chairman McCabe, distinguished committee members, friends, and guests.

I'm Paul Billings. I'm an internist, a medical geneticist, a professor of anthropology at the University of California at Berkeley, I'm the co-founder of GeneSage & Company, and for many years I've conducted research on the translation of biotechnology from lab to clinical care and in consumer issues in genomic medicine.

For instance, and this may be of interest to the Chair, given comments he expressed yesterday, I'm currently the PI on a grant funded by the Robert Wood Johnson Foundation that seeks to document and analyze legal and policy precedents affecting standards of care in genomic medicine. An initial database and draft analytic summaries are complete and I would be happy to give the committee, its members, and staff access to this project's initial results. The work will be reached at www.genewatch.org., a site sponsored by the Council for Responsible Genetics, and it is produced with the help of the National Council of State Legislatures and distinguished legal scholars.

I've also recently completed a large project considering the potential use in non-insurance health markets of genetic testing.

But I'm here today having recently become vice president and national director for genetics and genomics at Lab Corp. As you know, Lab Corp is one of the largest clinical reference labs in the country and is the provider of the highest-quality molecular and genetic tests and services nationwide. I'm pleased to be a part of such an excellent group of laboratorians.

I would like to make three brief comments, primarily in response to the discussions yesterday and the characteristic incisive comments by Reed Tuckson and Huntington Willard. It has been over 10 years since I testified before a subcommittee of the House of Representatives on genetic testing and the then-current ClIA-associated oversight practices.

First, the actors in genetic testing now -- consumers, patients, providers, payers, producers of technology, labs, standard setters, lawyers, and others -- are all generally united in the intent to provide safe, valid, and information-rich testing in the United States. While variance in quality of testing practices will always occur and a few enterprises may need to be curtailed, the system with its current mission standards and checks is working. I have seen no convincing data that documents current systematic harms or end user dissatisfaction with how genetic testing is now delivered.

Rather, demand for more products seems apparent, even with well-known public concerns about the uses of genetic information by third parties and privacy. Attention rightly ought to be paid to the needs of informed and, therefore, empowered consumers and patients who are changing the balance of power in demand and decisionmaking and who will be significant determiners of the market for tests and its qualities in the coming days. The role will likely be more important than some experts now feel comfortable with.

Second, clarification of nationally enforceable standards, oversight, and appropriate regulation is desirable. The issue has always been neither doing too much nor too little given this rapidly changing field, its evolving methods, and the wishes of components of the health care system. There are significant problems in the payment and coverage systems in our country that do not promote equitable access to the benefits of genomic medicine and sometimes contentious issues related to intellectual property ownership and policies currently affect which genetic and genomic tests are being delivered and their cost. As with regulation, these topics are relevant to your work as well.

Finally, I first took part in a debate about the special nature of genetic testing for policy purposes here in Washington many years ago as part of the Joint NIH/DOE Committee on Genetic Information and Insurance, which was chaired by Tom Murray. At that time, I argued that there are unique qualities of genetic tests and information, fully aware that this smacked of the so-called genetic exceptionalism and essentialism and that genetic data can be part of medical information generally.

Discriminatory practices using health-related, disability, or genetic information can be wrong and, if so, ought to be prevented. Pertaining to genetic information, the so-called laboratory of the states has legislated for years in this area and now developments at the federal level are occurring.

For the purposes of this committee's work, clarity about what, if anything, is special about genetic testing remains a challenge. As mentioned yesterday, the fact is that practices of those developing and delivering new imaging modalities are relevant and examination might cast light on non-unique aspects of the expansion in genetic services in this country. The need to collect data on the special aspects of genetic testing practices now and to let the information direct new policy and regulation is prudent conduct.

While new regulation and law can precede large-scale problems -- for instance, the banning of human reproductive cloning on primarily current safety grounds or the outlawing of certain discriminatory practices arise from genetic information simply because it violates basic American principles of fairness -- policy made anticipating problems or without enough data to direct it usually is fraught with folly.

Thank you for your time and attention. I would be happy to supply the committee with my comments in writing if you wish. Thank you.

DR. McCABE: Thank you, Dr. Billings, and we would appreciate it if you could supply your comments to us in writing. You can provide them to Sarah Carr. Thank you.

Our next speaker is Dr. Chin-To Fong from the University of Rochester School of Medicine and Dentistry.

DR. FONG: Well, I thank the committee for giving me the opportunity for a few comments to give some perspective. As a person that's spent a fair bit of time on what I call the front line of some of the teaching efforts, I spend about 50 percent of my efforts in medical education, and I'd just like to share a couple of points with you.

One is, to sort of put into perspective our programs, what I thought of is this hierarchy of interest. If you take this triangle as the pyramid of the general public, with sort of increasing degree of interest in genetics, this is probably, in Dr. Willard's analogy this morning, the iceberg, and somewhere in

the top half are folks who are highly more interested in genetics. This is the informed public that is interested in genetic issues, and somewhere in that upper group would be professionals that have to deal with genetic issues. At the topmost of this hierarchy are genetic professionals, who need to be interested in genetic issues. This is how we make a living in some ways.

So the amount of training needed to reach the top of the pyramid certainly increases the further you go, and what I really perceive as our real important agenda is to prevent the knowledge gap from the top and bottom of this pyramid to be widened as genetic technology becomes more and more advanced. In other words, if you don't watch it, this pyramid is going to get more and more elongated and we're going to fall further and further behind.

We take the current high school student as an example, as an approximation of the future public, and to think about high school education particularly. One of the issues that deal with high school education is that many of the existing programs that encourage genetic education turns out benefit kids who are already interested in genetics.

Secondly, professional development of the teachers is sorely needed, and thirdly, we need to really watch out to adhere to current state science curriculum standards, because this is what teachers have to deal with. Fourthly, we need to watch for availability of laboratory equipment to all school districts, which is easier said than done.

Fifthly, while we're interested in genetics, the current vogue in education in general at the school level is to develop student-centered education approaches that encourage critical thinking and problem-solving, and we think genetics teaching fits very well into that model.

So in Rochester, we have a new project called Project BEGIN. It takes advantage of an infrastructure in the New York State teachers. This is called the New York State Chemistry and Biology Mentor Network.

In deference to Dr. McCabe's call for brevity, I will skip through some of these specific items, but basically there is a hierarchy among the teachers in New York State that starts in the top with the highly motivated, so-called coordinating mentor teachers that would then evolve the curriculum development and teacher training. They work with the regional mentors and the regional mentors also work with local teachers. So this is the network of instructors within New York State that we capitalize on.

We take advantage of this network by bringing the mentors to the university and to summer workshops. We've developed a problem-based case that is very rich in ELSI issues that adheres to certain parts of the New York State science curriculum standard and that the coordinating mentors then take back to the regions to implement in the schools. So the mentors would disseminate these curriculum into the schools.

We also supply the participating schools with laboratory equipment to support some of these lab activities.

DR. McCABE: If you could wrap up in the next 30 seconds, please.

DR. FONG: I would refer you to the handout on the efforts we're doing with the medical education. So I'll probably quit here.

DR. McCABE: Thank you very much, and I would refer the committee to the handout. Thank you very much, Dr. Fong.

The next is from Ms. Dawn Allain, National Society of Genetic Counselors.

MS. ALLAIN: Good afternoon. Again, my name is Dawn Allain, and I'm the current president of the National Society of Genetic Counselors. It's my pleasure to speak on behalf of NSGC, which represents genetic counselors worldwide and is the leading voice, authority, and advocate for the genetic counseling profession.

Like others, NSGC recognizes we are entering an era where genetic services will increasingly have a significant impact on health care. Although genetic advances will benefit health care services, they will also pose challenges, particularly as they relate to access to genetic care. Specifically, many areas of the country have few or no geneticists or genetic counselors and many non-genetic health care professionals have minimal training in basic genetics. NSGC supports the expansion of not only the genetic counselor workforce, but also all genetic professionals. We also support increased training and education of health care providers.

I would like to highlight some key areas which NSGC believes could increase the genetic counselor workforce, as well as access to genetic services.

In regards to increasing the genetic counselor workforce, NSGC proposes three areas for consideration. First, we encourage SACGHS to support the renewed passage of and appropriations for the Allied Health Reinvestment Act. If this act is renewed and budgeted for and HRSA develops grants targeted specifically for genetic counselors, genetic counseling training programs could be eligible for grants promoting recruitment of minorities, development of didactic education and clinical internships, and programs for faculty development.

Second, NSGC recognizes that access to genetic counseling and testing services requires genetic specialists to practice in underserved patient populations and/or geographical areas. Inclusion of genetic providers into federal acts and programs would enable expansion of genetic services into these areas. For example, while federal programs, such as HRSA's National Health Service Corps, specifically aim to increase access to primary care physicians into underserved populations through scholarship and loan repayment programs, there are no programs specifically targeted for genetic providers.

NSGC would like SACGHS to support the development of federally-funded mechanisms for enhancing clinical genetic services in underserved populations. We also urge SACGHS to promote developing network infrastructures which will allow the provision of genetic services through telegenetics and web-based modalities to enhance access to genetic services in these areas.

Third, employability of an increased genetic counselor workforce and improved access to genetic health care is directly tied to reimbursement for genetic services. According to preliminary data obtained by NSGC, many medical centers subsidize genetic counseling services because there is no CPT code for genetic counseling services and there is no manner in which genetic counselors can bill.

The NSGC is committed to obtaining more comprehensive coverage for genetic counseling and testing services. We encourage SACGHS to make appropriate agencies aware of the current problems of billing and reimbursement and support state-based licensure for genetic counselors, the lack of which limits patients' access to genetic health care.

Separate from services provided by the clinical genetic workforce, it is clear that non-genetic health care providers are already providing some level of genetic services, including ordering and interpreting genetic tests.

NSGC strongly believes a well-informed and prepared health care workforce will lead to appropriate utilization of genetic services. We have and will continue to play an active role in educating health care professionals about genetic counseling and testing services through the development of professional guidelines and genetic education programs.

Based on our experience, NSGC believes that development of genetic curriculum or educational programs should be tailored to each medical specialty to ensure that each health care provider understands the immediacy and relevance to their patient populations. Education must also stress the underlying genetic component in common complex disorders, as well as single-gene disorders. In

education, educational programs must incorporate mechanisms for distance learning to help meet the needs of educating non-genetic health care providers. NSGC requests that SACGHS support the development and implementation of model educational programs.

In conclusion, genetic counselors currently provide a large percentage of direct patient care, genetic services, and education of health care professionals. Therefore, increasing the genetic counselor workforce will be critical to continuing translation and integration of genetics into routine health care. We encourage SACGHS, in conjunction with professional and consumer organizations, to continue to develop thoughtful yet practical strategies addressing the issues regarding oversight for genetic testing, education of health care professionals, and an increased workforce.

The NSGC membership has vast experience and expertise in direct patient care, laboratory services, research, public policy, and industry-based genetic services. We are available to work closely with SACGHS as you continue to address your important mandate.

DR. McCABE: Thank you very much, Ms. Allain, and while Dr. Lawrence O'Connor from the AMA comes forward for his presentation, I just want to make a comment that the NSGC has presented in public comment at every one of the meetings of the Secretary's Advisory Committee on Genetic Testing and now the Secretary's Advisory Committee on Genetics, Health, and Society. We appreciate your coming and commenting.

Dr. O'Connor.

DR. O'CONNOR: Thank you, Dr. McCabe.

My name is Larry O'Connor and I represent the American Medical Association. Now, the mission statement of AMA is to promote the science and art of medicine and the betterment of public health. With regard to genetics, we at the AMA are committed to providing educational tools that will enable physicians to better understand and incorporate medical genetics in their clinical practice.

This commitment is perhaps best illustrated in our genetics and molecular medicine website, which is shown here, the front page. This site is intended to serve as a portal by which physicians, as well as the general public, can obtain information on and links to the most current information in genetics.

Now, briefly, I'd just like to point out a few highlights that we have on our website here. For example, within our website we have a series of short educational primers that appear under the Number 1 heading there on various topics related to medical genetics, including gene therapy, stem cells, gene testing, and pharmacogenomics. These primers provide a brief summary of the subject matter and links to other sites where additional information can be obtained.

Now, we also have sections that are updated regularly listing recent advances in the field of medical genetics. These advances include not only research in clinical breakthroughs, but also news stories highlighting the social, economic, and legal implications of genetics.

In addition, there is always a special section within our website that provides more detailed analysis of current topics in genetics. As shown here, one such example is a topic on the recent anniversary of the double helix structure of DNA.

Now, the AMA also has a number of collaborations with a variety of national organizations to improve physician understanding of medical genetics. For example, we have recently co-sponsored a two-day seminar with the American Bar Association and the American Association for the Advancement of Science on applications of the human genome in clinical medicine.

Now, for physicians, we also have available a number of CME programs available online that can be used to better understand not only the science, but also ethical issues relating to the genetics, in this particular case shown here, of ovarian and breast cancer.

Now, the AMA continues to seek out new collaborations with federal, non-profit, and private organizations to develop new educational tools on genetic medicine. Currently, we are partnering with a number of organizations to help develop and disseminate educational programs in medical genetics.

Now, two examples shown here on our website -- actually, they're not on our website yet. They'll soon be posted on our website -- include a collaboration we have with the Dartmouth Medical School and the CDC to promote and distribute their educational program called "Genetics and Clinical Medicine." In addition, we're also working with the March of Dimes to promote their new education program called "Genetics and Your Clinical Practice."

Now, in the interest of time, I won't go into what each of these programs covers, but suffice it to say that each of these are intended for the primary care physician and are intended to provide tools and guidance to assist them in integrating genetics into their patient services.

