DEPARTMENT OF HEALTH AND HUMAN SERVICES

Eighteenth Meeting
of the

# SECRETARY'S ADVISORY COMMITTEE
# ON
# GENETICS, HEALTH, AND SOCIETY
# (SACGHS)

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**Thursday**
**March 12, 2009**

**– VOLUME I –**

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Hubert H. Humphrey Building
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Washington, DC

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# C O N T E N T S

C O N T E N T S *(continued)*

C O N T E N T S *(continued)*

P R O C E E D I N G S

[10:02 a.m.]

**Opening Remarks**

**Steven Teutsch, M.D., M.P.H.**

DR. TEUTSCH: Good morning, everyone. Welcome to the 18th Meeting of the Secretary's Advisory Committee on Genetics, Health, and Society. I'm Steve Teutsch. I think I have met most of you.

As most of you are aware, the public, as usual, has been made aware of this meeting through notices in the Federal Register, as well as announcements on the SACGHS website and listserv. We want to welcome all of the members of the public in attendance, as well as the viewers who are tuned in via the webcast. We really appreciate all of your interest in our work.

Please note that we have scheduled two sessions for public comment. One is at 12:45 today, and one is at 9:00 a.m. tomorrow morning. Two individuals have registered to make comments at that time, but there is still an opportunity for others to do so. We would just ask you to sign up at the registration desk.

I want to begin this session by introducing and

welcoming the new faces around the table. We have five new members who have been appointed to SACGHS.

First, Gwen Darien. Gwen is down here. Gwen is the director of Survivor and Patient Advocacy at the American Association for Cancer Research. She was previously the editor of MAMM, a consumer magazine dedicated to women with breast and reproductive cancer.

We are delighted that you are here. Thanks so much.

Dr. David Dale, who is sitting across from me, is an internist and professor of medicine at the University of Washington, and president of the American College of Physicians.

We are delighted that you could be here and join us as well.

Sheila Walcoff will be here shortly, I believe. We welcome her back in her new capacity. Sheila is now a partner with the law firm of McDermott, Will and Emery. You will recall that Sheila served as counselor for Science and Public Health to Secretary Leavitt. In that role, she presented to this committee the Secretary's Charge on Oversight of Genetic Testing.

Another new member is Dr. Sam Nussbaum, who will be here tomorrow. He is executive vice president of Clinical Health Policy and chief medical officer at WellPoint. Sam also has responsibility for HealthCorps, WellPoint's clinical outcomes research subsidiary. You will be seeing Sheila and Sam when they arrive.

One member who could not attend this meeting is Dr. Charmaine Royal. She is associate research professor at Duke University's Institute for Genome Sciences and Policy. She is a former post-doctoral fellow in the Bioethics and Special Populations Program at the National Human Genome Research Institute. We look forward to having her here at the next meeting.

Welcome to all of the new members. Your expertise will serve us well as we move forward with our new priorities. The full bio sketches for the new members can be found in your briefing books.

I would also like to introduce a few ex officio members of our Committee. Dr. Naomi Goldstein, who was at our last meeting, is our ex officio from the Administration for Children and Families, where she is director for the Office of Planning, Research, and

Evaluation. She has previously served as director of the Division of Child and Family Development in the Office of Planning, Research, and Evaluation.

Dr. Peter Kirchner, in the Office of Biological and Environmental Research at the Department of Energy, is filling in for Dr. Dan Drell.

Dr. Alberto Gutierrez is our new ex officio from the FDA, where he is the deputy director for New Product Evaluation in the Office of In Vitro Diagnostic Device Evaluation and Safety.

Stuart Ishimaru is the new ex officio from the Equal Employment Opportunity Commission, the EEOC. Sharon Alexander, special assistant in his office, will be serving as his alternate.

We are glad to see you here today.

Kerry Leibig, whom we met in December, is here to give an update from them later on. Finally, Dan Wattendorf is filling in as the ex officio from the Department of Defense until a permanent ex officio is assigned.

As always, we really value the input from all of our ex officio members and appreciate all of your

contributions.

One more update on our roster. As many of you may know, Professor Paul Miller has begun a short stint as special assistant to the President, and has therefore resigned from the Committee.

We have five main goals for this meeting. First, we have asked our ex officios to give us brief reports on their agencies' missions and relevant developments since our last meeting. This afternoon, we will receive an update on activities relating to DTC genomic services and discuss what steps, if any, the Committee would like to take to address issues of concern. After that, we will be updated on informed consent issues for sharing genomic data and consider what steps, if any, to take in that area.

At the end of today, Barbara McGrath, who chairs our Genetics, Education, and Training Task Force, will provide some preliminary findings from the surveys that we have been conducting.

