Public Comments

DR. LEONARD: So we're now moving into a public comment session and one of our critical functions is to serve as a public forum for deliberations on the broad range of human health and societal issues raised by the development and use of genetic technologies. So we greatly value the input from the public. We set aside time each day of our meeting to hear from the public and we welcome and appreciate the views they share with us.

Today we will be hearing from five speakers and the first will be Michelle Schoonmaker from Association for Molecular Pathology or AMP.

DR. SCHOONMAKER: Hi. Good afternoon. My name is Michelle Schoonmaker and today I'm speaking to you as a member of the Professional Relations Committee of the Association for Molecular Pathology or AMP.

For the record today, AMP is an international medical professional association representing over 1,400 physicians, doctoral scientists and medical technologists who perform genetic testing as well as other testing based on the knowledge derived from molecular biology, genetics and genomics. AMP members practice their specialty in academic medical centers, community hospitals, independent clinical laboratories and federal and state health facilities.

On behalf of our membership, the executive counsel and the professional relations committee of AMP thank the committee for the opportunity to provide commentary on the issue of intellectual property right and patent protection of genetic information.

Our members have had to cease, curtail or alter clinical laboratory testing due to restrictive gene patents for an ever growing list of diseases, including Alzheimer's disease, hemochromatosis, neurodegenerative disorders, congenital deafness, familial breast and ovarian cancer, lymphomas and treatment resistant leukemias.

The Association for Molecular Pathology believes that the human genome sequences are in the public domain and, therefore, there should be open access to them for any clinical application.

Genetic test services are medical procedures and, as such, they should be widely available to promote optimal patient care, medical education and medical research.

The restrictive use of patents or exorbitant licensing fees prevents physicians and clinical laboratories from performing genetic tests, limits access to medical care, jeopardizes the quality of medical care and raises its cost.

Exclusive licenses that limit genetic testing to a single provider are detrimental to the public interests by limiting patent access to testing, medical education and practice, the advancement of medical knowledge and the enhancement of public health.

AMP urges the committee to investigate the clinical impact of gene patents and develop recommendations for steps that can be taken so that patients continue to have broad access to the benefits derived from ongoing and future research on the genetic basis of disease.

Consequently, AMP makes the following recommendations:

All clinical laboratories should be exempt from gene patent restrictions for diagnostic testing in the practice of clinical medicine.

Research funding agencies should oppose patent licensing agreements that inappropriately limit clinical care, the use of medical procedures, medical education and medical research.

Organizations, including universities, that hold patents and require licenses for use of their technology for genetic testing should offer non-exclusive licenses and make available to any—make these available to any qualified clinical—CLIA certified high complexity laboratory on an equal basis.

To ensure that testing remains widely available and affordable, financial terms for test licenses should be reasonable. License agreements should also be free of any terms that limit the number of tests that can be performed by a laboratory or regulate the technical performance or clinical use of the test.

License agreements should, likewise, be free of terms that inappropriately limit research related to the testing or the public dissemination of the resulting research findings.

AMP appreciates the opportunity to address the committee on this very important topic and we invite you to contact Dr. Wayne Grody, who is the chair of our Professional Relations Committee, if we can provide any further information.

Thank you.

DR. LEONARD: Thanks, Michelle.

Any questions for Michelle?

Thank you very much for coming and we appreciate AMP's willingness to continue to provide comments to the committee and work with the committee. We may be calling on AMP for input.

The next speaker is Elissa Levin from DNA Direct.

MS. LEVIN: Good afternoon. To briefly introduce myself, my name is Elissa Levin and I am a board certified genetic counselor and the clinical director of DNA Direct. DNA Direct is a web-based company that has been offering direct to consumer genetic testing and genetic counseling services since February of 2005.

Having worked at two major academic medical centers, I recognized that the current model of delivering genetic services was not meeting the needs of all patients. As a representative of DNA Direct and as a genetics professional, I strongly believe that genetic testing services can be offered responsibly and reliably by a direct to consumer approach. In no way will direct consumer testing services ever replace the traditional models but if direct to consumer is offered responsibly by appropriately trained professionals, these services can be a valuable and viable complement to the traditional model.

I would like to highlight a few critical reasons behind why some consumers have chosen to utilize direct to consumer services just to name a few.

Limited access to genetic services by qualified professionals. This may be due to regional limitations, the overall paucity of trained genetics professionals or current health care networks, and referral patterns that makes it difficult for people to access genetic testing services.