Now, in conclusion, we at the AMA feel that our access to our physician members places us in a unique position to help increase awareness of how genetics can be integrated into clinical practice. We welcome the opportunity to collaborate with other organizations to create new educational tools or to assist in publicizing existing educational tools that may already be out there.

Thank you.

DR. McCABE: Thank you very much, Dr. O'Connor.

We'll now move on to Ms. Sharon Olsen from the Oncology Nursing Society.

MS. OLSEN: Thank you, Dr. McCabe.

Dr. McCabe and committee members, thank you very much for having us here. My name is Sharon Olsen. I'm an oncology nurse. I've been in oncology since 1969 and I've very proud of it. I'm here on behalf of the Oncology Nursing Society.

ONS is one of the largest professional organizations in the world. It's composed of over 30,000 members. These are registered nurses and other health care providers. All are dedicated to excellence in patient care, education, research, and administration in oncology nursing. We'd like to thank the advisory committee for this opportunity to provide testimony on workforce issues regarding genetic testing and counseling.

The Human Genome Project accomplishments have made it very clear that in the genomic era health professionals in every medical specialty will utilize genetic technologies and genomic information in the prevention, diagnosis, and treatment of disease. Oncology nurses are pivotal to the interface between genetic technologies and the patient. As such, in 1995, the Oncology Nursing Society put together a strategic plan that set as a priority the integration of genetics into its educational and practice initiatives.

You have in your packet our written testimony. There is also testimony available at the table outside. I will highlight here only a couple of our accomplishments and outcomes to date.

In 1996, a Cancer Genetics Special Interest Group was formed for nurses working in cancer genetics to provide a forum for networking, education, and professional growth so as to improve our genetic services provided to our patients. As a member of this group, we are spearheading efforts to help all oncology nurses recognize the relevance of genetics for their practice.

With the support of a five-year grant from the National Cancer Institute, the Oncology Nursing Society offers a three-day continuing education accredited course that is entitled "Genetics Short Course for Nurses." It provides basic genetics education for nurse clinicians.

Third, with the support of ELSI funds from the Human Genome Institute, a bench-to-bedside science lecture series has been offered at the Oncology Nursing Society Congress for the last three years. Typically, we have over 3,000 individuals attending this session each year.

Just this year, the Oncology Nursing Society released a practice and education-based genetics tool kit for oncology care for our members, and lastly, the Oncology Nursing Society has collaborated with our colleagues at the International Society of Nurses in Genetics and with the American Society of Clinical Oncologists on the publication of position papers and standards of practice.

While the Oncology Nursing Society has played and will continue to play a key role in educating and training oncology nurses in the provision of quality genetics care, ONS maintains that these issues cannot be addressed solely by private and non-profit sector entities. The federal government must contribute in these critical public efforts.

To that end, the Oncology Nursing Society advocates the following. Full support and sustained funding for the efforts of HRSA and the Human Genome Institute's ELSI program project that is entitled "Assessing Genetic Services and the Health Workforce." This is a project in which ONS will be actively participating.

Second, sufficient funding is needed to model interdisciplinary educational efforts that will facilitate the rapid and the safe transfer of genetic technologies and therapies to the service arena.

Third, federal support is needed for efforts to educate nurses and to encourage collaboration between the myriad of stakeholders in patient care for purposes of developing and advancing initiatives to integrate genetics and genomics into health care.

Lastly, provision of scholarships, loan forgiveness, and other incentives are needed to recruit and retain nurses practicing in the area of genetics.

As many of you probably know, we are expecting in the next 10 years a shortfall of 800,000 nurses in the United States. This is not just a U.S. problem. This is an international problem. The Oncology Nursing Society urges Congress to provide a minimum fiscal year '04 allocation of $163 million for the Nurse Reinvestment Act and other federal nursing workforce programs that are housed at HRSA.

The Oncology Nursing Society respectfully requests that the committee recommend to the administration as well as to Congress that they support a significant increase in funding for these programs in fiscal year '05, as educating and training nurses takes time and insufficient investment today will leave our nation without the care and the genetics workforce that it needs.

In conclusion, the Oncology Nursing Society very much appreciates this opportunity to discuss workforce issues pertaining to genetic testing and counseling. Please know that the Oncology Nursing Society maintains a strong commitment to working with this committee, and also the administration, members of Congress, other nursing and genetics-related societies, patient organizations, and other stakeholders to reduce and prevent suffering from cancer and to ensure that all people and their family members have access to quality genetic testing and the counseling that they need as well as deserve.

Thank you very much.

DR. McCABE: Thank you very much, Ms. Olsen.

Our next speaker is Mr. Michael Rackover from the American Academy of Physician Assistants.

MR. RACKOVER: Good afternoon. I represent the American Academy of Physician Assistants, and it is the only national organization that represents physician assistants in all specialties and all employment settings.

While PAs practice in at least 61 specialty fields, 44 percent of this year's respondents to our national survey reported that their primary specialty was in one of the primary care fields: family general medical practice, general internal medicine, OB/GYN, general pediatrics. The other prevailing areas that PAs practice in are surgery and their subspecialties, emergency medicine, and the subspecialties

of internal medicine.

As a university professor at a PA program, I make recommendations to the Association of Physician Assistant Programs, which is called APAP, established in 1972, and this is the national association representing physician assistant education programs in the United States.

The Accreditation Review Commission on the Education for Physician Assistants is the accrediting agency that protects the interests of the public and the PA profession by defining the standards for PA education and evaluating PA educational programs within the territorial United States. This organization is beginning the standards review process from this day until December 1st, 2003, and it is soliciting comments or suggested changes regarding the current standards.

I will try to make the appropriate recommendations, as my role is in physician assistant education and nationally is genetic literacy, to improve the education of PA students and postgraduate physician assistants in clinical practice with respect to the integration of genomic medicine. My concern is that we are having a difficulty as the students are coming out. There are no mentors to help them take what we are training to apply it to practice.

My other concern here is that when we talk about family history-taking and family practice, it should be synonymous. The correlation I present to you as a former x-ray technologist in diagnostic radiology, because of the shortage of radiologic technologists, you wouldn't expect a mammography to be done onsite. So it should be synonymous that in family history, the public assumes that the practitioners will understand their family history to be able to make applicable testing and to be able to take care of preventive medicine for themselves.

Thank you.

DR. McCABE: Thank you very much, Mr. Rackover.

Our final speaker from the public comment session is Dr. Fred Ledley from Mygenome.

DR. LEDLEY: Hi. Thank you for having me, and my apologies for not being here yesterday. I had the same choice most physicians have in practice, whether you pay attention to the 16-year-old who crashes into the goalie and doesn't think she can walk or whether you spend time talking about genetics, and I chose the same thing most health care providers choose.

I want to take the first 10 seconds and congratulate the committee and the people here on the work that was done to get the Senate to approve legislation on genetic privacy. Hopefully, this will go through the House and this will be a landmark, if it passes, in reducing one of the great barriers that has prevented consumers from having access to genetic testing.

My message to the committee is that this is only one barrier of many which prevent the health care system today from playing a proactive role in meeting consumer interest in predictive genetic tests, and I will restrict my comments today to predictive testing and not prenatal or genetic disease testing.

In addition to the inadequate training of professionals, there is the issue of the interest of the health care providers, many of whom are also contributing to meetings about the need for increased emphasis on social and behavioral causes of disease, increased recognition of spousal abuse, and of course, in pediatrics, where I trained, injuries and family issues.

The demographics of health care utilization is a critical issue, I believe, for this committee. Even if every physician was providing adequate genetic services, CDC data says that only 37 out of 100 people in our society go to a health care provider for preventive medicine in any given year. You can multiply it out. In your lifetime, that gives you about 15 hours of preventive care. There are not that many hours in that for in-depth genetic background and training.

This is particularly acute for young adults. For women, the number is a little bit

higher, but they're using their OB/GYNs for practice, and in the period of time in which predictive testing would be most useful and most important, few of us, few in the 15 to 44 demographic, go to health care providers. More of us, ironically, in our society are going to alternative providers for health and wellness care.

There has been very little incentive for payers or providers to build the infrastructure for genetics. I think there are a number of academic centers that have shown how outstanding that can be when that investment is made, but it's expensive. Forrester Research has identified fear of the cost of genetics as one of the things that's inhibiting growth of genetics.

Consumers have an enormous lack of trust in the health care system with genetic data. The genetic privacy legislation is useful, but I believe that consumer concerns for privacy are primarily emotive and not practical, and that family history is traditionally something that's out there. In our surveys of consumers, there is enormous fear of this in the hands -- they trust their provider. They don't trust the HMO, the PPO, the employer, and, very importantly for people at this table, the government is not a good guy in the eyes of many consumers when it comes to privacy.

Consumers believe that genetic history is very important. It's something they want to do, but the same disparities in health care that plague everything else in our system are even more profound for new technologies like genomics.

The cost, the reimbursement, education, literacy. We did a quick survey of all of the documents on the web related to cystic fibrosis and only one tests out at a readability level appropriate for consumers, and I congratulate anyone who is here from ACOG that they wrote the one that in fact passed.

Race is a big issue. Geography is a big issue. Trust of the local resources is a big issue.

This problem that we're discussing today -- I believe we already have inadequate resources -- potentially is only 1 percent of the demand for genetic services that we will have within a decade.

Let me give you some context. The Kaiser Family Foundation on their website lists all the public opinion polls that have been done over the last 10 or 15 years in health. In every one of them, more than half of the people asked say they would have genetic tests to know what diseases they'd have later in life, whether or not it's treatable, whether it's curable, whether it's fatal. No matter what adjectives you put in front of it, it's over 50 percent. Our data suggests that when you actually put a specific disease in front of people which runs in their family, acceptance would be in the 85 to 90 percent range, and this includes things like drug sensitivity.

Privacy concerns are so profound that in some studies we and others have done, people say they would not do genetic testing if it's reimbursed, only if it's self-pay. We see a 2 to 1 preference for self-pay if it's associated with the ability to decide and have control over where that information goes and who sees it. As I said, this is largely personal.

We find that consumers are smart. I had a professor in medical school who said, "When in doubt and you don't know what's going on, ask the patient," and what we see is that consumers have an awful lot to say. They want control. Autonomy is very important. Access is very important. Quality information, knowing the laboratory provides quality results.

Consumers want to provide informed consent. I think it would be a shame if this is something that's passed away as too complex. Even for simple tests, things in pharmacogenomics, I think consent is something consumers want to be asked for and want to provide.

Counseling, follow-up. The follow-up issue is an extremely important one, as Neil Holtzman and many others have pointed out.

DR. McCABE: Please finish up in the next 30 seconds.

DR. LEDLEY: So my charge to this committee is that we're looking at the tip of the iceberg here, that the solutions perhaps aren't even on the table yet, and that it's very important that the policies that are established do not, even accidentally, create any barriers. I think that there needs to be tremendous innovation and open-ended innovation, not only in clinical practice and training, but in information sciences and the application of expert systems, in business practices.

There needs to be partnerships with the market to bring investment to bear to support the kind of works that need to be done. One needs to look at the financial incentives and disincentives very carefully. There is some troubling language in the Senate law that could actually prohibit financial incentives for patients to have genetic testing, and we need to be very careful that no accidents are made in the future about things like this.

So we believe the consumer has a lot to say. I think they're going to be in the center of it. They're facing a health care system that today is not providing what really any of us here are saying are adequate services, and today's consumer is only about 1 percent of the people who we're going to be facing in the next decade.

I think the success of this committee is not going to be decided -- the question was asked before, how do you define success? I think success is not how do you prepare the health care system for genomics. It's in 10 years have we delivered the health benefits of genomics to the maximum number of consumers?

Thank you very much for the opportunity to talk.

DR. McCABE: Thank you, Dr. Ledley.

With that, that concludes the public comment. We appreciate all of the presenters from the public.

I would ask our three speakers to come forward now, and while they're doing so, first of all, I want to thank all of you for being responsive to our request at the first meeting. I know it required putting quite a bit of information together to prepare for today.

I'd also like to point out a discussion that we had at the first meeting of this committee where there was some concern about the designation of genetic counselors, and I want to bring everyone's attention to a pertinent letter received from Kerry Paige Nesseler, associate administrator for health professions at HRSA. You'll find a copy of this in your folders.

Based on questions that surfaced in our meeting in June, we sought clarification from Ms. Nesseler about the eligibility of genetic counselors and counselor training programs for HRSA program funds that support development of allied health professionals. Ms. Nesseler makes HRSA policy in this regard quite clear, and that is that genetic counselors are allied health professionals, schools that educate and train genetic counselors are eligible for Allied Health Program grants, the education and training of genetic counselors is a funding priority in the Allied Health Program, and HRSA encourages genetic counseling and education programs to apply for HRSA Allied Health Program funding.

As someone who's participated in the training of genetic counselors, I know that this is something that has not been understood, and I hope we can help get word out to the genetic counseling community that there is funding for the genetic counselors. So that's extremely important information, and I think serves one of our purposes, to improve communication between the agencies and the public.

Thank you very much.

Dr. Boughman.

DR. BOUGHMAN: Thank you, and I'd like to take this opportunity to thank the committee for letting us bring some of this information to you. You all have in your packets the written

materials. I'm going to hit some of the highlights and give you some framework this afternoon.

Actually, I put the slide up here with the American Society of Human Genetics logo on it to remind you that today I am not speaking for the American Society of Human Genetics in this context. I'm speaking as a member of the genetics community and having been involved in genetics education in many aspects for a long time and having chaired the previous work group for SACGT.