Tomorrow will be devoted to our work on one of our new priorities, genetics and the future of the healthcare system. We have organized a roundtable of

public and private payers to learn their perspectives on new approaches to coverage and reimbursement, particularly as they relate to genetic technologies and services.

Now let me turn to Sarah, who will remind us of how conflicted we actually are.

[Laughter.]

MS. CARR: Thank you, Steve. Good morning, everyone. I just want to remind you, as I do at every meeting, that you are special government employees when you serve on the Committee, and you are subject to the rules of conduct that apply to regular government employees. You are aware of all these rules. You have a document called Standards of Ethical Conduct for Employees of the Executive Branch.

I just want to take a moment to remind you about two of the rules. One is about conflicts of interest. Before every meeting, you provide us with information about your personal, professional, and financial interests, which is information that we use to determine whether you have any real, potential, or apparent conflicts of interest that could compromise your

ability to be objective in giving advice during Committee meetings.

While we waive conflicts of interest for general matters because we believe your ability to be objective will not be affected by your interests in such matters, we also rely to a great degree on you to be attentive during our meetings to the possibility that an issue would arise that could affect or appear to affect your interests in a specific way.

In addition, we have provided each of you with a list of your financial interests and covered relationships that would pose a conflict. That should be at your seat this morning. If they became a focal point of Committee deliberations, we would ask you to recuse yourself.

I also want to mention the rules about lobbying. Government employees are prohibited from lobbying. We can't lobby, not as individuals or as a committee. We advise the Secretary of Health and Human Services, not the Congress. If you lobby in your professional capacity or as a private citizen, it is important for you to keep that activity separate from the

activities associated with this committee.

Thank you very much. We appreciate how attentive and conscientious all of you are about these rules. Thank you.

DR. TEUTSCH: Thank you, Sarah. Just a few more announcements before we get into the body of our discussions today. At our December meeting we reviewed the draft report prepared by the Gene Patents and Licensing Practices Task Force. That report was released to the public on March 9th. The public comment period will be open until May 15th. We sincerely welcome public feedback so we can take that into consideration.

It has been an enormous amount of work on many people's part, and particular thanks to Jim Evans and all the Task Force members and staff who worked so long and hard to get it to this point.

Since we last met, a number of organizations have held meetings that are of interest and relevant to our work. I just want to highlight a few of those.

In February, Paul Billings served as one of the keynote speakers at the kickoff symposium for the Center for Translational and Policy Research on Personalized

Medicine. The Center is at the University of San Francisco and was founded and is directed by Catherine Phillips. At the symposium Paul informed attendees of the Committee's work and recommendations concerning establishing the clinical utility of genetic tests.

The Advisory Committee on Heritable Disorders in Newborns and Children, one of our sister committees, held a meeting in late February that was attended by SACGHS staff members. Just as a matter of process, our committee no longer has a formal liaison to the group, due to a change in the charter of that group, but SACGHS staff will continue to attend the meetings to stay informed of their activities.

The Institute of Medicine's Roundtable on Translating Genome-Based Research for Health held a meeting in early February that I attended. The meeting included a workshop on developing systems for evidence generation, focused primarily on clinical utility. Members of the Roundtable also developed a plan to begin exploring three subtopics in greater detail, namely the effects of genetics and genomics on drug development, the process for translating research discoveries into genetic

diagnostics, and the potential value of genetics to medicine and public health.

In addition to our roster changes, we have some new staff announcements. Yvette Seger left SACGHS at the end of January. She took a position at Discovery Logic. We wish her the best in her new position. She has made great contributions to a number of our reports.

We also have a new member of the SACGHS staff to welcome. Kathy Camp joined the staff in January, after 20 years of combined clinical and academic work in pediatric nutrition. In addition to caring for children and families with genetic disorders, most recently at the Walter Reed Army Medical Center, she has been serving and providing leadership on a number of committees and organizations related to genetic education and newborn screening.

Appropriately, given her impressive background and interest, Kathy is now the staff lead to the Committee's Task Force on Genetics, Education, and Training.

Welcome to the team, Kathy.

Let me turn to the first order, which is to

hear from our ex officio members. We will be hearing from many of them today and tomorrow. We have particularly asked Barry Straube, who is the chief medical officer for the Centers for Medicare and Medicaid Services, to talk to us.

As many of you are aware, we have done extensive work with CMS on issues related to coverage, reimbursement, and related issues. CMS has been working diligently on many of those. We wanted to have an opportunity for Barry to talk to you about that, with the understanding that CMS works within a closely regulated framework and has authority to do some things. Others, of course, come at them from congressional mandates.

I think that what Barry has to say will be very enlightening. I know you have a deadline on the other end on some meetings, but we are delighted to have you here. We appreciate your continued interest in the work of this committee. Now I turn it over to you.

**Update from Centers for Medicare and Medicaid Services (CMS)**

**Barry Straube, M.D.**

[PowerPoint presentation.]