There are also concerns about privacy, confidentiality and genetic discrimination. Again no matter how legitimate the risk of discrimination is based on genetic test results, public fears exist and represent a major barrier to someone who could significantly benefit from testing. Anecdotally, I can say that the majority of consumers who have used DNA Direct services have not sought testing through us for that reason.

Genetic interpretation and support services is another reason. We have had patients come to us because they have received incorrect or incomplete information and no follow up or referral support references through their physicians. In addition, we've also had physicians refer their patients to us. One example is that some physicians refer their cystic fibrosis patients to us for cystic fibrosis carrier screening because they are not comfortable offering or interpreting the test results.

Finally, cost of testing and services. Testing for physicians' offices or academic centers may actually be more expensive than going direct to consumer in some scenarios.

There are many more reasons that I will not go into right now.

Direct to consumer companies need to be responsible for developing and maintaining standards of practice. By setting the bar for reliable and comprehensive services provided by qualified health care professionals, the industry will hopefully move in a direction that will ultimately benefit consumers. In doing so, it is critical that health care providers and policy makers acknowledge that a spectrum of direct to consumer services has emerged.

Consumers must be able to distinguish between these companies and it would be a disservice to consumers if policy were to lump together all direct to consumer services.

There are there key factors that currently distinguish the market.

One: Selection of tests offered.

Two: The laboratories utilized.

Three: The level of services provided and who provides those services.

So, first, all genetic testing is not equal. Under the rubric of genetic tests is a significant range from neutrigenomics and fitness profiles to paternity and ancestry testing to clinically validated tests under medical guidelines in a dozen or so approved tests.

At DNA Direct we offer clinical genetic tests that are routinely offered through most genetic centers. We evaluate each test option in light of its clinical validity and utility and we utilize renowned experts in the medical and genetics fields as well as relevant guidelines to help us convey this to consumers.

Second, it is not up to the direct to consumer industry to defend the validity of laboratory testing. That is an issue that needs to be resolved through CLIA, the FDA and other relevant agencies.

Critics have suggested that FDA approval would be a safeguard for consumers but since the FDA is not currently regulating genetic tests, consumers should be made aware that reliable testing can still be obtained often using the same laboratories that their primary physician would use. Until this issue is clarified, providers of any tests need to be more transparent about where laboratory testing is done, not just direct to consumer. DNA Direct addresses this by partnering only with large reputable CLIA certified laboratories.

The third point relates to the level of services provided and I think this is an extremely important one. Some direct to consumer companies enable consumers to test or order on line, providing no human contact or support by genetics professionals, often referred to as DAT or direct access testing. Pre-test education may be unavailable or incomplete. Informed consent may not be required and post-test results may be disclosed without access to genetics professionals and without interpretation.

I can speak to the fact that DNA Direct has gone to great lengths to address these issues. We have developed a website that offers patient oriented information. All of the materials on our site have been developed to reflect the same core information that is conveyed in genetic counseling sessions, including the limitations of testing.

We all know that people learn differently and the web enables people to seek information in ways that makes sense to them in a time frame that works for them. The web is dynamic and enables us to adapt consumer feedback, updated standards and guidelines, and to add new interactive tools to help people work through the decision making process.

Further, all test orders are reviewed and authorized by a board certified medical geneticist. Signed informed consent through DNA Direct is required. Test results are conveyed via personalized reports that include interpretation based on the individual's personal and family history. All interpretation is reviewed by both genetic counselors and a medical geneticist. Our disclosure protocols are test specific so for some tests reports are only released once they've been disclosed through a post-test discussion with a genetic counselor and the patient. Genetic counselors are available to patients at any point before, during and after the testing process, and all of our clinical interactions are HIPAA compliant.

One last point I would like to raise as a genetic counselor is that I feel it is my professional duty to provide consumers with accurate information. I receive no economic incentives for people to test, though I acknowledge that providing services is our core business model. However, direct to consumer is certainly not the only example of a conflict of interest in the workplace. Other scenarios have proven that genetic counselors can maintain their professional standards even if their salary is paid by the company offering testing.

Why is direct to consumer any different?

It is the responsibility of direct to consumer companies to hire and maintain staff who are competent, qualified and who are morally and ethically dedicated to the patient's best interest. When testing is offered responsibly in these ways, consumers can benefit. Indeed, we are finding that people are appropriately selecting whether or not to test. We know this because currently 38 percent of all people who have tested through our services have had a positive test result. This trend is consistent and this number is significantly higher than is seen in most academic medical centers and it is significantly higher than would be expected in the general population.