I want to remind you of the stakeholders and that Dr. Fong's very nice pyramid in fact could be superimposed on this. What I'm going to do actually, I'm going to speak about genetic specialist positions and just a moment about public education, but in fact allow the other speakers to focus on those areas.

What I would like to do is talk to you and remind you yesterday afternoon that one of the comments that was made late in the afternoon talked about the responsibility or whose job is it? Who is ultimately responsible for various aspects? Yesterday's focus was on oversight. Today, I'd like to put that framework around education, if you will.

In general education, of course, we have all of the school boards and all of the curricula.

Let's move on to undergraduate education, which can, of course, be extremely variable, but the American Society of Human Genetics Information and Education Committee has done one survey and is getting ready to do another survey on undergraduate courses around the country and their content. The bottom line there is that the content is not sufficient, especially on the basic concepts of genetics that can be applicable to real-life situations, and in fact there's been one paper published that you have the reference to and there will be another paper talking about suggestions for content there.

When we move in the undergraduate to medical school, in the premed requirements, we're now talking about the AAMC, the Association of American Medical Colleges. It is interesting that medical students who have been surveyed rank genetics as the third most important content area, yet no medical school in the country requires genetics as an undergraduate course for entry to medical school. However, there are some other interesting requirements still out there for us. This is a question that has been raised with the AAMC and they are looking at it, but change is sometimes difficult.

The medical school curriculum themselves, once again the AAMC and various other accrediting agencies who will look at the medical school curriculum. However, we have other groups who have been involved in the development of those, and we'll get to that in just a moment.

Postgraduate programs or residencies. Now we start really getting into alphabet soup. We have the ACGME, the graduate medical education accreditation organization, residency review committees for every specialty, and the American Board of Medical Specialties, which incorporates all of the major boards for all of the specialties and that serves as the umbrella for each one of those organizations.

For continuing medical education, we have the American Board of Medical Specialties, working through the individual boards, and in our case that would be the American College of Medical Genetics.

For geneticists themselves, both M.D. and Ph.D., their training, the content of their training, the accreditation of their programs, and their certification comes from the American Board of Medical Genetics. For counselors, that is the American Board of Genetic Counseling.

For health professions in general, I'm not going to go into that area. Joe McInerney will be covering some of these things, but from a slightly different angle in a little while.

Now, let's talk about the activities themselves. In the medical school curriculum, the American Association of Medical Colleges looks at those general curricula, but it is the Association of

Professors in Human and Medical Genetics, a subgroup of geneticists, that have worked very hard over the last few years to in fact develop curricular content for medical schools that now has been adopted by the AAMC and in fact is out there and being integrated into many of the medical schools around the country. Constantly under change as we move to a problem-based learning curriculum, we have to make a variety of changes, but at least the curricular elements are there.

Then when we get to national board examinations, we bring in another organization, the National Board of Medical Examiners. Now, as geneticists, we have to deal with another organization to get ourselves appointed to the right exam-writing committees to make sure that we test on genetics in order to affirm that genetics was important in the curriculum in the first place, because the dean of a medical school, wanting to know whether his or her medical school is meeting the guidelines, will look at the results of the national board exams.

So now, over the last few years, in fact we now have genetics as a subset. You can actually look for genetics as a group of questions. This has been a hard-fought battle, but we're getting there.

In residencies, once again, the ACGME, the accrediting body, and the residency review committees are the ones that work on the residency curriculum, and this is not only in the didactic curriculum, but in the practice curriculum itself, how in fact the topics are woven into the various residencies, and I'll make a few more comments about that a little bit later.

Continuing medical education, which has been, in theory for a very long time, a very important concept. Now, it's written in stone because via the American Board of Medical Specialties and the individual boards who certify individuals, certifications are now time-limited. So people in fact, to continue to be board-certified, have to demonstrate competency in a variety of areas.

This is a window of opportunity for genetics, as I see it, because in fact genetics is one of those areas that all of the practicing physicians out there who are garnering board certification in their own specialty, we can in fact provide them with the elements to teach and examine on genetics in a variety of specialties. This is referred to as maintenance of certification and is a very important item on the docket of the American Board of Medical Specialties.

We've talked about the number of individuals certified and so on, with clinical geneticists being 1,075. Please notice over the last few years the numbers are dropping in the number of board-certified clinical geneticists.

If we look around the room and count the number of board-certified clinical geneticists who have been in the room in the last two days, I would suggest that nearly 1 percent of those qualified have been here. Ergo, they are not seeing patients at this point in time. They are not teaching medical students. They are not teaching other places. It is this cadre of people that are responsible for doing all of these tasks.

Now that the American Board of Medical Genetics actually has a residency in genetics -- it's not just a fellowship on top of other residencies, and I'll talk about that option in just a second -- there are 175 slots available for medical genetic residents around the county.

That number that I have there of 95 is an error. It is only 78 out of the 175 slots that are filled. There were many more applicants than those 78. They simply do not have the funding to fund those residency slots because from the hospital's perspective, they get so many dollars for residency slots and in order for genetics, a new residency, to get a slot, another residency slot has to be taken away from some other specialty. So from the training point of view, the mechanism and the process is there. The funding is not.

You'll hear more about the counselors again in just a moment.

We're trying. We're trying very hard in the genetics community. We have been working very diligently with a broad range of specialties on RRC, with the residency review committees, on their curriculum, and in fact in the American Board of Medical Specialties board programs, through geneticists being members of these other boards, we now have developed approved genetics curricular elements in pediatrics residencies, in internal medicine residencies, in neurology, in family practice, and in OB/GYN. So now any resident going through any accredited residency program would have those elements in their training process.

In addition, we have worked on formal combined residency programs. Right now, there is a medical genetics and pediatrics combined residency program, a medical genetics and internal medicine combined residency program, and a medical genetics and pathology special training in molecular genetics that has been approved by both boards and approved by the American Board of Medical Specialties. We are now working with psychiatry and neurology, the American Board of Obstetrics and Gynecology for general OB/GYN, as well as a way to integrate the maternal/fetal medicine program in genetics, and the American Board of Family Practice has approached the American Board of Medical Genetics.

I talked to you already about maintenance of certification. I will remind you that it is board-specific. So of the 24 boards out there, the geneticist will have to work with the members of each one of those boards to include the materials, to make programs available to them, and then to assist them in figuring out ways to develop the competencies.

This is where one of the really important transitions comes in. I've talked about the American Board of Medical Genetics, but the American College of Medical Genetics is the group that not only helps develop many of these curricular materials, but in fact produces guidelines, practice guidelines, that puts the genetic example into the practice situation of the individual who needs to practice that.

For example, the American College of Medical Genetics and the American College of Obstetrics and Gynecology with the CF testing. It is when that material comes into the OB/GYN's office and they see how it is supposed to work for a patient, if there then is a knowledge gap that he or she has, they can figure out a way to fill that knowledge gap.

But unfortunately, the idea of build it and they will come or put it on the web and they will learn it is not going to work in many situations. I commend the AMA and I use the AMA website for a lot of different kinds of things. It is really extremely well-done, but we're talking about those doctors out there that only have 10 minutes per patient as it is, and it is a challenge to find time to utilize those.

The ABMS now has a committee on the management of maintenance of certification, the maintenance certification police, as some of us are fondly calling it. In fact, right now, from the genetics community, we have nominated one of the public members to be a member from the Genetic Alliance, who in fact I think will have a lot to say about maintenance of certification in all specialties in the way genetics fits into the pediatrician, the OB/GYN, the family practitioner.

We need teaching models. There are a lot of curricular guidelines and elements out there. I suggested the professors group before and you'll hear more about NCHPEG. Professional guidelines. The American Society of Human Genetics and the College continuously are developing guidelines. Programming for the specialties, as I suggested, and faculty development, geneticists with individuals in other groups like the Genetics in Primary Care model.

I talked to you already about the undergraduate and medical school requirements and it's heartening to realize that genetics is rated as the third most important subject, but one of the challenges once again we have in the medical schools is often genetics is taught as a basic science. When you in fact ask medical students if they want to specialize in biochemistry or physiology, they'll probably say no as well. They are not yet seeing -- in the time, genetics is not yet into the fabric of practice.

We also need to deal with patient and consumer education, obviously, both with informational materials at the time of service and many of the websites that are being developed now, and groups such as the Genetic Alliance, the GeneTests, and the National Library of Medicine. Their new Genetics Home Reference is a great website.

We're working with teacher training with the National Association of Biology Teachers and the National Science Teachers Association, giving lectures and workshops at every annual meeting that they have, providing materials to the teachers from the NHGRI website and the American Society of Human Genetics and NCHPEG.

We have a mentor network set up with over 700 genetics volunteers available around the country for any science teacher or student to get on the website and find a geneticist in their area who is willing to come and talk to a classroom, work on a project, and so on. We have more work to do there.

I left the media slide blank because at this point I don't think we've begun to tap the resources that the media might provide us, and I would suggest to you that that is an area that does need to be looked at.

In the written materials, I suggested ways of addressing each one of these. I'm not going to do that orally here. We can do this during the discussion, but I would suggest to you that we do have big barriers, and one is the perceived lack of necessity or relevance that the everyday practitioner out there -- once again, what do you want me to do as a physical therapist or a family practitioner? What am I supposed to do differently?

We need to shift our focus from rare disorders to the common ones. We have the challenge of, once again, the overcrowded curricula and the complexities of the probabilistic material that we're trying to get across.

We have a lot of gaps left to fill. We aren't teaching enough concepts in college and probably not in high school or junior high school yet. Right now, we're facing a declining specialist pool. We don't have enough training faculty out there in all of the disciplines who are excited enough about and know enough about genetics to train other faculty to train the students to provide the service.

There is a tremendous gap, of course, in understanding genetics and common disorders, and while my bottom line here is that we are improving -- I've tried to show you the way that we're working every day to increase the amount of genetics information that is out there at all these levels -- remember the curve of genetic knowledge that is coming out, and in fact one of the other speakers talked about this gap, and as the genetic knowledge goes up and we're working so hard to make this curve up, the gap still gets bigger.

So with that, I will sit down and we can continue the discussion a little later. Thank you.

DR. McCABE: Thank you very much.

Our next speaker is Joe McInerney from NCHPEG, the National Coalition for Health Professional Education in Genetics.

MR. McINERNEY: Thank you very much, Dr. McCabe, and thanks to the committee for paying so much attention to this important issue.

I'm going to do essentially what Dr. Boughman did, and that is just hit some of the highlights here. There is no way we can cover all of the information that we have available to us about the status of education. I'm certainly not going to talk about K-12 education, although I would be happy to respond to questions about that during the discussion session because I have spent a fair amount of time doing that. I will talk primarily about what we're doing with non-physician health care professionals, but let me just tell you a little bit about NCHPEG first.

I think many of you already are familiar with us. At this point, we have 127 member organizations and the real benefit of NCHPEG is that it's an extraordinarily eclectic collection. We have all of the professional societies in human genetics, many of the medical societies that are related to genetics directly, and even those that seem only tangentially involved, at least on the surface, and we also have many of the allied health professions, and we also have groups such as the National Association of Catholic Chaplains and the Association of Professional Chaplains, all of these organizations recognizing the importance of genetics for their constituents, but what that allows us to do when we decide to take on a project is bring the genetics expertise to bear in the context of the advice and counsel from the members of other professional societies.

Now, I use this slide a lot from Drs. Hayflick and Eiff because I think it does a nice job of encapsulating the gaps and the challenges, and I've highlighted in yellow here what I see as the significant challenges and gaps. This notion of specific content is an echo of what we've heard several times already. It is quite clear as I travel around, which I do a lot, and talk to a lot of different health professionals that what you've heard already is absolutely true. People want to know what do I do now? Don't tell me what the Genome Project is going to do for me 10 years from now. The technology is very cool. The science is cool. I admit that. It resonates in my heart of hearts as a biologist, perhaps, but what do I do differently tomorrow when I go into the clinic? So that is an absolutely important challenge.

But the broader-based educational challenge, and this perhaps is the difference between education and training, is to change the way people think in what is referred to here as the "'usual' cognitive strategies." I would assert to you that if in fact genetics is going to become the substrate for the practice of medicine and for the maintenance of health in the future, we do have to change the way people think. It's what people such as Barton Childs have called "health care through a genetic lens."

Now, perhaps the most significant thing we've done to date to try to address this gap is to provide some "Core Competencies in Genetics," and I think most of you are familiar with these already. They've been out now since January of 2001. We've distributed about 2,000 copies of them. We recently did an evaluation of the effectiveness of the core competencies, and I'll be happy to share some of that evaluation data with you.

For those of you who have not seen these, I brought about 50 copies. They're on one of the tables outside. You can pick them up if you wish.

Now, I won't assert that all the educational efforts that are underway now have been influenced by the NCHPEG core competencies, but certainly many of them have, and I just put this list up to give you some indication of the range of organizations that are involved in the development of educational programs in genetics. We've heard a number of these discussed already today and I won't go through examples of each of these because I will necessarily have to offend some organization or individual by leaving a program out.

But I will tell you that one of the things that comes through here -- and I don't know yet whether this is a good or bad thing -- is that there's no coordination. People are developing educational materials for specific health care professionals, but I will tell you that there's no coordination. There's no direction that says this is what ought to be included in terms of content or in terms of clinical objectives or in terms of a basic definition of genetic literacy, and I will tell you very straightforwardly that almost none of this, so far as I have seen, takes place in the context of a response to the question Dr. Willard asked before, and that is what do we want to be like 10 years from now? Those kinds of questions to me are not at the forefront of the development of these programs.