DR. STRAUBE: Steve, thank you very much. Good morning to everybody. I apologize for having to leave a bit early and not being able to be here the whole time.

As you can imagine, we are heavily involved, with my other colleagues from HHS, with the Recovery Act, with the CHIPRA bill, with implementation of MIPPA, and a bunch of other statutory mandates, in addition to helping the new administration with the preliminary efforts on healthcare reform. We have a number of things on our plates right now.

Genomics, in my mind, has been one of those issues that is seminal to healthcare reform. It is certainly something we haven't talked about in a more broad setting in the past. We are going to need to engage on that. This committee has done some exemplary work over the last number of years that I think sets a wonderful base for a broader national discussion on genomics and how that fits into healthcare reform in general.

What I wanted to cover this morning, in my time frame here, were several things. First, I wanted to talk a little bit about the history of genetic testing in the

Medicare program. We will go through that and I will get into some issues.

If you could go to the second slide there, please. We will then cover some of the specific things that you see listed on the screen here that we have been involved with recently, more specifically over the last year or so since I got more involved with genomics in the agency and wanted to elevate this to a much higher priority.

Go to the next slide, please. The first area here we will talk about is coverage for genetic testing and some history of this. For those of you who have been on this committee, this is probably a frustrating and mysterious area in terms of why does CMS do what they do. Quite frankly, I'm still figuring it out myself, having been at CMS for a few years. This is educational for me, and hopefully for you, also.

Currently, referring specifically to genetic testing services, we cover Medicare beneficiaries for genetic testing services when it is used specifically for the diagnosis of specific diseases. This is propped up historically. I will try to make the case that it is

time for us to be rethinking some of our positions, guidelines, and policies.

We cover cytogenetic testing under a national coverage determination. We make national and local coverage decisions at CMS. About 15 percent of the coverage decisions that are made under the Medicare program are made referable to national coverage decisions. About 85 percent of the coverage decisions are made referable to local coverage decisions, which I will get into, also. The bulk of coverage decisions are being made by a contractor medical director at a local level, sometimes guided by national guidance but often guided by local coverage decisions that are made locally.

Next slide, please. In terms of cytogenetics, over the years there has been at a national level a definition of what we cover referable to cytogenetics. As you can see right here on the slide, the definition of cytogenetics sounds more like something that would have been relevant when I was in medical school, cytogenetics being the "microscopic examination of the physical appearance of human chromosomes." There has been a tremendous change in genetics since we were in medical

school, but that is what is on the books now. I would posit that we need to refine that.

The second bullet defines what cytogenetic tests are deemed by Medicare historically to be reasonable and necessary for coverage. "Reasonable and necessary" is another very confusing term that has never been very well defined. There have been multiple attempts by the agency to redefine this. We are in the process of trying to do that one more time and present that to the new administration.

Basically, the way the statute and subsequent regulation has defined "reasonable and necessary" for coverage is basically as a service, treatment, or device that will lead to improved outcomes in a patient population that is relevant for the Medicare population. It is not just does something work, is it safe, et cetera. It has to actually lead to improved outcomes for Medicare beneficiaries.

The official ones that are listed as definitely being reasonable and necessary I have listed here: genetic disorders in a fetus, such as Trisomy 21 analysis; failure of sexual development; chronic

myelogenous leukemia; acute leukemias; or myelodysplasia. Obviously, this is a very short list of relevant cytogenetic tests. I think we will be struggling in the very near term, let along the long term, with how to expand this list. In doing so, we have to adhere to this definition of reasonable and necessary.

Next slide, please. That is the national coverage-guided cytogenetic testing decision. The local carriers for each of the Medicare administrative contractors, who pay the bills for us, will interpret that national coverage decision at a local level. They have authority to have some leeway. We can give them some guidance but we can't overturn their decisions. They will interpret the national coverage decisions. They have the additional opportunity to make local coverage decisions.

This is a key debate that has been going on for decades. Some people would say, why have local coverage decisions at all, especially in this modern age. Why not have national determinations. What is the difference between the Northeast and the Southwest in terms of genetic testing and coverage. I happen to personally

fall onto that end of the spectrum. I think we should be centralizing things more.

This process of allowing local coverage determinations has evolved over the last three or four decades. When you start delving into it, it does have some relevance. It had more relevance in the past, in my opinion, where decisions did vary regionally. It was standard of practice that governed how people practiced medicine, not evidence-based guidelines.

As we have evolved to the latter, I think that there is some argument for making more national coverage decisions, particularly in complex areas where the subject expert resources are not available at the local level.

Be that as it may, this is the way the law and the regulations currently stand. The Medicare administrative contractors can determine, absent national coverage delineation, which Medicare benefits, including genetic diagnostic tests, are covered within their regions.