In summary, whether or not genetic testing is offered direct to consumer or through other avenues has almost become irrelevant. Genetic testing can be obtained not only through academic centers, physicians offices and internet based direct to consumer companies but also through wellness centers, spas and drive through clinics.

Our society is moving towards more virtual services. Almost 32 million adults in the United States go to the web first for health care information. The media continues to bring genetic testing into the spotlight. Isn't it critical that consumers be able to distinguish between valid testing services?

The direct to consumer industry, health care providers and policy makers should be collaborating to set the bar. We should be asking questions like does the current model, regional medical services, meet the public's needs? Is it effective? Can the traditional system keep up with the advent of new clinical tests? Is it morally and ethically responsible to withhold new validated testing that may reduce morbidity and mortality due to issues of dissemination and access? In these areas, direct to consumer testing is a viable alternative.

We have common goals and we need to work together to ensure that testing is provided safely and effectively to consumers.

Thank you.

DR. LEONARD: Thank you. If you have a moment—question, Kevin?

DR. FITZGERALD: Thanks very much for your presentation and just one quick question. From your experience you mentioned you did some genetic counseling in medical centers. Is that correct?

MS. LEVIN: Yes, correct.

DR. FITZGERALD: So from your experience, are there disadvantages to doing the counseling over the phone without the face to face interaction and then how does your group then address those disadvantages?

MS.. LEVIN: Well, essentially the direct to consumer model is clearly not for everyone and we recognize that and we are very transparent about that. I am continually doing assessments by phone counseling and if someone is highly anxious, if they are not understanding the core information that we're conveying, if we need additional medical records and assessment, or if we don't offer the testing that they're requesting, we refer them to a regional medical center. I try to hook them up with a local genetic counselor or genetics group. So there really is an opportunity for additional referral networks.

Similarly, we have had genetic counselors and physicians refer patients to us, for example, for at risk family members who don't have access to local testing services.

So, yes, I do think that effective genetic counseling can be done by phone but again it needs to be—there need to be additional referral services to make sure everyone is covered.

DR. LEONARD: Joseph?

DR. TELFAIR: Thank you very much for your presentation. The question that I have is that it seems that one of the things that you're advocating very strongly is that there need to be—there's a real need in the industry, the DTC industry itself, to have very high standards in terms of practice.

MS. LEVIN: Correct.

DR. TELFAIR: But there seems to be—that doesn't seem to be a consistent perspective across the board. In your experience so far, and this may not be something you can answer but I think if it comes to this committee, we'll have to make some recommendations, what do you see—first, what do you see as an attraction moving in the direction of kind of raising the bar as you're advocating and what do you see as sort of one or two or maybe a few key things that would begin to move it in that direction given—I don't know if my question is clear.

MS. LEVIN: I will try to answer that to the best of my ability.

DR. TELFAIR: Okay.

MS. LEVIN: We have taken the steps to create our own standards and guidelines that are posted on our website if anybody is interested. It's www.dnadirect.com. And under "about us" we have posted standards and guidelines and that addresses our own internal practices and our own internal standards as well as guidelines for patients that utilize our services. That addresses issues, everything from marketing to testing to accuracy of information, support services, everything that we have heard and we know that are critical of the DTC industry. So we think that there are many different areas that can be focused upon. I just named a few. I hope this is getting towards what you're asking me but I think that there is a lot of room to essentially set that standard and then have others have to move towards it. I think that one of the things that needs to be considered by this committee is how to get that concept out there.

DR. LEONARD: So if I can just follow up or sort of overview, you—from what you have described—are providing fairly state of the art genetic testing and counseling services that are fairly complete and sensitive to whether the patient is really getting what you're saying in genetic counseling, and you'll refer outside, et cetera, but not everyone is providing this level of service.

The testing that you're also providing is pretty much clinical standard testing. It's not nutriceutical type testing.

So in that you're saying that there is a problem out there and it does need to be addressed. We just need to be careful not to lump everyone into the same level of concern category but really look at the individual practice of each direct to consumer provider.

MS. LEVIN: Right. I really would like to emphasize that we view the services that we are providing as a complementary service, an adjunct service, and not a replacement. There are—it is never going to replace the traditional model and I think that what we hear both through the media and through consumers is that they don't—that everything does get lumped together.

There aren't easy ways to distinguish between the good and the bad companies. I think that that really needs to be addressed on a more formal level.

DR. LEONARD: Well, hopefully, the FDA and FTC are listening and taking note of your comments. Thank you very much.