Again, I don't know whether it's a good thing or a bad thing that there is no coordination. I don't know whether centralization in this place is a good thing or whether decentralization

is a good thing. I do know that the decentralization is a good thing to the extent to which it allows the individual disciplines to specify the way genetics plays out for them and is relevant in their own practices, but whether we need some overarching mechanism that specifies what the basic content should be is another issue.

So I'm just going to give you some very quick examples of some of the programs that already have been developed, and I will apologize. Some of these did not copy well from the web.

This is a program for speech and language individuals, speech and language professionals, speech and language therapists. This is "Genetic Syndromes in Communication Disorders." This is a program developed by the American Occupational Therapy Association, one of NCHPEG's members.

This, of course, many of you have already seen from HRSA and a number of other organizations, "The Report of the Expert Panel on Genetics and Nursing: Implications for Education and Practice."

This is a special issue of Family Therapy magazine produced by the American Association for Marriage and Family Therapy, a NCHPEG organization. Again, an indication of how eclectic our membership is, marriage and family therapists clearly seeing the need for genetics education for their membership.

This didn't copy very well at all, but this is a publication from Mr. Rackover's group. Again, a special issue devoted to the importance of genetics for physician assistants.

Now, it is important to have commitment from the top, and this is a statement from the House of Delegates of the American Physical Therapy Association. I'll just leave it up for a second, but I will tell you that that's not enough, simply having commitment from the top. This is a nice statement, but so far as I've been able to tell, nothing much has happened yet in terms of influencing the curriculum and the training of physical therapists since this document was produced or this statement was produced.

Roughly the same thing is the case for radiologic technologists. These are from the current curriculum guidelines for the American Society of Radiological Technologists, and you see just two objectives here.

Now, again, it's not clear how these are likely to play out in the curriculum. I don't really know what these mean. Certainly, there's no national curriculum, so far as I'm aware of, for radiologic technologists, although they do have to pass a certifying exam, but I think these kinds of guidelines get played out in different ways in different institutions.

Perhaps the discipline that's further ahead than any of the others is nursing. You've heard a lot about nurses already, but just let me share with you some of what has been happening in nursing in terms of trying to bring some structure to genetics education and training of nursing professionals.

This began with this statement in 1998 by ISONG and the American Nurses Association, and we heard a little bit about this earlier today.

I thank Josh Carlson from the University of Washington's Public Health Genetics Program for providing these data. He did this as part of a master's thesis.

There are in fact a number of different genetics credentials within nursing. We heard about advance practice nurses this morning. There is also a genetics clinical nurse for those trained at the bachelor's level, and perhaps Dr. Feetham can correct me if I'm wrong, but I think this credential is actually provided by the Genetic Nursing Credentialing Commission, which is a subsidiary of ISONG. So you see, they are bringing some structure to the credentialing process here, which I think is missing in some of the other organizations.

There are actually certifications in a nursing specialty where there is a genetic component in certification or core competencies. There are 11 separate credentials here and these credentials are provided by the individual professional societies.

Of course, a lot of different graduate programs or certificate programs that emphasize genetics. Short courses, for example. We heard about the GIFT program already.

So nursing is really I think quite far along as compared to some of the other disciplines.

Social work is also coming along, and the National Association of Social Workers has developed a set of standards for integration of genetics into social work practice, and you can see the broad range of issues with which they are concerned.

Now, this is just a list of organizations that are asking for help or professions that are asking for help, and I won't touch on each of these.

I did just want to mention international groups. Some of you may have received this publication within the last few days from the U.K. It's called "Addressing Genetics and Delivering Health." I was happy to see NCHPEG cited in here. The folks who worked on this document came to the NCHPEG annual meeting last year, and I'm hopeful that we will be able to continue to collaborate with Dr. Burton and her colleagues on the development of educational programs for health professionals here and in the U.K.

There also will be a meeting in Santiago, Chile, in late November where we will begin to take a look at genetics education for health professionals in Latin America, focusing first on Chile and Costa Rica.

But again, we have requests from a fairly broad range of individuals and professions, and these are some of the things they request. Now, I should jump right down to the fourth bullet and tell you that in most cases, and this again reflects what Dr. Boughman was talking about, it is very unlikely that in most of these professions we're going to get a new course devoted to genetics. The task will be to supplement the curriculum in some way, to integrate genetics into the curriculum. Often, that requires that you have to leave something out and that's, of course, very difficult because each professor has his or her own favorite topic and you step on turf toes.

Some practical constraints. Of course, we've already heard some of these constraints alluded to before. Individuals are being trained at different levels. There are vagaries in state-by-state regulation of practice.

Scope of practice is a big issue. For example, when I talk to the radiation technologists, they were extremely interested in genetics, but a number of them said to me, well, it's hard for us to imagine how we can even integrate genetics into our interaction with patients because their work is very circumscribed by scope-of-practice regulations in their own states. They often don't even get to talk with the patient.

Now, I just wanted to finish up talking with you about some new NCHPEG programs, some things we are trying to do to fill some of these gaps.

Many of you likely already have received this CD-ROM we developed on psychiatric genetics. We distributed this free of charge to all members of the National Society of Genetic Counselors and about another 3,000 copies to other health professionals. That was funded by the ELSI program at the Department of Energy. We have a follow-up grant to that where we're focusing on genetics and common chronic disease, and the target audiences here are primary care providers and public health professionals.

Another program for which we are seeking funding at the moment is a program to try to train obstetric and neonatal nurses about the cystic fibrosis guidelines developed by the American College of Medical Genetics and ACOG.

Now, just a word about this program that we're doing on common chronic disease for public health professionals and primary care providers. This has been a real struggle to find case study vehicles that are relevant to both groups, and we have another meeting of the advisory committee coming up and then the writing team coming back in January to help us with this.

But we do believe, as it says here, that genetics does help us build bridges between population thinking, which is the purview of epidemiologists and public health people, and the primary care provider, and I borrowed this quote actually from Barton Childs at Johns Hopkins. "The epidemiologist asserts 'This is a risk factor,' and the geneticist says, 'For whom is it a risk factor?'" So we're trying to meld the two in this particular program.

A couple of other things. We actually have approached the notion of family history as the first genetic test and we think it is vital that all health care professionals be sensitive to the importance of family history in health care.

So under a five-year contract from HRSA and the Genome Institute and the Office of Rare Diseases, we are developing a number of programs, and this is one, a family history newsletter. We're doing three issues a year. They are appearing on the web, but we've also produced some hard copies. I brought about 50 copies of the summer issue. It's out on the table for those of you who want to take one home with you.

Now, I should tell you that the original charge to the Family History Working Group, which is chaired by Robin Bennett -- and I'll just digress to tell you that we really impose, is probably the right word, on the NCHPEG representatives to do a lot of work for us. They do an enormous amount of work as volunteers.

We're going to double your salary next year, Robin, actually.

(Laughter.)

MR. McINERNEY: We'll even triple it, if you like.

The original charge to us was to develop a generic family history tool, and for reasons that I won't go into now, that simply was not going to work. We didn't have the resources and there were a number of other issues, but what we did decide to do with the Family History Working Group was focus on the importance of family history itself and provide information about family history that we think health professionals can use.

Now, this is a project that actually began with Alan Guttmacher at the Genome Institute in 1999 and has moved over to NCHPEG, Genetics Resources on the Web, and we are hopeful that this will ameliorate some of the problems we heard about yesterday with respect to really egregiously erroneous information on the web.

This is a search engine that will search only the websites of the members of GROW. There are about 30 of them at the moment, including the American Society of Human Genetics, the Online Mendelian Inheritance in Man, GeneTests, the Genetic Alliance, and so on. Our intent here is to provide membership criteria that will serve as some certification that the information that you will access through this search engine is going to be reliable and accurate.

Certainly, the NCHPEG website is a resource that a lot of individuals use. We have lots of information on there, including a list of educational resources.

We also are undertaking a collection and review of genetics education programs. The reviews will be on the web. We've got an extensive list of materials submitted to us by a variety of individuals and agencies.

I should tell you that there's actually two pieces of this contract that I left out. One is our annual meeting, which will occur at the end of January in Bethesda, and the other is that each year

we're developing a web-based program for a particular discipline or group of related disciplines. Actually, this year, the focus is on dentistry, and we actually have done this in response to requests from dental faculty and dental hygienists to try to improve the genetic literacy of the dental community.

Now, Dr. Boughman mentioned some work that the American Society of Human Genetics has been doing in undergraduate genetics, and I just wanted to mention this very briefly, if only to say that I think we need to keep in mind that genetics education for health professionals and genetics education for the public need to be conceptually congruent. We can't be teaching the public one thing and teaching health professionals another. That is not to say that the public needs to know all of the details of genetics that health professionals need to know, but we need to be approaching it from the same conceptual base and the same conceptual assumptions.

What we did in this paper was take a look at the genetics content of introductory biology courses for non-majors. This is a huge course. Many students who have only one science requirement, for example -- they are non-science majors -- they take biology. That's it. That's the last formal science they're going to take.

So we analyzed the content of genetics in these introductory biology programs and we made some recommendations about basic concepts in genetics for undergraduate non-majors. We specified concepts in these six areas. I'd be happy to provide a reprint of that paper if any of you here wish to see it. The next task, which we'll undertake at the Los Angeles meeting in a couple of weeks, is to look at the content of human genetics courses, elective courses, again for non-majors at the undergraduate level.

Now, this is probably as close to flat out heresy or blasphemy as one can get in this room, but this is my modest proposal. I use the phrase "modest proposal" in the way that Jonathan Swift used it in his essay on the amelioration of poverty in early 18th century Ireland. He used it to be a little -- well, more than a little provocative, but also to make a point.

What I'm proposing here is that if we really believe, as we've heard about 100 times in this room over the last two days, that genetics is really the substrate for the future of medicine, then we should stop talking about genetic disease and genetic disorders, and we should talk instead about the role that genes play in the expression of disease, in the variable expression of disease, in the onset of disease in individuals at particular times.

Now, again, this is a bit of hyperbole, I guess, but I think we send mixed messages, and I think what is says is it continues to reinforce the notion that there is genetics over here and that genetics doesn't necessarily have anything to do with everything else that is over here. So I would like us to really be consistent about promoting our assumption about genetics as the underlying substrate for health care.

Thank you very much.

DR. McCABE: Thank you very much.

Our next speaker is Robin Bennett, who is past president of the National Society of Genetic Counselors.

MS. BENNETT: Thank you for the opportunity to present to you today. There is a formal copy of this report in your packet and also on the back table. So I'm just going to go through some of the highlights.

We were asked last June by the Secretary's Advisory Committee on Genetics, Health, and Society to give a report that develops a plan for gathering data on what is needed to increase the number, diversity, and quality of training of genetic counselors. This is a specific focus of the SACGHS under their general priorities to supply diversity, certification, and education and training of the health care workforce in genetics, and this discussion pertains to master's level-trained genetic counselors. Master's level-trained genetic counselors have extensive training in human genetics and counseling skills.

You also asked us to try to put some dollar amounts with this, and so I will be putting some of that in this report.

The methods that we used were we collected this information from July to October of 2003. We did a survey in September of the Association of Genetic Counseling Program Directors, and we also did an interview and met with the American Board of Genetic Counseling in September. We also had a consultation with Judy Cooksey, and you've heard about her 2000 workforce study and we reviewed that also.

We did try to look at the answer of how do you spell success in having enough genetic counselors, and I did try to do some investigation into programs in Canada and the U.K. to see if programs with a more uniform health care plan, if they had actually addressed this issue, but I couldn't find any evidence that that had been looked at in that way.

We also have a lot of information from the National Society of Genetic Counselors database. Our membership is 2,100 individuals all over the world, but I focused mainly on the training in the U.S., given the scope of this committee, and we believe that NSGC, 85 percent of practicing genetic counselors are members of this organization.

We also worked with our Industry Special Interest Group to see some ideas of where the technology was taking the field of genetic counseling. The NSGC, every two years we do a professional status survey. So that provided a lot of the information for this report, and then our Executive Office provided us information on our membership.

So this report is a brief review of the field of genetic counseling right now, with the focus on the U.S. population, the current status of the genetic counseling training programs, suggestions for expanding the existing programs while maintaining a high quality, and suggestions for developing new programs. We also were asked to focus on enhancing the diversity of the genetic counseling profession.

This report does not look at the workforce issues of other genetic specialists. You've heard a little bit about genetic nursing and the medical geneticists.

It's also not an independent report. It's through the National Society of Genetic Counselors. We did look into the cost of maybe having a report that would be independent and we estimated that if you wanted such a report it would probably cost anywhere between $90,000 and $150,000.

This represents the geographic distribution of the genetic counseling programs in the United States. There are currently 25 genetic counseling programs in the United States. There are three in Canada. Canadian citizens have priority in those programs, and so few genetic counselors are trained in Canadian programs, although genetic counselors trained in Canadian programs serve in the U.S. workforce.

There are 11 programs, represented by the green dots, that are in consideration, and the "RNPS" stands for "Recognized New Program Status." So they haven't been accredited by the American Board of Genetic Counseling yet, but they are anticipating that they will become full programs. There are two states, California and Utah, that have licensure for genetic counselors, and there are several that are pending that.

I think that there will always be some dearth of programs in certain parts of the country because these tend to be associated with academic medical centers. So for example, in Washington State, we are the only academic medical center for Washington, Alaska, Montana, Idaho, and Wyoming. So unless some of those kinds of things change, I think there are challenges to having programs everywhere.