Some have said an advantage of LCDs is that they are more flexible. Some would say that they are too

flexible and not prescriptive enough. They may be more responsive to local needs and situations. That has been true historically, but again, the counterbalancing is that we may not need local influences beyond opinion. Finally, they permit local input about coverage. That could possibly be relevant because of certain populations in a geographic area that other parts of the country aren't sensitive to, for instance Indian health centers on an Indian reservation. There might be people in other parts of the country who know nothing about Indian health, Indian customs, et cetera.

There are clearly disadvantages to having LCDs. That includes lack of consistency across the MACs. We get different determinations having been made between different MACs. There is less national input at the local level. They can set precedent for covering something where, if we had broader national input, there might have been a different decision put in place. Finally, I mentioned earlier there are local resource constraints. They just don't have the subject expertise at the local level.

Next slide, please. If you go on the

CMS.HHS.gov website, we do have a national and local coverage decision database. I have referenced here where you can go and find out what LCDs are present or not present. Really, right now they are somewhat limited in terms of local carriers having put local coverage decisions in place. I have listed the two main ones that are referable there. Again, this is woefully inadequate compared to the number of issues that we have talked about at this Committee.

Next slide, please. These are some caveats, or some general principles. If you go to our Coverage Decision Handbook, again which has been developed over a number of years, these are what are in place now and I would say are, arguably, up for review. When we get into tomorrow's session about going forward, I think this is one of the things that Mara is going to charge us with addressing. Keep these in mind for tomorrow. These are some areas that we ought to be reconsidering.

First of all, genetic tests for cancer are only a covered benefit for a beneficiary with a personal history of an illness, injury, or signs or symptoms thereof. A person with a personal history of a relevant

cancer is a clinically affected person, even if the cancer is considered cure.

The caveat here is that genetic testing is considered non-covered for patients who do not have a relevant illness, injury, or sign or symptom of a disease. If they are asymptomatic and don't have any historical evidence of having a genetic disease, under the current statute and regulations that genetic test is not covered. That has been problematic. We have discussed that here.

Next slide, please. The second caveat is, predictive or presymptomatic genetic tests and services in the absence of a past or present illness in the beneficiary are not covered. Specifically, an issue that has come up here frequently has to do with family history. Again, under the statute and regulations Medicare does not cover genetic tests based on a family history alone. How we are going to integrate family histories into coverage under the Medicare program I think is a challenge for us. We need to get beyond that.

Next slide, please. The third caveat is, a covered genetic test must be used to manage a patient.

We do not cover a genetic test for a clinically affected individual for purposes of family planning, disease risk assessment of other family members when the treatment and surveillance of the beneficiary will not be affected, or in any other circumstance that does not directly affect the diagnosis or treatment of the beneficiary.

Again, we all know that all of those issues come up and in some cases are quite important. But under current statute and regulations we are limited by that.

Next slide, please. Question?

DR. EVANS: Does diagnosis constitute a form of management? I'm interested in the wording that you had in the previous slide, "that does not directly affect the diagnosis or treatment of the beneficiary." Is simply making a diagnosis [covered]?

DR. STRAUBE: It can be used in making a diagnosis.

DR. EVANS: Then it falls into the rubric of management.

DR. STRAUBE: The patient has to have signs or symptoms to lead you to want to get that test to make the diagnosis. Mara?

MS. ASPINALL: This also focuses on the fact that it has to be the patient and not a family member of the patient, as well as the other key point to that slide.

DR. STRAUBE: That is correct. That is, obviously, potentially a limitation in some circumstances.

The fourth caveat is, the results of the genetic test must potentially affect at least one of the management options considered by the referring physician, and it must be in accordance with accepted standards of medical care. Some examples that we have listed here in terms of management options might include that surgery would be done or that you might judge the extent of the surgery being done. You might change your surveillance pattern afterwards. You might implement hormonal manipulation or a change in drug dosage. All of these are examples that might justify, again, genetic testing.

Next slide, please. The fifth caveat is, pre-test genetic counseling must be provided by a qualified and appropriately trained practitioner. I think there are several rubs that we have had here at this Committee.

One is, what is a qualified and appropriately trained practitioner. Second, and perhaps more importantly -- I think we can agree on that one perhaps -- do they get reimbursed or not to do the services that they are allowed to do here.

The sixth caveat is that an informed consent form must be signed by the patient prior to testing. That informed consent must include a statement that he or she agrees to a post-test counseling if that is required. Medicare has to supply this form to whoever needs it.

Finally, the next slide, genetic analysis must be provided through a laboratory which meets the American Society of Clinical Oncology-recommended requirements. I have listed those here.