MS. LEVIN: Thank you.

DR. LEONARD: The next speaker is JoAnn Boughman from the American Society of Human Genetics.

Welcome, JoAnn.

DR. BOUGHMAN: Thank you very much.

I'd like to sit down because at this point my comments are actually informal until I get them written up and submitted.

I am JoAnn Boughman, the executive vice president of the American Society of Human Genetics and I would like to just make a couple of comments regarding your discussions this morning on what ASHG is doing about some of these issues around patenting and licensing.

I can tell you that I can't quite come to this issue without bringing my experience of ten years as vice president for research and development and responsible for a tech transfer office that I hope did listen to me at least from time to time.

The executive committee of ASHG is very concerned about the issues that you've been talking about this morning surrounding patents and licenses and are investigating and discussing responses that our organization, that ASHG, may use to address the research clinical interface, ASHG having researchers and clinicians and genetic counselors as members.

We are looking and discussing these downstream effects in clinical testing from the research laboratory perspective. This domino effect if you will.

I would just like to reiterate to the committee as I do to our committees in discussion that there is a difference between legal and ugly. This applies both in patent law and in the contract law situations that we are referring to with the licensing processes. We have to be very careful in discussing practice guidelines, actual practices and certain players that are not playing by the general rules as we have just heard pointed out in another situation.

Our primary focus at this point is that we recognize that our member scientists need to be educated and informed and some first need to be made much more aware about these issues surrounding the patent and exclusive licensing issues so that they can deal from their own scientific perspective as a research scientist understanding that there is going to be downstream effect as these tests are translated.

The scientists should have a role and should have a part in this discussion with their tech transfer and their licensing departments. And if they don't know, if they don't know what this landscape looks like, we cannot depend simply on the tech transfer and the licensing officers to handle this.

We are also looking as a third party when you have the investigator and the companies and so on looking at this. We're also asking questions about our rights as a third party organization and our limitations around advertising or publication issues at our meeting, in our journal, and the idea of what can we do in order to help our members differentiate between practice guidelines and practices of certain organizations or individuals.

We are also asking about what the impact of these discussions themselves and/or commentaries about these issues might have. We think that, in fact, bringing light to this issue, bringing these issues to the fore and making people aware of these differences is a step in the right direction.

I would finally say that ASHG highly values all of its members, whether they are academicians, whether they are clinicians in a different practice, whether they are industry scientists, whether they are consumers, and we clearly recognize the importance of science in academia and from the industry perspective.

Still our leadership is concerned about the actions of the few and the impact that that may have on the many, i.e. the patient community, in the long term. So, in fact, we are actively discussing and looking at these issues and will work with you in any way that we can to help the committee in any further deliberations.

Thank you.

DR. LEONARD: Thank you, JoAnn. Could I ask a provocative question having been president of the Association for Molecular Pathology? One of your points was to look at the advertising and publications and meeting support, et cetera. Are you willing to piss off some of your members by making open statements about their patenting practices and things? Is that what you're implying? Because most societies are not. They want to keep all their members happy and don't want to upset any particular group. So I'm not quite sure what that point was.

DR. BOUGHMAN: We certainly don't want to upset groups of individuals but as you have heard from a variety of people, even within industry, there are many folks, both in the industry and the university side, that are willing to play by guidelines or rules that, in fact, would encompass a more accessible model in the long run. In fact, if you look at our website, for example on somebody who could put a booth up at our meeting, and we are getting a legal reading on this right now, it clearly says that if there is not space available or at the discretion of the organization we could deny somebody the ability to, in fact, rent a booth at our organization. Now whether we have the right to and whether we're going to do that are two different things but I'm saying that, in fact, we are concerned enough about this issue that we are at least asking that question.

DR. LEONARD: Any other questions for JoAnn?

Thank you very much.

Our next and final speaker is Joanna Rudnick from Kartemquin Films. Before Ms. Rudnick begins her remarks, I'd like to point out that her testimony is being videotaped as part of the documentary film she is producing. As a committee governed by the Federal Advisory Committee Act, our meetings are open to the public and may be covered by the media.

Ms. Rudnick?

MS. RUDNICK: Good morning. Thank you. I'd also like to introduce Beth Iams who is my associate producer and filming me right now.

This has been very interesting for me. We also, as film makers, talk about intellectual property issues all the time so this has been highly informative.