The American Board of Genetic Counseling is the official accrediting body of the genetic counselors. As I mentioned, there are 25 programs. You can see that, unlike in some of the other

genetic specialties, there has been a steady increase in the number of genetic counselors. Because of this and because of the licensure of genetic counselors that is pending, the American Board of Genetic Counseling will now be offering the cycle every two years, instead of every three years, and hopefully that will continue to see the same advance.

The accreditation of programs, the genetic counseling training involves 27 areas of competency within four critical domains. The critical domains are communication, critical thinking, interpersonal counseling and psychosocial assessment, and professional ethics and values. There is considerable didactic course work involving human and medical genetics, cytogenetics, developmental biology, embryology, teratology, statistics, quantitative and qualitative research, counseling theory, interviewing techniques, communication skills, ethics, and public policy.

The time-intensive part of this training is the field work of over 800 hours per student. There is also a commitment to having teaching experience in all the students and as part of the accreditation process, and also all the programs have a research component. The average program length is about two academic years.

It's hard to predict the demand for genetic counseling. It's hard to predict what people are going to want when tests become available. For example, in Huntington's disease, when people were polled how many people wanted to be tested, up to 79 percent of people intended to be tested, but worldwide only 9 to 20 percent of people have chosen to be tested. So it's hard to say when people are asked what would you do if this theoretically existed and then predict their behavior when that becomes available.

As a profession, genetic counselors have been very able to meet the demands of the field. For example, in 1994, there were only 10 percent of the profession practicing in cancer genetics, and in 2002 over 42 percent of the profession is practicing in cancer genetics. I think that the broad base of training in genetics makes this group particularly adaptable to changes in the market.

We also have adapted to increasing our patient load without necessarily getting more resources to do so. So between 2000 and 2002, there has been a 66 percent increase in the number of patients seen. So you can see, as compared to what was given for the medical geneticists, about 572 is the average annual patient volume for a genetic counselor.

I can say in my own practice at the University of Washington, in 1996 we saw maybe 26 people for cancer genetic counseling. This year, we'll see over 400. So there is a big demand for services that's increasing.

I think that the demand for genetic counseling, there is always going to be specialties that want to refer to a genetic counselor. So some people are going to become competent. Some professionals and others are going to want to make that referral.

It's also very much tied up with billing and reimbursement, and the best example I have is from a letter I actually got from a surgeon that he sent to his insurance company that was for this patient.

It says, "To whom it may concern: I am responding to your letter refusing genetic consultation on" this patient "and suggesting that we do pedigrees, et cetera, on her. You are forgetting who you are writing to. I am a surgeon. Remember, we are not cognitive professionals. I don't know how to do pedigrees. The only people that do pedigrees are genetic counselors and dog breeders. Since she is not a dog, I thought the geneticist would be the best fit, and thus my request."

It continues, "Perhaps you will still refuse our small and humble request. Then, in keeping with 'Frank and Earnest,' we will send her to you for genetic counseling," and you can see the comic: "You want a second opinion? Call your insurance company."

The demand for genetic counseling may not be that you see a genetic counselor office in every block, like you might see an attorney or a real estate agent, but I think that the number of jobs that are listed on the National Society of Genetic Counselors job postings shows that over 75 percent of genetic counselors find a job within one month of graduation, and in Myriad Genetics in 1996 -- this is a major cancer genetic counseling service -- they had one genetic counselor in 1996 and they have 33 employed now. There has been a steady rise in the number of jobs. The last column, it's not a full year yet, so there were about 143 jobs posted so far this year.

Genetic counselors are not only clinicians, but they serve as a resource for the community. For example, 71 percent of genetic counselors serve on an advisory board of a consumer support group. They also are very involved in organizing conferences and workshops, particularly for health professionals, and also for consumers.

Genetic counselors are also a major workforce in terms of teaching. You can see that 77 percent were involved in teaching medical students and 34 percent taught other health professionals. They also are involved in many of the nursing curriculums, with over 70 percent teaching nurses, and there has been a specific focus on physician assistants and social workers, probably because of some of the things you heard in some of the earlier talks about their initiatives.

In the current genetic counseling programs, most of them are in academic medical centers. There are six that are at private schools. The rest are public. The average size is 16, with the range being six to eight to 46 students for the total enrollment over the two years. Estimates are that 550 students applied for genetic counseling programs last year. The majority of these are women and also Caucasian. These people are very qualified applicants with high GPAs and GREs.

So when we get down to the limits of the genetic counseling training programs, the biggest limit that overall was expressed was quality field placements for that 800 hours of field placement, and if a program loses a field placement, they have to often reduce the number of people enrolled in their program.

There is also an enormous volunteer effort that goes into genetic counseling training. There are over 400 hours per year for the average supervisor if they do four rotations per student per year. The rotations are usually six to eight weeks or so, and they also do a lot of teaching that is uncompensated. So the lack of funding for programs and limited scholarship opportunities are the biggest gaps that were reported.

The annual cost to train a genetic counseling student, again, this is just based on talking to the program directors. It wasn't going to their actual centers, their programs, but they range from $25,000 to $50,000, with the average being $30,000. The higher range tended to be the private institutions over the public institutions. There are a lot of physical resources and in-kind contributions that are also given that aren't accounted for in this number.

So here's what we are proposing. We have sort of a two-pronged approach to increase the number of genetic counselors in the workforce and the capacity of training programs. That was to look at the existing training programs and funding of new programs.

So the biggest thing that the existing programs need is additional training sites, and there also are very limited student stipends for getting people to -- if we were going to expand the number of sites, they would have to be often out of town, and so if there was a student stipend of maybe $3,000 per year to cover housing and travel expenses to get to these off-site rotations, that would be a big help.

Also, the supervisors, as I've already said, are giving a lot of volunteer time, and so if you're thinking about 100 hours of rotation per supervisor, if you were looking at a $25 an hour base pay, which is based on the average salary from the professional status survey and wouldn't account for people

with a lot of experience in genetic counseling, some of the more experienced supervisors, that would be about $2,600 per student or, if you were looking at it in FTE mode, that would be $10,400.

Also, there is a need for additional faculty. There is currently limited funding for clinical and research supervision, and this limits the number of students that can be admitted. We feel that adding additional faculty would also benefit educating other health professionals because you could have joint appointments in physical therapy, social work, nursing, whatever.

So an FTE for a genetic counselor would be $80,000 to $100,000, including benefits, and for a medical director/geneticist it would be $150,000 to $175,000.

To have some sort of training grants for students, similar to doctoral candidates in many other types of allied health professionals, would also help the existing programs, and you could consider tying this support to limited obligation to practice in an underserved area.

We felt that a diversity scholarship was an acute need of the profession, and some sort of a student stipend to cover books at about $5,000 per student would be another approach to enhance these programs.

If you were going to increase new programs, we suggested three to four years of start-up funds, and you might require the institution to have a matching fund. So this is the estimated cost for a new program. I talked about a genetic counselor program director of $100,000. An M.D.-Ph.D. geneticist, $175,000. Program administrator, supervisor stipends. If you were going to count the average size of a program as being 16, so there would be probably 32 placement rotations, that may be a few individuals, and then continuing education and resources, and then the diversity scholarships that I mentioned for the existing programs.

One way to increase the existing programs and to enhance new programs would be to do more teaching through webcasting. So you could have Dr. McCabe teach all of the genetic counseling students in the country at one time if you were going to do webcasting. This would help eliminate faculty time and expense.

It would also perhaps enhance student communication with journal club and case conferences, so that programs that are very small may have that benefit of conversing with other students in other programs. The cost of that, I didn't have a way of estimating that, but many medical institutions do have this kind of equipment and the charges are really for the land lines and the technical support.

The benefits of targeting funding for genetic counseling training are many, and I think if you look on Appendix A at the end of your packet, we ask the programs if we had funding -- we didn't say how much -- how big could you grow your program? And they all were willing to expand with the exception of maybe one or two programs. Most programs could expand at least 50 percent and some could actually double in size if you could deal with the supervision sites, the training site issues.

We already have at least five programs that have talked to the American Board of Genetic Counseling that are waiting to exist and I had 11 on that actual map. So there are certainly plenty of new programs that are just waiting to exist.

Having more training for genetic counseling would increase the diversity of the field if we had funding to get students who couldn't afford to come to the genetic counseling programs as they're offered now. We would get more diversity, and having more diversity gives more role models for clients to see genetic counselors besides white Caucasian women. There would also be increased access to genetic counselors by other health professionals for teaching.

So I think the genetic counseling workforce is an enthusiastic workforce willing to do clinical work, teaching, public policy, work with genetic industry, and I think that even though there may be a limited number at this time, I think it's a good bang for the buck in terms of the money.

I just wanted to end by saying that the NSGC is available as a resource for future workforce assessments or to provide expertise in developing initiatives to support genetic counseling training programs and increasing the genetic counseling workforce, and the American Board of Genetic Counseling and the Association of Genetic Counseling Program Directors also expressed their dedication to providing data for such initiatives.

Thank you for your time.

DR. McCABE: Thank you very much.

Dr. Tuckson, I'll now turn it over to you as facilitator of the roundtable.

DR. TUCKSON: Thank you.

Well, we've got quite a robust amount of information. I guess as the moderator, I'll just ask -- I'll shock everybody. I'll just ask one question and then actually let some other people get in it.

I think, Joann, I really enjoyed your presentation earlier, where you sort of laid out a variety of roles that need to be played.

By the way, I want to keep in my mind always this constant theme we keep coming back to over and over again, which is what's different about this? What's new? What's special? How come this ain't just medicine and where is the delta between why this isn't just medicine or will be soon to this special little nugget, and what is that nugget and how do you define it?

But if you take the roles of family history and risk assessment and genetic counseling and information gathering and then there are diagnostic issues and risk assessment and then management and care coordination and supportive services -- I mean, there is this variety of things -- I guess I'm still struggling with how do we then decide who does what? How do you know who has the qualifications to do what? And therefore, as so much of the thesis of this, part of this, is that people keep saying they want the reimbursement to follow, you've got to have some idea then what is this service that people want reimbursed? Who's eligible for doing it? How are they accountable? So that when money is paid to people for doing things, you actually have some idea of what you're buying and some idea that the person that's doing it is qualified.

So I guess it's a large question all in one, but at the end of the day it's how do we decide who does what? What's the certification that folks ought to be able to have? Who makes those decisions? And then maybe we can start figuring out something about what it is that people get paid to do.

DR. BOUGHMAN: There was a question in there.

(Laughter.)

DR. TUCKSON: Yes. How do we approach that? I mean, that's what I guess it boils down to is at the end of the day, how do we make progress in making these determinations? I guess I'll make the question easier. Are we getting there on answering these questions?

DR. BOUGHMAN: I think by necessity we are getting there, just as in any other field services or tests or diagnostic procedures, whatever it is, are in fact coded and reimbursed for the procedure. In the CPT codes, it doesn't say "as done by" and then say an oncologist had to order or perform this test or interpret a test. The code itself for the activity or the service doesn't include that process. So I think there are ways that we will get there by actions alone.

The question about who is competent, really, to do these things I think will depend on the definition of the service provided. The interpretation of a single carrier CF test is different, I believe, than doing the risk assessment and counseling an entire family. The coding process is going to have to be done in such a way that there would be differentiations, and I know that there is much discussion going into these kinds of things and separating out these issues in the level of service.

DR. TUCKSON: One last little quick question. Robin, maybe you can help me then.

Actually, that helps me a lot. Thank you, Joann.

The genetic counseling, then, at some level, there's a service called genetic counseling. Not everybody today in terms of reimbursement will pay for a genetic counselor unless it's ordered by a physician. If the counseling is ordered by a physician, then there is a genetic counselor who can then provide that service.

Should genetic counselors be able to provide this service of genetic counseling without a physician's order? Should they be able to generate their own demand today?

And secondly, should anybody else out there be able to do genetic counseling other than one of the people who graduated from one of these 25 accredited programs that therefore have some official standing as a genetic counselor?

MS. BENNETT: I think those are complicated questions because the billing and reimbursement issue is something that the National Society of Genetic Counselors is trying to address right now to figure out some of those issues about are we getting reimbursed now.

In general, the licensure bills so far have said that genetic counselors will work under the supervision of a physician. That doesn't mean that they can't order the testing, but that there is different oversight in terms of working with a physician.

I think that right now, given the complexity of the genetic testing, that it's good to have that relationship working with someone else. I think genetic counselors in general work as a team, although some are in private practice. Most of them are working with a physician, maybe in primary care or oncology, and recommending genetic testing that they're authorizing and then the genetic counselor is interpreting it, or they may be working with a medical geneticist.

DR. BOUGHMAN: Let me give you another example. Several years ago, there was a real challenge about molecular genetic testing, and the perception of the geneticists that there was an underlying amount of understanding of genetics that would be required to perform and interpret these tests correctly.

Pathologists, on the other hand, had the perception we've been doing complex tests for years and years. We can adapt and incorporate. We were head to head. In fact, the American Board of Pathology and the American Board of Medical Genetics were able to work out a one-year training process that a pathologist can go through and have certain specialized training and be equally certified to perform that level of complexity of tests.

Let me just throw out another example. We've heard that in nursing there are certain specialty certifications that are done, for example, in the oncological situation, oncological genetics, that in fact could or can be, if we so choose, integrated in such a way that there would be circumstances where everybody can agree if we work hard enough together.

DR. TUCKSON: Excellent answers.

Debra, and then Hunt.

DR. LEONARD: In fact, I'm one of those pathologists that's now certified by ABMG and ABP in molecular genetic pathology.

A question. Can you clarify, Joann? You said that there are now medical genetics residency programs.