Again, this is what has been built up over the years. There are reasons for why they came to be. I think there are barriers that ensue from some, if not all, of these that have been mentioned here before and are challenges to go through over the short to intermediate term. We have new political leadership in place with a different Congress on the Hill. We need to think about whether they should be changed or not and, if

so, how are we going to change them.

Next slide. The next thing I wanted to talk about was some of our activities in terms of how we are looking at new diagnostic technologies.

If we go to the next slide, there is within CMS a special Council on Technology and Innovation. This was established after passage of the Medicare Modernization Act of 2003. It is chaired by a political appointee from within the agency. I am the co-chair, as the chief medical officer for the agency. We have relevant staff from a variety of parts of the agency, mainly those responsible for coverage and payment policy.

This council is supposed to facilitate the exchange of information about new technology as it comes up, particularly as it raises questions about coverage and payment policy. It is supposed to also help enhance a coordinated response to inquiries from the general stakeholder community when there are questions about new technology and whether or not it is covered, coded, or paid for.

Next slide, please. One thing that this council has done is to publish a guide last fall. It is

on the website. The references are at the end of the slide presentation. We published an Innovator's Guide to Navigating CMS. It took several years to actually document how to navigate CMS. This was done, of course, by us internally.

[Laughter.]

DR. STRAUBE: You can understand that there is a need for such a document.

Again, this guide is publicly available now. It is a start assisting stakeholders trying to understand the processes used to determine coverage, coding, and payment for new technologies under the fee-for-service program. It does provide summarized and simplified versions of existing statutes, regulations, and other policy materials for guidance. It tries to facilitate timely introduction of innovative technology for care of beneficiaries.

This is a reference for you. I think, again, though, this only goes so far. Again, I'm fully committed to trying to link up the Council on Technology and Innovation with this Advisory Committee. I think that we have to take some things back to the CTI from

this Advisory Committee and have that council deal within CMS with some of the recommendations that are made to the Secretary without even having to wait for the Secretary to determine whether he or she wants us to address those specifically.

Next slide, please. As part of the CTI, I also established, about nine or twelve months ago, a Genomics Working Group. This is a multicomponent workgroup that would support CTI specifically on issues of genomics and personalized medicine. Obviously, there are many, many technology and innovation topics that come up to CTI, but in my mind, we are the most behind on genomics and personalized medicine. We are going to be more affected, arguably, by genomics and personalized medicine technology over the next several years. I keep referring to it as a tsunami that is going to affect the agency, and it is getting very, very close.

Again, the issues that we deal with I have listed here. In addition to coverage and payment coding, we include CLIA issues in this working group. We have to look for alignment across not only Medicare fee-for-service, which most people see CMS as running, but also

the Advantage Care Program under Medicare.

I'm happy to see that with the new administration there is going to be something a lot of us have wanted to do for some time now, and that is to get the CHIP program in alignment with Medicare, both fee-for-service and managed care. We are going to see that, and hopefully we can focus on genomic issues across all those product lines, if you will.

We have personalized healthcare issues that go beyond just genomics, but this is a focal place where we are going to coordinate those within CMS. Again, we have increasing relationships with our sister agencies in HHS to try to collaborate more on these topics.

Next slide, please. That covers that particular focus within the agency. It needs to be done, I think, in a much more rigorous and focused manner than we have been able to achieve so far. Again, a change of administration is a perfect time to get refocused on certain issues and take them to the next level. We will report back on that to you in the future.

Now, evidence and coverage of testing under Medicare is another topic that we work on every day. In

addition to the national and local coverage decision process I briefly mentioned earlier, we have other technical advice that is being given to CMS. I wanted to talk about some of the genomics issues that have come up recently here.

At Medicare we have what used to be called the MCAC, the Medicare Coverage Advisory Committee. We changed the name about a year and a half ago to the Medicare Evidence Development and Coverage Advisory Committee to put a focus on the generation of evidence for all decision-making in the agency.

This advisory committee entails 100 people that we appoint. They sit for three-year terms. They are very broadly representative of the healthcare stakeholder community. We pick from that 100-member panel the specific people who have the most subject expertise to advise CMS on specific topics, with the MEDCAC meeting several times a year.

It is a FACA-compliant committee. The Federal Advisory Committee Act has very specific prescriptions about who sits on it and how they can advise us. This is a FACA-compliant committee, so it is similar to the

Advisory Committee.

We also seek outside technical advice, not only from this Committee but through the Agency for Healthcare Research and Quality, AHRQ. We also contract with academic medical centers and other contractors to provide us with technical assistance in any number of clinical and scientific areas.

The MEDCAC, you will see, we can charter to focus on specific issues. Again, pursuant to what we have been doing over the last six to twelve months, I sat down with staff. We had a MEDCAC meeting on February 25th that reviewed current recommendations about evaluating sources of evidence for the patient-focused health outcome benefits from diagnostic testing for genetic testing. We had them focus on diagnostic applications, prognostic applications, and pharmacogenomic applications at this meeting. I will talk about the highlights in a second.