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My name is Joanna Rudnick and I am a documentary film maker with Kartemquin Films. I don't know if anyone here has heard of "Hoop Dreams" but that's our most famous, famous documentary. We've been around for 40 years making social issue films.

I'm here today both as a film maker—that' better—and as a woman at high risk for hereditary breast and ovarian cancer.

When I tested positive for the BRCA mutation five years ago, the last place I ever imagined I would be at—would be was on television sharing my story with millions of people. I went to great lengths to keep it a secret. I tested anonymously. I was very worried about privacy and discrimination issues. I told my employer that I just happened to have many doctors appointments, more than anyone else, and I told my boyfriend at that time--he thought I was avoiding him because I didn't tell him I was testing for the mutation.

When the only friend who knew that I was getting tested asked what my results were, I lied and said I was negative because at the time at 27 I really didn't have the right language to be able to deal with this.

I moved home to Chicago in 2003 to be closer to my family and to finally come to terms with what it means to carry this mutation. I began to realize that being BRCA positive was not a curse but an opportunity and I slowly revealed my BRCA status to those around me and set about seeking information to help inform my future decisions about how to protect myself.

As I learned more about the BRCA mutation and those it affects and interviewed over 50 women for my original film proposal, it became clear to me that there really was a very important story here. And "In the Family" is a film about predicting breast and ovarian cancer, the consequences of knowing and knowing that you live with the risk.

In addition to telling my own story, the documentary also chronicles the story of other high risk—stories of other high risk women like Martha Haley. Martha is a three time breast cancer survivor who tested for a variant of unknown significance on the BRCA2 gene. She wonders openly why so few African American women like herself have tested for BRCA mutation. I follow her and she takes this information to her "Y-Me" support group on Chicago's Southeast side and pleads with more women to test so that we can have a better understanding of the role of genetics in African American women's breast cancer.

We also meet Linda Pedraza, a 43 year old BRCA positive mother of two, who is diagnosed with both breast and ovarian cancer while screening herself carefully for both diseases. Knowing she is losing her battle with cancer, Linda worries about the fate of her 16 year old daughter, Nicole. What will this information mean to her future? When should she get tested? You will see more of Linda's story in the upcoming clip I'm about to show you.

Often when you read stories or watch media pieces on BRCA, the mutation characters are reduced to vignettes used to illustrate a purely scientific over simplified ethical statement about genetic testing. The women who live with this information day in and day out and make life altering decisions based on it have much more to teach the general public policy makers, other high risk women and health care providers about the implications, fears and concerns of living with predictive genetic knowledge. They are having discussions in their homes and on their support websites about some of the same issues you're discussing here like fears of genetic discrimination, direct to consumer marketing and patenting.

"In the Family" is the first comprehensive documentary dedicated entirely to telling their stories while asking some of the larger questions about the legal, social and ethical implications and complications surrounding genetic testing for adult onset diseases.

By coming here today, I'm hoping that members of this distinguished audience will connect to the film and see its potential as a resource for public education and professional development.

We are in the process of forming a coalition for outreach and education and want to find partners with expertise in the field and establish networks to help us create and distribute targeted video modules and workbooks.

As the committee works to increase our understanding of how human genetics is impacting society, we want to partner with you to bridge the gap between policy and the people whose lives it most directly impacts.

So I'm about to show a two minute clip here and then I'll show my full 18 minute sample from the film in room 8 so please bring your lunch and watch. We'd love to hear your comments.

Thank you.

(Video presentation.)

DR. LEONARD: Thank you very much.

Any questions for Joanna--for Ms. Rudnick?

So as Ms. Rudnick just stated, a 17 minute clip of "In the Family" will be shown during the lunch in conference room 8, which is out this door. It's the conference room to the right. So your lunch is in 7, the film will be in 8. Committee members, ex officios and members of the public are welcome to view the film and participate in a discussion to follow during lunch.

Ms. Rudnick also will be on hand to answer your questions and comments.

So we are not moving into lunch. Committee members and ex officios, the lunches you ordered are in conference 7, to the left out the door.

The members of the public and those members who did not order lunch, there is a cafeteria on the first floor.

We will reconvene at 1:28. You wanted to be reminded that there is a fire drill scheduled for 1:30. If we do not start at 1:28 then those of you may get caught. So that's why I'm moving it up two minutes.

I didn't ask Sarah for permission to do this so we'll start at 1:28. Otherwise, you'll have to walk up the six flights of stairs. So if anyone comes in here out of breath we will know you were late.

(Laughter.)

(Whereupon, at 12:44 p.m., a luncheon break was taken.)