DR. BOUGHMAN: Yes.

DR. LEONARD: But there are 175 slots and only 78 are filled, and you made a statement that you can't fund the residency slots. I'm co-director of our residency program, so I understand. We have 35 slots and we can't expand that, come hell or high water. It's mandated by somebody and I don't know who.

So who would allow the medical genetics residency programs that are now new programs to create new slots at those locations for that residency program and can we do something to facilitate that process?

DR. BOUGHMAN: Actually, Dr. McCabe may be able to help us here, in his role as a chair, but some combination of the hospital and the medical school itself in designing residency programs, but because the money comes back to the hospital, I believe that is the place where the number of residency positions overall, the cumulative number across all specialties, is determined.

So in your 35 slots, if at your hospital a new program came up, you would hate to give up one of your slots to medical genetics.

DR. LEONARD: So the number of slots per institution, per hospital, is set by CMS? Who sets that and can that be asked to be expanded for medical genetics, new medical genetics programs?

DR. McCABE: It's set by Medicare funding. So I assume therefore it's set by CMS. It's amount of dollars and that is linked to the number of residents there are, but it's also a complex formula that involves inpatient and outpatient experiences as well. Then you have to recognize we're not one of these, but as a pediatrician, I have to point out the freestanding children's hospitals until very recently did not receive the Medicare funding, and it's under threat again is my understanding. So the way we pay for trainees is quite arcane and it's a serious issue.

The other thing is that with these combined programs, my understanding is that it only pays for the first training, so that if you're taking three years to do pediatrics, but now you can do pediatrics and medical genetics in five years, the last two years are on the back of one of the departments in the medical school, not paid for by the hospital.

DR. LEONARD: So is there a way to change that?

DR. McCABE: My guess is that there are a lot of constituencies that want to change this arcane formula, and that this would be a relatively small constituency. Sure, one person can change the world. I don't know if this is a world that one person could change.

DR. WILLARD: Just to clarify a little bit, my understanding, at least as of a few years ago when I was playing that role, is that it's the dollars that are set, not the number of slots. So any hospital can add more slots. It just has to self-fund them. So you can add five more residents for anything the hospital decides is important to it, but it's going to have to find its own way of financing it, and some hospitals do that.

DR. LEONARD: Right, and in fact we have many more than 35 residents that we fund.

DR. WILLARD: Right, and some hospitals and programs do that and others, of course, don't, depending on their priorities and financial situations.

DR. LEONARD: Right.

DR. TUCKSON: Debra, do you have another question?

DR. LEONARD: No.

DR. TUCKSON: Hunt?

DR. WILLARD: Thank you.

Two questions for Robin. One, I wanted clarification on your assessment of cost because you referred to genetic counselors volunteering to teach and volunteering to train genetic counseling students. Are not most genetic counselors salaried at their institution or are they paid on a per visit basis like a barber?

MS. BENNETT: Many of the genetic counseling programs will have maybe one or two funded positions for training genetic counselors or they'll have a program director, maybe some

medical geneticist's salary, but they bring in many other outside speakers who are genetic counselors, and I'm aware of very few -- any funding for the supervisors when they go to the clinical rotations.

So people are being paid to be a genetic counselor, but they're supervising students, which is taking more time. They're supervising their theses projects --

DR. WILLARD: But it's not hurting their take-home pay. It is simply a question, within that institution, to juggle whether enough patients are being seen and whether the division chief or the chairman of the clinical department or whatever it is feels he or she is getting enough bang for the buck in terms of the outlay that's going to pay three genetic counselors and one supervisor.

MS. BENNETT: Well, I think that it limits the number of placements. People aren't willing to take a student on because they're bearing the expense of educating that student and taking time to review cases with them.

DR. WILLARD: Right. Fair enough, but for this committee, it would focus, and least my thinking is, we're trying to identify what's a federal role here. What are the issues Secretary Thompson's going to care about? I would view that issue, as a former clinical chair of a clinical department, I would focus that attention on enlightening clinical chairs and to what degree they prioritize not only genetic counseling as a service, but also genetic counseling training as a training exercise for the development of new professionals.

So I see it less as a federal issue as I do an academic leadership and hospital leadership issue. But that's just my comment. I'm just one person.

The other question I wanted to get clarification on from the standpoint of the NSGC, and it was Reed's question but I didn't hear the answer, which was, again, what is the model? What's the vision here? That all genetic counseling should be done by a professional genetic counselor? And if the answer is yes there, then that's going to focus us on numbers of training programs and the workforce projections, et cetera.

Or is it going to be similar to what we discussed before lunch, where very complex issues, there needs to be professional genetic counselors, uppercase GC, to do that, but that all kinds of people, if we educate all kinds of health professionals well, all kinds of people can do lowercase GC?

Those to me are two very different models with two different long-term outcomes.

MS. BENNETT: I think that the National Society of Genetic Counselors feels that there's certain competency, that if you're going to call yourself a genetic counselor, that you should have. Some of that may be a credentialing process, but in general, if you're going to provide the counseling component with the genetics component that is necessary for many complicated tests, then that should be done by a genetic counselor or a medical geneticist.

DR. WILLARD: As a quick follow-up, that's going to be a long uphill battle, it strikes me, because just as we saw before lunch for all things genetic, I would think that there are all kinds of health care professionals who think that at some level they can do lowercase genetic counseling. You may want to give it another name, but that they can provide simple estimates of risk and interpretation of certain kinds of tests.

MS. BENNETT: I agree with you. I'm not disagreeing with that at all.

DR. TUCKSON: CDC, Tim. Timothy Baker, for the record.

DR. BAKER: Yes, as we were talking about health professionals -- you know, our favorite subject here -- looking at the continuum of health professionals as we train health care providers all the way through the various disciplines of public health, I want to remind the committee and the audience here of the variety of disciplines that need to continue to be engaged in the public health world, and we at CDC have been trying to marshall this notion of fitting genomic knowledge that comes from all sources --

and it's been mentioned, family history is certainly genomic knowledge, and much of the discussion we're having here -- in answering existing questions that we know there's variability and risk of fundamental epidemiology. They don't need a new science. This is helping answer existing questions within that science.

Then the notion of health education. How do you explain to families that they have differences in disease risk in common diseases? Well, we're all expecting that this is going to help answer some of those questions.

So I just I guess would bring to the attention of the discussants here that as we're looking at answering one of the questions I think that Dr. McCabe brought up earlier -- he was commenting on one of the reasons perhaps students don't go into medical genetics is because of this notion that you're pointing out a problem, rather than something that may be helpful or promising -- to some extent, the reason we named our office back in '96 the Office of Genomics and Disease Prevention was this notion of trying to tease out that knowledge and recognize when it fits into answering those questions about risk factors of disease, so we can target interventions effectively and bring it to the health of the public.

So at some juncture, we need to move more carefully into that, along with the discussions you're having here.

DR. TUCKSON: Terrific. Good comment. Thank you.

Emily?

DR. WINN-DEEN: So I was struck when I read through the book there was a plea from the person who wants to start a genetic counseling program in the State of New Jersey for funding. You know, how can this committee help get funding?

I have to say, my immediate reaction is why isn't the State of New Jersey stepping up to the plate? I mean, the argument was the state needed counselors. Most people when they're trained tend to stay geographically near where they're trained for their jobs. Why are the state medical colleges not stepping up to the plate and creating training programs for genetic counselors and providing the diversity scholarships and all the other things that are needed to have the counseling community reflect the diversity of the community they would be serving?

DR. BOUGHMAN: Let me jump in and put a former hat of mine on as provost at one of those state universities, and we actually did get a genetic counseling program started.

It's simple economics. You have a situation where state medical schools, in many situations, are actually funded primarily from NIH research grants and clinical income, and the state portion of what goes into those budgets is relatively minimal. In the School of Medicine at the University of Maryland, only 12 percent of the School of Medicine budget was state dollars. Everything else was everywhere else.

Now, the individual states, you know that the budgetary constraints that we're having -- we've most recently heard about California -- bring down the size of the state and the number of millions or billions of dollars may be less, but the state budgets are going down, and with the NIH budget, unfortunately after years of going up, now being flat, the state medical schools especially are getting crunched from two directions. So the initiation of new programs while you are trying to maintain programs you have is an incredibly big challenge.

DR. WINN-DEEN: So having also lived in Maryland and watched the really good job that they did with creating undergraduate programs to graduate people with skills in biotechnology to meet the local employment needs of the biotech sector in Maryland -- I mean, if a state has a need for these professionals, I really think the state should step up to the plate and take care of their needs. You know, this was a half a million dollar start-up grant. This was not something that was billions and billions of

dollars.

DR. TUCKSON: Thank you.

Ed, we'll come right to you in one second, but Robinsue was right before you. You're from the Office for Civil Rights.

DR. FROHBOESE: Right. Robinsue Frohboese with the Office for Civil Rights at HHS. I wanted to pick up a little bit on Emily's theme of diversity, but take a little bit of a different angle, and that is during the past two days we've heard this thread throughout of lack of racial and ethnic diversity across the board in terms of the population for research and testing, as well as when we get to the provider side in terms of clinical geneticists and lack of diversity.

Robin has touched a little bit upon the issue of providing scholarships, and one question that I had wanted to ask in the morning, and I don't know if Dr. Cooksey is still here, but perhaps -- oh, great. Good, because one thing that you talked about that I think might address this issue, because we've been hearing this identified as a challenge but really haven't heard about solutions, and you had mentioned a promising grant at the University of Texas that HRSA is funding that will deal with border populations between Texas and Mexico. I wonder, just from the brief description that you gave, if this was a project that is going to get at some of the racial/ethnic diversity issues in terms of service population, research population, testing population.

DR. McCABE: We can have a brief comment.

I was in Texas for a while. The Rio Grande Valley is 85 percent Latino and underserved, critically underserved, and I know the services were being provided by the state with geneticists flying in, but there is a tremendous need in that community for services that will do more than just fly in geneticists, but will actually bring services to bear and provide role models for the young people in those communities.

MR. McINERNEY: If I may -- I'm sorry, Judy -- you might want to look at a program at the University of Cincinnati. Cindy Prowse, a nurse there, is running a program to educate nurses from minority and underserved communities and populations in genetics. I think that might be a nice model for this group to take a look at.

I think it's important to think not just about training genetics professionals from minority and underserved populations, but to think about how to reach underserved populations, whether you're reaching them with individuals who are themselves minorities or not. I think that's important.

DR. TUCKSON: Judith, did you want to make a quick one?

DR. COOKSEY: I believe it's a short, funded supplemental program that I think is looking at and trying to clarify some of the access issues and who is getting genetic services and who is providing it. There are many other efforts to try to improve diversity, but I don't think that one is specifically focused on improving diversity. I think they'd like to do that, but their focus is different than that.

DR. TUCKSON: Thanks for answering the question.

Ed?

DR. McCABE: Yes, I just wanted to remind everybody, as we were assembling the group, that HRSA does consider training of genetics counselors as training of allied health professionals, and does have funds for those training programs, and so I think it's very important that we recognize that and that that be a source of funding for new programs as well as existing programs.

Having been a part of the program at the University of Colorado, these are incredibly personnel-intense training programs, and that's part of the problem to a health center administration.

DR. TUCKSON: Ed wants us to break off, but Debra, you want to get one last

comment quickly?

DR. LEONARD: Could somebody clarify, either Dr. Sullivan or Robin Bennett, is there a CPT4 code so you can bill for genetic counseling and what is the Medicare reimbursement for that? And then what gets reimbursed in other settings, you said you were looking into.

DR. SULLIVAN: I left my CPT code book back in Baltimore, so I don't know exactly the answer to that, but I can find that out. I am a certified professional coder. Also a CPCH.

DR. TUCKSON: But there is a code. If a physician orders it, there is a mechanism by which physicians' offices can tell payers what happened, that something happened called genetic counseling. I just don't know what the reimbursement is.

DR. SULLIVAN: The answer is 28V, 28 Victor. And how much does that get you?

PARTICIPANT: It depends. It's time and hourly.

DR. TUCKSON: Time and hourly-dependent.

All right. Well, thank you. Let me just thank Joann, first of all, for your coordinating of both of these sessions today. We really owe you a debt of gratitude.

I think that Hunt raised an important focus that Ed picked up on, which is starting to try to drill to what can the Secretary do, and Ed sort of gave a very specific example of that in terms of HRSA and those resources.

I'm not sure how the rest of the committee feels, and we've got some time, I guess, where we're going to talk about what the next steps are, about whether or not we feel that, having been given this excellent foundation, now the question is what do we do with it?

I know, for one, that you've whetted my appetite for wanting to know a lot more about if we were to project five years from now, what is the work of the workforce for genetics? How does this get integrated into what a cardiologist, an oncologist, and so forth -- what do they do in five years? What will their work be? Therefore, what support will they require to be able to exercise their clinical practice?

Some of that support it sounds like from time to time may be a counselor. Some of that support may be an Internet site that provides a very real, functional, moment-by-moment set of supplementary information that a physician may need. I mean, there are a variety of supports that are going to be required.

I think one other thing I'm taking away from this is that I am left with some concern about how well the academic establishment in each of our disciplines are retooling the academic enterprise quickly to be able to meet some of these challenges. It sounds like a lot is going on. Everybody's kind of happy that things are moving. Whether or not those activities are adequate to meet the needs of the nation in five years, certainly it seems to me a big question mark, and I don't know what to do with that anxiety.

Thank you.

DR. McCABE: Thank you very much, Reed, for this session, and thank you to the speakers.

While you're stepping away from the table, Alyssa Johnson is from the National Council of State Legislatures, and so perhaps she can just briefly give us some insight into the state issues.