We also plan a second MEDCAC on genomics on May 6th. These are the first two MEDCAC meetings that CMS has had focusing on genomics. I anticipate that this will be a regular thing. I would guess once a year we

will probably have to strive for a genomic issue to be on the docket.

Next slide, please. The participants in the February 25th meeting did recommend to CMS that we ought to use a standard framework and methods and we ought to delineate this in the form of a guidance document in terms of how we are going to evaluate evidence about diagnostic uses of genetic testing. I think that is one assignment that we will have on our plate here over the next year or so.

They also recommended that we encourage evidence from clinical studies with high internal validity about patient-focused health outcomes due to the use of genetic results in care management.

Finally, they recommended we encourage collaboration among CMS and other federal agencies involved with research and healthcare policy pertaining to genomics. As you all know, we sometimes come out with slightly different viewpoints about how to interpret clinical studies. I think that is most prevalent between CMS and FDA. We have different statutory functions, and some of that is natural, but we are trying to get in

alignment with what we are striving to seek from clinical trials in particular.

I wanted to stress the second bullet again. We are really going to focus as we go forward on evidence-based, patient-focused health outcomes in terms of driving our decision-making regulations and so forth about genetic testing.

Next slide, please. This is a side thing that I don't think I have mentioned to this Committee before but that we are working on. Preventive services under Medicare originally were not provided. If you look at the original Medicare statute, there were no preventive services as covered benefits under Medicare. Congress has added these services, interestingly started in the early 1990s or the late 1980s. It has only been over the last 10 or 15 years that preventive services have been added by statute by Congress on an individual preventive service basis.

As you can see here, some of the areas we now have preventive services in include breast cancer, colorectal cancer, prostate cancer, and cardiovascular diseases. There are still a whole host of preventive

services that have not been implemented or covered under Medicare. I think genetic testing as a preventive modality falls into this area. The statutory authority is not there. We either have to get statutory authority or use what I put on the next slide.

Under the Medicare Improvement for Patients and Providers Act of 2008, which passed in July of this past summer, Section 101 gives authority to the Secretary and to CMS to consider additional preventive service benefits through the Medicare national coverage decision process. Interestingly, this is one of the few areas that Congress has actually in the language allowed us to use cost-effectiveness in our decision-making process.

This is a very important modality that may allow us to now start addressing genetic testing issues without waiting for Congress to actually give us the specific mandate.

As I mentioned earlier, in May of 2009 the MEDCAC will meet again to consider screening uses of genetic testing as a preventive service benefit for Medicare beneficiaries. This MEDCAC will be advising us on some of the ways we might take Section 101 of MIPPA

and address some of the issues that you all have been recommending for some period of time.

Next slide, please. I want to do a brief overview of what is happening with CLIA. That is another important area that you have tried to get your arms around and made some good recommendations.

Under the current CLIA regulations, we continue to certify labs where CLIA is applicable, we update the CLIA database with the various issues that I have listed here, and we provide standards for all moderate- and high-complexity laboratory testing, which includes genetic testing.

You may recall that there have been some discussions about whether genetic testing should be separated out as specific different high-complexity laboratory testing. So far our policy has been that whatever applies to other high-complexity testing should apply to genetic testing. If there are relevant things to genetic testing that could apply to other high-complexity testing, we should align those, but we have not found a reason yet to separate genetic testing out as a special circumstance.

Next slide, please. To promote a high-quality, expert level of laboratory performance we have been meeting with federal agency partners, represented here, with other professional societies, advisory and standard-setting groups, and other partners and stakeholders. We will continue to do that, particularly with guidance from this Advisory Committee.

Next slide, please. We are continuing to try to adapt current regulations to the changing needs of the laboratory testing industry. I think this is something we have to continue to work on with this Advisory Committee and our CLIA folks, who have been very open to this. We will be talking more over the next year or two about how we can update our current regulations to meet the needs of genetic testing.

Next slide. We have some additional CLIA projects that I have listed here. I won't go into those in detail. They are in your paper.

The next slide, please. This is, again, educational standards writing, revision of regulations, and so forth.

Next slide, please. What about going into the

future, looping back full circle to national coverage decisions. Currently, we have a pending national coverage decision on genetic testing for Warfarin responsiveness. In August 2008 we opened a national coverage decision, at the national level again, to consider coverage for genetic testing to determine Warfarin responsiveness. We had a technology assessment done through the Agency for Healthcare Research and Quality. They evaluated current evidence from published articles. We anticipate that our proposed decision memo should be out relatively soon, no later than early May of 2009, to meet statutory deadlines.

I was hoping we would have this out by this meeting, but we don't so I'm not at liberty to discuss what the proposed decision may be. I can say, again looping back to what I said earlier, under our coverage decision process with that term "reasonable and necessary," things have to lead to an improvement in health outcomes. Just measuring a porcelain level to determine whether porcelain is high or low in a body is not sufficient, even if it is a safe test and it is an effective test. It has to be able to be used for health

outcomes improvement.

The key question in whether we are going to cover genetic testing to determine Warfarin responsiveness is, does the evidence show that the use of that test leads to improved outcomes in patients who are placed on Warfarin.

There will be a proposed decision put out. Be on the lookout for that. Public comment is then engaged for 30 days after the proposed decision is there. We can change our proposed coverage decision if the public input is sufficient to sway things. That can include new evidence, evidence that was not brought to our attention, or perhaps pointing out that we misinterpreted evidence that was used in any coverage decision. That is soon to come.

Next slide. People should know that at all times we invite public participation in determining and prioritizing topics for consideration of NCDs. Anyone can request a national coverage decision. We have had people coming to talk to us about genetic testing, particularly in the area of pharmacogenomics and screening for heritable forms of cancer. We are open to

having questions about these and [suggestions as to] how people might go about proposing national coverage decisions to be open.

The key stumbling block there is that people have to present to us at least some preliminary evidence that might be interpreted in a way to actually verify that we should have a national coverage decision on a specific test. You can't just send in a comment saying, "We would like you to consider such and such," without having done some homework and giving a reasonable possibility that we might be able to make a coverage decision. We can't do all of the research involved with that up front.

Next slide, please. Complementary to this, we published, in December of 2008, on our website, 20 potential national coverage decisions that we put out for public comment in terms of things that we came up with based on suggestions from people in the general stakeholder work or that we did with our internal brainstorming.

Two of the areas that we put here that we think are ripe for potential NCD topics include gene expression

profiles in oncology, and also pharmacogenomic testing. As I said, we already have the Warfarin testing decision, but there are obviously other ones that we could consider under these two entities.

I think we have given notice that we are going to be considering, going forward, areas in genetic testing for national coverage decisions. These would be the two areas that we would propose doing so.

Next slide. This just lists for you some of what we take into consideration when considering possible future NCDs.

Next slide. I think this is the end. These are references to what I had before.

The final slide is contact information if you want to get a hold of me.

In conclusion, we are doing a lot of things. Two years ago, we were hardly doing anything at CMS on this because we were focused on other issues. This is so important, and this Committee has brought increased awareness to us at CMS, as well as other HHS OPDIVs, about the need to focus on these issues. I hope you are reassured that we are doing something. We have a lot

more to do, and with your help I think we will do the best we can to address these issues.

Steve, I'm open to comments and questions.

DR. TEUTSCH: Thank you very much, Barry. That is extremely helpful. I think those of you who have been with this Committee for a while will recognize the responsiveness that CMS has undertaken to many of the things that have come before this Committee.

I think some of the clarifications about what are the things within your span of control and what are the things that are a little beyond those and more in the domain of Congress will be very helpful. We really appreciate the level of responsiveness and particularly your leadership in moving those things forward. These are many of the things we have had conversations about.

It is really terrific to see some of these actions and, particularly near and dear to my heart, the MEDCAC expanding its role from not just reviewing specific coverage decisions but beginning to look at the criteria. That will be extremely helpful not only for those helping inform those decisions but, I think, for helping people who need to develop tests to get a better

handle on the kind of evidence that is going to be necessary.

Many thanks to you for all of that. Do you have a moment to take a couple of queries?

DR. STRAUBE: Yes.

DR. TEUTSCH: Great. Marc, do you want to start?

**Question-and-Answer Session**

DR. WILLIAMS: I have two comments and a question. I certainly share Steve's congratulations on a really nice presentation about what you are doing.

The first comment relates to personal experience with local coverage decisions. I was on the Wisconsin Carrier Advisory Committee for 14 years, understanding that there is a high degree of variability about the quality of carrier advisory committees and whether or not the local carriers are doing it as a pro forma to be compliant with the regulation or actually using it.

I will use the cytogenetics as an example. It was our local carrier that really initiated a revision of cytogenetic coverage to reflect the current use of that

within medical care as opposed to the historical use in the statute and regs.

I think the advantage that the local carriers have, if they are constituted correctly, is the ability to be much more nimble. If you look at large companies that are innovative, the innovation usually does not come from the top. It usually comes from units within the company that bubble things up.

One of the changes that occurred over the 14 years that I was on there was the venue for the medical directors of the local carriers to get together and talk about what they were doing with local coverage decisions. If a number of people were grouping around a certain area, they could actually identify topics that could then bubble up to the MCAC at that time. That was dismantled at some point in the early to mid '90s.