DR. FEETHAM: Before we move to that, I would just like to say that Dr. Joan Weiss from the Bureau of Health Professions is here, and from the Division of Allied Health, and those of you at a later point may want to approach her and talk about availability of TA, technical assistance, from Allied Health in regards to funding, resources, and applications, et cetera.

DR. McCABE: And Joan Weiss is sitting over here, so Robin, you can't see her when she stands up now, but perhaps you two can huddle at the end of this session.

So, Alyssa Johnson?

MS. JOHNSON: Thank you, Dr. McCabe.

I'm happy to offer any insight that I might have and answer any questions, but actually I have a question for Joann. I do work in the health care program at NCSL, and not education, so as a follow-up to the discussion on funding a genetic counseling program at the University of Maryland, I'm wondering, when those dollars were appropriated, if you used state funds, are they specifically given for, for example, a genetic counseling program or is that up to the university itself?

DR. BOUGHMAN: It was out of university funds, the state dollars that came to the university, and then allocation from below that level.

MS. JOHNSON: Okay. So it's not a legislature decision. Thank you.

DR. McCABE: Thank you.

Well, we have some time now set aside for the committee members to discuss next steps, and I'll basically break this down into two tasks that we have to deal with over the next 20 to 30 minutes, because we've already begun to see an attrition of the committee and I know from previous experience we will see more over the course of the next hour as people head for airplanes.

The two tasks that we have to do is talk about this report that we had agreed that we would send to the Secretary, and then the second task is the prioritization of items for the committee's agenda, and I want to make it clear that's the committee's agenda, not the next meeting's agenda, but it's setting our agenda of our effort in the near future.

So first, I know that Sarah and her staff have done some work on what would be the key issues. While they're getting that up, there are some things that I don't know that they're in the items that I saw before, but some points from the meeting in June that we had identified for further discussion in the future.

Large population studies and the resources needed to advance genotype/phenotype correlations, and we had asked that the NIH present some details. Because of timing, they actually asked to hold off on that because it wasn't the right time, but eventually we will have that report and be able to use that to generate advice to the Secretary.

Genetic discrimination. We said we should explore with the EEOC and consumer and professional groups the extent of genetic discrimination cases, with the Department of Labor to provide audit data on genetic preexisting condition violations by group health plans.

This is all just to remind us of what we have talked about before.

Health care-related issues. Integration, insurance coverage and reimbursement, affordability, and disparities in access, and we had talked about departmental efforts to address health disparities to ensure that genetics is included and receiving proper consideration in these initiatives, and AHRQ to organize a presentation on the diffusion of innovation and its implications for access to genetic technologies, as well as effects of patents and licensing practice on access to clinical genetic technologies.

So as we proceed with our discussion, I just wanted to remind us that we have some other items that we said we would discuss.

Sarah, you want to comment?

MS. CARR: What we've done is just tried to summarize what we heard you all discuss yesterday afternoon, and I think you indicated that you want to put a small task force of the members and ex officio agencies together. The members that I think volunteered so far are Reed and Debra.

DR. McCABE: And Hunt.

MS. CARR: And Hunt, okay. Well, this is why the questions are there. We'll add Hunt, and then anybody else, too. Then the agencies were CMS, CDC, and if this were to be a broad-focused report, HRSA. Well, I haven't even said what this task force is going to do yet. I'll get to that. So

HRSA is a maybe at this point, and then there are some questions about other agencies. We can talk about that.

But I think -- well, at least what we heard was that you want to prepare a draft report or we should prepare a draft report for consideration at the March meeting that would be based on the minutes of the first meeting, the presentations and deliberations of this second meeting, and, as necessary, additional fact finding and analysis, and that we would prepare a report to the Secretary describing the current status and future promise of genetic technologies to benefit health and society, and specifically articulating the steps that need to be taken by government, the private/profit and non-profit sectors, the public, and society at large in order to realize the full promise and potential of the Human Genome Project.

This is very much, I think, taken from the way Emily described why she got into public policy, but how you described that, Emily, seemed to capture a large part of the discussion.

The committee hasn't really talked about it in these terms, but the report could articulate goals that need to be achieved and pursued at a broad level, and then some of the issues and concerns that we've talked about. There are things here that we can talk about, but they're really kind of place holders and they reflect some of the things that you've been discussing these past two meetings and some of the things that you've heard from presenters. We can go over them in more detail if we want to take the time now or we want to defer that to the task force, but then these would be the specific steps that are needed and some of them would take the form of recommendations to the Secretary and so forth.

So if you want, I'll walk through this a little more. We could do that or maybe, at the face of it, you need to say whether you think we've kind of at least gotten close to what you were after.

DR. McCABE: Cindy?

MS. BERRY: I think of it in actually a broader sense first in terms of our task, and I don't know if this helps folks or not, but I've kind of outlined, and I always have a tendency to do this at the end of these meetings, what are the problem areas based on what we've heard in the last few days and also in previous meetings.

I've identified some, and I'm sure I've left some off, and I can go through those, but then the big question is which of these require further action and recommendation from our committee and which just require further monitoring? I mean, I think we have to really understand -- rather than sort of develop the tasks, I'd like to go backwards and do the concepts. I saw them, many of them, identified up there as you were scrolling through there. I can just run through them really quickly.

Workforce, obviously. Student training is one issue. Then the other issue is getting to the practitioners who are already out there in the field. Integrating the genetics knowledge base into clinical practice. Who should do it? How? Also, there are diversity issues under that.

Testing. Junk science was talked about. Unregulated labs and tests, advertising and the Internet, those kinds of things.

I'll give you all this, Sarah, because I'm just sort of running through it in the interest of time.

Access was another issue that has been talked about. What are the barriers to access? Well, coverage, health insurance coverage and reimbursement, and maybe it's worth hearing from folks from the insurance industry. What do they do typically in terms of covering genetic testing and screening and genetic counseling services? What are the thresholds for allowing coverage? What are the concerns that they have? So we could also hear from federal health programs. Then health disparities also falls under access.

The research void. We talked about that a little bit, the pharmacogenomics. Is enough research being done and is it appropriately targeted? Then there was a diversity issue under that as well,

that so much research is being done on certain groups, but not on certain minority populations.

Public awareness. We touched on it a little bit, which is is the public equipped to understand these services that are out there and how it can benefit them and do they know the risks and benefits?

Confidentiality, non-discrimination. We talked about that. The HIPAA and privacy regs. Are they sufficient? There's the genetic non-discrimination legislation. Beyond those two things, do we need to think about developing a set of principles or has some other organization already done that in terms of genetic services?

Then there are ethical concerns, which we really didn't delve into too deeply, but I listed as something that is worth talking about.

So I have all these so-called issue areas or problem areas that we touched on, and from there I think we should determine which of those require action on our part, where we can put forward something constructive to the Secretary, and which just require us to pay attention to.

DR. McCABE: And I think it's interesting because the ex officio agencies also gave us a list of what their issues were, and we're beginning to hear some themes. Number 1, with eight of the agencies stating that this was important, use and misuse of genetic information in insurance, employment, education, and law. Number 2, with six citing this one, ethical, legal, and social implications associated with the use of genetic technologies to screen or select for desirable or undesirable traits.

I won't go through the rest. After that, there's a very large group of five that come up, but I would think that this group that will be looking at both the report and the prioritization of issues should also look at what the ex officios have recommended.

Cindy, I think you've done such a nice job of organizing that that I hope you will contribute to this group as well.

MS. CARR: Ed, could I say one thing? Cindy, it seems like what you've done is -- there were sort of two tasks that we were going to talk about. One was to put a group together to identify the priorities of the committee, the next things that the committee needs to work on, but the committee also indicated an interest in issuing a report rather quickly to the Secretary that would in part be based on the minutes of the first meeting, the substance of the first meeting, and we could bring in the substance of this second meeting, and tell the Secretary where things are with genetic technologies now, where they're going in the future, what you think the promise is, and where you think some of the pitfalls are, and you've laid them out there.

Then you could consider in that report telling the Secretary, perhaps through the initial discussions of the task force, identifying what the committee thinks needs to be worked on in a more intensive way, and then we could include that in this first report, so that the Secretary knows where the committee thinks are the issues that further study is needed on, and then he'll know also in a very concrete way where you're headed as a committee.

Do you see what I mean? So we could kind of combine everything into one report that would also set the stage for your future efforts.

MS. BERRY: I was just questioning whether a report is premature, given the fact that we haven't stepped back and figured out the answers to some of these questions, but I have no objection to it if people think at least a preliminary report summarizing what we've heard -- you know, that's fine, and if the Secretary would find that useful, that would be a good first step. But I was sort of thinking that we needed to do this other exercise first before we provide him a report.

MS. CARR: Well, I think you could do it either way for sure. I mean, there's value in either approach.

DR. McCABE: We won't send out the report before the next meeting. So a large part of the agenda of the next meeting will in fact be looking at this report and then I'm sure, knowing the folks sitting around the table, that there will be some reordering of the points and prioritization, and I really see that exercise as both serving as an update to the Secretary on what we've accomplished and also helping us to set our agenda for the next one to three meetings.

DR. WINN-DEEN: Yes. I think we ask our President to give us a State of the Union. We ask our governors to give us a State of the State. I think basically what I'm hearing is let's give Tommy Thompson sort of a baseline. We've had all these reports and here's where the U.S. is today, maybe some comments on what other countries are doing and where we rank -- you know, where they're ahead, where we're behind, where we're ahead and they're behind -- and then what specific things do we feel we should address.

I think that would be probably very helpful to him to give him that top-level overview to carry forward in his job and also to focus our efforts on the gaps. Where are the gaps and where are the things where we can do something to make a difference?

DR. McCABE: Reed?

DR. TUCKSON: I guess I'm still struggling on this one. I don't feel that we know enough yet to be able to say anything to him that's definitive enough for our action. I see that we have walked down a road here and opened up some important doors and laid out some important questions.

I think, Number 1, again, and I know it's been the overwhelming, dominant theme that keeps coming up, is I think we all want to know more about this idea of genetic exceptionalism and integration into clinical medicine. I think that we really need a much firmer foundation or at least a shared vision among us as a committee about what we think that means, and I don't think that we have a shared vision about that and I don't think we know how to interpret then the thorny issues that are before us until we get that.

So I would just say I think that we need to have the next meeting and that we're going to do some work before the next meeting to have that shaped with a finer point before us.

Number 2, we spent a long time yesterday on the issue of regulation and oversight. I think we made a conclusion that we don't want to get back into that too deeply, but we raised some real issues around the pharmacogenomics issues and how does pharma work, the relationship between the public sector and the private sector, and we had pretty specific questions that came out of yesterday's conversation that I thought I was hearing that we wanted to get a little bit deeper in and then be able to then report back with a definitive conclusion, but I think it meant that we were to do a little work outside of the next meeting to bring that forward.

Then Number 3, we have opened up this big can of worms around the workforce issue, and we were given a great foundation today, but I think we all have questions.

So it seems to me that we've got some fine-tuning outside of the committee meeting to have an even more intense conversation at the next meeting. So I see three clear work products that aren't brought to closure enough -- I don't think, but I'm looking for guidance from others -- to be able to say to the Secretary, hey, here are three things. Now, they ain't everything, but we've got three things on our plate right now and we want to get to some others coming up, but we've got some more work to do.

DR. McCABE: Dr. Sullivan?

DR. SULLIVAN: Yes, thank you.

I tend to agree with Reed on this issue. I don't know the Secretary well and I doubt whether he knows me, but I do think he wants something that's more finalized rather than here's what we're doing. He wants to know, well, what's the answer?

I'm a student of business history and one of the books I read last year was "Big Deal" by Bruce Wasserstein, who is an investment banker in New York City who has done very well and he's worked with industries, whether they be health care, food industry, manufacturing, aviation, and he defined five factors which are essential to the success of any enterprise, be it a project like this -- we could call ourselves Genetics, Inc. -- or General Electric.

You have to master these five factors. I've heard them here. I could put them all together in a little package for you. You have to be able to master the regulation, the technology -- I've heard regulation and technology -- vision, strategic vision, leadership, and finances, and I've heard finances mentioned a lot.

So somehow or another, that has, over many years of investment banking and multiple deals on Wall Street, those five factors have to be addressed in order to come to a successful conclusion in any business or industry, and I consider this a little business venture we have.

So I believe defining our strategic vision, coming up with answers about the questions we mentioned regarding finance, and certainly at least addressing regulation -- we can't get too deeply into that or we'll be here forever -- and also putting our arms around the technology are all factors that we have to come to some closure with at some level before we put this forward to the Secretary.

DR. McCABE: Thank you.

I guess what I was thinking was that this exercise in the interval with the task force -- maybe it's more than one task force -- would begin to accomplish that strategic vision, that that would really begin to set where we were going because I think we don't know exactly where we're going and we've got to establish that vision. So that's really what I saw it doing. I think those are all important aspects that would need to be incorporated.

If we look at Reed and the three things that you talked about, the concept of the shared vision, which really has to do with the prioritization and what are the issues, what are the big bang issues, and what are the gaps, pharmacogenomics, and then workforce issues were the three that Sarah captured here in her notes, the question is whether we'd have the one group looking at this and incorporating all of those three issues and deciding where they'd fit into the prioritization and the shared vision or whether we'd set up individual task forces.

So I could use some guidance from the committee. Again, having been through the work group issues, where we had four work groups that went off in very different directions, then ended up coming back together and stepping on each other's turf, that's partly why I was trying to keep things unified, but perhaps that's a mistake on my part. So I need some help here.

DR. LEONARD: I'm just concerned that each time we meet, we bring up new issues, and we had prioritized issues in the first meeting and we were trying to address some of those at this one, and now we have new ones.

It's also frustrating hearing about Australia and the U.K., and I know their budgets were huge and they had maybe different mandates than what we have, but they produced products that I'm wondering -- I mean, last time we produced a letter to Secretary Tommy Thompson, and this meeting I'm not sure we've produced much of anything.

So I don't want to get three years out and not feel like we've accomplished anything. So I'm not sure of the process because I've never been on a committee like this before.

DR. McCABE: I can tell you what we produced with the first. We produced a white paper, which I would encourage people still to go back to, that was sort of like the basics and I know people are still using in their undergraduate classes. We produced some letters and we had begun to put together the reports of the working groups.

We also put together, on top of the white paper, a document that was really important in regulation and I'm pleased to hear that it's continuing to be implemented, but what we wrote down and what we finally came up with at the end of the three years was very different, and we sent that to then-Secretary Shalala. I guess that's where that went to.

In essence, we then had to write a letter saying that we had tried it out and it didn't work. That was a fairly big deal because that wasn't just a communication. That was a report and we basically had to retract it or at least say that it needed a lot more work.

I think it is important that we have a work product. I was saying, whether we call it a strategic plan or prioritization, you know, of the issues that we've heard about so far, of the issues that we all have individually, what do we think are the biggest issues for the health of the American people? And are there any actions that we can recommend in March? Are there any recommendations that we would make for further information that we need to fine-tune some of these issues?

Go ahead, Emily.

DR. WINN-DEEN: Well, I was going to say I would recommend that we don't try and make one gigantic report that has everything in it.

DR. McCABE: Right.

DR. WINN-DEEN: I would say let's identify our three to five key issues and work on taking each one of those up to the point where we can say we've looked at the background, we've looked at the vision of where we need to get, here's our assessment of the gaps, and here's our recommendation for how to close those gaps.

So that's a framework that applies whatever the issue is, and then we can very strategically become educated about the issue through these kind of lectures. I don't think these two sessions have been productive primarily because we have been receiving information, but we haven't had enough time to digest it and process it back into a recommendation. I don't think they're a waste of time, but we're not ready to Reed's point.

Now, if all Tommy Thompson wants from us is just sort of an update, we can give him an update. If he wants a recommendation, then I think we should very carefully, and I would say it probably makes more sense, at least for individual task forces, to not try and do everything, but try and segregate it into specific subsections where they can go. They can do this summary. We've all heard the summaries, but to just digest that down into a more coherent couple of pages, and maybe we can as a committee discuss the vision and the gaps, and then that committee can then again go back and put that discussion down on paper, so that we actually have a work product.

I wouldn't recommend personally that you have subgroups getting into the strategic vision and the gaps because I really think that's where you lose the consensus process and the multiple -- so any subgroup you have will miss viewpoints, and the whole point of having the 13 people that were selected for this committee is that it brings a diversity of viewpoints, which I think is valuable in framing any of these issues.

But the background stuff that's just work, not discussion, but just sitting down and summarizing, I think that subgroups can do that without any loss of value to the committee.

DR. McCABE: Reed, and then Hunt.

DR. TUCKSON: I'm beginning to get clear I think what I'm looking for, but I still may not be right, and I'm scared about your prior experience, Ed, around the notion of the complexity of multiple small committees, and also, Sarah, I don't know what the staff resources are anymore in terms of how much you have to do it.

But specifically, I think that we had the opportunity to be presented with information

around the adequacy of the oversight of genetic tests and particularly the interagency coordination that's under the Secretary's purview. I think there needs to be a few of us who sit down offline and try to digest that information. I am sure that there will be a few follow-up questions and data points that we all need to be able to say whether or not we feel like there are some reasonable recommendations that can be made back to the Secretary about how well that oversight seems to be going or things that he ought, through his role, be paying attention to based on real thought.

I think that's a doable thing. It may require, after a subgroup looks at what we've learned in the last two days and thinks about it offline and there may be some questioning of the various folks in the interim, one or two very focused presentations at the next meeting that sort of say, specifically for the record, Joe Smith, answer this question because we're not sure about exactly this point that we really need to have understood. So they don't come in and just talk about everything. They answer a very specific question that then allows us to say, okay, we feel comfortable in making this recommendation.

Related to the oversight issue was the second question of the use of pharmacogenomic data to hopefully provide safer, more cost-effective, and better-quality drugs, and that that was a specific issue that came out of our discussion. There are some people that need to sit offline, I think, and digest that. I think we understood that we needed to bring the pharma people in and ask them some very specific, pointed questions around how they behave in this regard so that we would have all the data we need to then be able to say is this a concern or is everything fine?

And then we could either say to the Secretary in this area of cost-effective, better-targeted, safer drugs, the climate exists for appropriate research. There is no need for more safe harbors. There is no need for public/private partnerships for research. Everything is fine. Return to your homes. Or there are some issues that you need to pay attention to, given that everybody's scared to death about the cost of drugs and all this sort of stuff.

Number 3 is the workforce. We specifically then need to do that work, zone in on the questions that we asked, and then, and this is where I get scared because it's too many committees, the fourth one then is again this overarching committee, which I wish Hunt would chair, which has got to do with helping us understand about this exceptionalism deal and how do we bring the people we need into the next meeting to put an even finer point on that?

So I see this as part of a journey. We've started that journey. We've moved the ball from the 0 yardline to the 50 yardline, and between now and the next meeting, I think the committee is going to get it to the 25, and then we'll score a touchdown at the next meeting and then send the results of the game to the Secretary, and then open up a new game.

DR. McCABE: I would just caution us that I think there's still some diversity of opinion in terms of what those three to five big issues would be, and that's what I really saw a group in the interval between now and the next meeting helping us decide what those three to five big issues were, and then if we decide that we need to break those down, that would come after that. But that's my opinion.

Hunt, you wanted to say something?

DR. WILLARD: Yes. You may think we're at the 50 yardline. I think we're sort of putting our sneakers on still.

(Laughter.)

DR. WILLARD: But as I was coming into this committee -- because as I read the charter, it was much broader than what we focused on for two meetings, which has been genetic testing, and I had sort of mentally had three sets of technologies which would be the points of departure to then look at genetics, health, and society as viewed in those three areas.

One is genetic testing, clearly, which we've started, and using genomic information for

risk assessment and anticipating the day for the Secretary when we have a $1,000 genome and how will we deal with that. As I said before lunch, I would come at that from the standpoint of, well, there are several different models that we might consider, and given Model A or B or C, each one of those will have different regulatory, educational, and workforce implications which we're beginning to wrap our arms around.

My frustration this morning was that we started with the implications, but in a vacuum with respect to what the model was, but even when we're done with that, and we're not yet done with that, but even when we are done with that, to me that's only the first of the genetic technologies that are part of our purview and part of the charge.

There are reproductive technologies, genetic technologies which haven't been addressed in depth yet much at all, and then the third is probably therapeutic genetic technologies. I'm thinking along the lines of stem cells, the ability to reset the genome, and to take advantage of the genome, our knowledge of the genome, in order to think of a variety of therapeutic interventions, stem cells being just one of those.

But that to me is very much part of the range of genetic technologies that fall into our charter, and so what I would ask for or that we get to and where I think we need a shared vision, or at least an acknowledged vision, for the group is what does the outline look like that we wish to address over the course of three to four years or whatever our lifespan is, so that I have a sense of whether we're on the 50 yardline or still putting our sneakers on, and to carve it up from that standpoint that says, great, we're part way through with Step 1 of four or Part 1 of eight or Part 1 of two. I think we need that broader structure in order to then sort of frame an agenda going forward.

DR. McCABE: You know, given the hour, we aren't going to get there today or this meeting. We could wait and do that at the next meeting, but that's why I really think it would be worthwhile to have a small group help to guide us at the next meeting, and that was the purpose of the task force, really, was to help to guide that.

DR. WILLARD: But then the output of that task force might be a presentation to the committee as a whole and to the public, rather than "a letter to the Secretary," unless at the end of that process we came up with that outline I was just referring to saying this is now what we consider our -- we're fleshing out the charge that you gave us.

DR. McCABE: Right. There may be some benefit, if we are going to set the course for the next three to four years, to pass that by the Secretary, so that we don't find that we've spent three to four years and we've gone off in a direction that doesn't really interest him. So that was part of the purpose of that.

Barbara?

MS. HARRISON: Just at the risk of being redundant, and I'm trying not to be, but I just want to make sure I completely understand this concept. You're talking about putting together a smaller group of us to decide on key issues for us to address. Are they coming up with a list of 20 possible topics for then all of us to vote on or are they really just coming up with the five topics? And if so, then I hesitate with that because I feel like then they're making this decision for where we're going and we're not really participating in that equally.

DR. McCABE: Realistically, if we're going to be effective, we've probably got to identify two to three topics that we're going to have some answers on within the next one to two years.

MS. HARRISON: And I can respect that. I completely agree with you. I just want to make sure we come to those two or three topics together as the 13-people group, not as a two- to three-people group that talked in between our meetings.

I can also very much appreciate this may be something that needs to happen in between our larger meetings, and I don't know, from the public's perspective, is that something we're allowed to do a

conference call or have something in a different format?

DR. McCABE: We can do it as long as we finalize nothing in those discussions and that the product of those discussions comes back to this group.

DR. WILLARD: I'm guessing we would have no trouble talking and not finalizing something.

(Laughter.)

DR. LEONARD: Well, the question also is could we email in our suggestions of each of us what we think the top three issues would be and build a consensus?

DR. McCABE: Sure. Sure.

MS. HARRISON: I was even thinking maybe even to make it more, I guess, easier for the count again to come up even with the list that you have up here or the list that Cindy was able to come up with and say, you know, check off your three most important topics.

DR. LEONARD: But I think we also have to go back to the SACGHS functions that we were given, and in addition to the things that you named, there's biogenetics and bioterrorism. There's gene patenting and patent policy. I mean, there are other things on this list that weren't even mentioned.

DR. McCABE: Well, let me make another -- Reed? Sorry.

DR. TUCKSON: No, no, just given that there is a growing consensus that I think is emerging, I will withdraw my suggestion, which is getting run over anyway.

(Laughter.)

DR. TUCKSON: But I would just ask, Sarah, in terms of the team, if there are some ways in which through staff work we can just sort of -- I guess what I'm just so terrified of doing is losing what we did these two days. I just don't want to have all that work done just float off into space and get lost because whether we like it or not, we did prioritize a couple of things already, and I just don't want to lose all that effort.

So if there's just a way that staff could help us to attend to that in the interim, and then I will withdraw the suggestion of having separate subgroups that work a little finer on that, until and unless they emerge again as compelling priority issues.

DR. McCABE: Let me make a suggestion that I hope is where the group is. What we will do is that Sarah and her staff will, together with all of you -- so if you have ideas that you feel are not covered in any of the lists that we've heard, get them to Sarah.

We will then finalize a list. This won't be a voting list, but it will be a list to send around to see if we've missed any key issues that you think are critical. We will then take that list and we will have the straw vote on that list.

That will then probably narrow it down I would guess to between five and seven, and then we will have the group that we were putting together work on that, because one of the things I've seen also is that there may be ways of creatively looking at that list of five to seven and it may turn out that it's really two or three, and it's really looking at various aspects of that.

Then we will bring that back and have a lot more time for discussion with less time for presentations. One of the key presentations will be the presentation of that group, who will have been thinking offline and then will bring that thinking back to the public as well as this committee for discussion at that time.

Is that acceptable to everyone?

Sarah, who's on the group? Who was on the list?

MS. CARR: It was Reed, Emily, and Debra so far.

DR. TUCKSON: I think that the task has changed somewhat and I think that there are

some other people who have emerged as very strong voices.

MS. CARR: No, I'm asking for volunteers.

DR. TUCKSON: Yes.

MS. HARRISON: I hate to define, but what is the group doing? The task?

DR. McCABE: The purpose of this group to discuss offline is to have us --

MS. HARRISON: To come up with the list. Okay.

DR. McCABE: You know, I would like to have more of the committee involved with this. If we don't have this group working offline, then what it will be is a few of you emailing Sarah and Sarah and I and the staff developing this, and I would prefer to have it broader than myself helping to develop this agenda.

So it's really helping us to develop the outline of our discussion for the next meeting and from that discussion deciding where we're going to focus, how we're going to organize, and how we're going to pursue the products.

So we have the folks that had agreed. Hunt had also agreed, so I would hope that you would do it. Cindy, you've given some thought to your own priorities, and if you were willing to do it, but anyone who would like to be a part of this, please let Sarah and her staff know.

Thank you.

Do you have enough guidance, Sarah?

MS. CARR: There was the discussion of turning -- I hate to bring this up again -- the minutes of the first meeting into a report. You had suggested that as an idea.

DR. McCABE: I think we've backed off from that. So why don't we decide at the next meeting? A lot of that will have to do with the deliberation of this smaller work -- task force. Not work group. Task force. But we'll shelve that decision for now and make a decision at the next meeting.

MS. CARR: Okay, but just to be clear, the minutes as minutes are approved and they'll be posted on our website.

DR. McCABE: Yes. The minutes are approved as minutes.

Any further discussion?

(No response.)

DR. McCABE: If not, travel safely, everyone. We'll see you March 1st and 2nd in Bethesda at the Pooks Hill Marriott. I'm sorry. The Bethesda Marriott on Pooks Hill Road. It changed its name.

Thank you.

(Whereupon, at 3:47 p.m., the meeting was adjourned.)