It seems that that is an opportunity that we really should look to take advantage of. It certainly opens the opportunity for self-interest. It was interesting in our group that if somebody came and presented something that was clearly self-interested and not based on evidence and guidelines, it was the

physicians around the table that did the policing of that. It was not the carriers. I think it potentially could work.

The second comment is, as a medical director of an insurance company for a period of time, I struggle with the same issue of definition of medical necessity and reasonable and necessary. I did find a pragmatic definition. It was Lewis Carroll. To paraphrase Humpty Dumpty, medical necessity means exactly what I say it means, neither more nor less. I think that actually does reflect how we use that term.

Finally, more seriously, the question relates to the upcoming meeting of the MEDCAC in May of 2009. As you are aware, one of the recommendations from the Coverage and Reimbursement Report was to specifically engage the MEDCAC around evidence-based family history and analyzing whether or not it would be possible to take family history where there is evidence and in fact recommendations that this is important in terms of generating testing. That should be used as, if you will, a surrogate for a history of disease.

Is family history in bounds or out of bounds in

the upcoming meeting? I would just point out that, for breast and ovarian cancer testing in particular, there isn't a recommendation relating to family history as defined within USPSTF that would meet the criteria that you list there. I'm very curious as to whether or not this would be an opportunity to actually act on that recommendation from this group.

DR. STRAUBE: That is a great point, Marc. It is timely in that we haven't fully set all of the agenda and the content of that meeting. In my opinion, that is one of many topics that it would be helpful to get some input on, even if it is preliminary.

I think the family history issue is still extremely problematic. It makes logical sense in just a discussion about the issue, but when you try to operationalize things, particularly when you get into payment and reimbursement that might be based on somebody's recollection or misinformation that has been passed on down in the family, especially as we are having to focus on keeping costs under control here for the healthcare system, that is the most problematic part of it.

Jeff Roche, who is my colleague here, will have to make sure we go back and be sure that is included. Thank you.

DR. FERREIRA-GONZALEZ: I also want to thank you very much for your very comprehensive presentation. I'm very excited for all the issues that you are actually working on. I'm glad that you are working on all of these.

Let me bring a point to light. I have been working with several local Medicare directors in genetic testing throughout the country. I have found a difference not only in the understanding of the use of the testing but also the local policies. One of those policies, for example, will cover genetic testing once per lifetime. When you look at genetic testing, which covers not only heritable diseases but also some somatic changes that you are monitoring, there have been a lot of denials for that type of technology.

Since there are significant differences in the local policies throughout the country and some testing is being denied in certain areas, I think that needs to be addressed.

Secondly, you have the Center for Technology and Innovation. I was wondering if there is any exchange or interaction between the work done in that group with the directors at the local level to start bringing them up to speed on some of those issues that you are discussing at the national level.

DR. STRAUBE: I will take the second one first. There increasingly has been more interaction in that regard. I have been meeting with a lot of outside stakeholders that have input into that. I just met the other day, for instance, with the Association of Academic Health Centers, AAHC. They had some concerns that our clinical trials coverage and payment policies were out of sync and/or inconsistent. So we are going back and trying to get all the folks involved there to make sure we can do that.

Similarly, I think that it is a venue where, when we find our payment and coverage is not in sync, we can take it back. First we have to rectify it internally because often it is out of sync because different parts of the agency aren't aligning their policies in the same manner. If we find that they are and it is a problem at

the carrier level, then we go back to the carriers.

I think that gets back to your first question. One of the factors that generates a national coverage decision is in fact inconsistency among our carriers. If it is brought to our attention that they are implementing policy either out of compliance with national coverage decisions that have been made or if they are not consistent with each other, we may open a national coverage decision to make sure that there is uniform coverage thereafter.

If the issue that you are raising is that they have made a national coverage decision but you don't agree with their coverage decision, the first rectification there is to go back to the local carrier to have them reopen their coverage decision if their evidence is wrong. I hope that is clear.

DR. FERREIRA-GONZALEZ: The second one is, how do these directors at the local level get ahead or stay abreast of some of these new technologies, not just reacting when it hits their door. How are they starting to advance or get more education or information about what some of the technology centers that you have are

actually exploring?

DR. STRAUBE: Again, that is a very good question. It is problematic. That is partly based on statutory requirements and partly on regulatory requirements, also.

The local carriers, by law, have discretion. Every case that they are reviewing for any coverage decision technically is a local coverage decision. It may not be written down, but they have the authority to look at a given case and interpret Medicare law, et cetera, as to whether Medicare should be paying for a service. Most of the time, obviously, claims come in and they are paid, and people don't look at all the details. They assume that people are doing things correctly. They may drill down into an individual case.

The rationale behind having that discretion is that on individual cases they would have some flexibility. We are not able to tell them what to do on each individual case.

Since we can't tell them what decision to make, we are also not empowered or authorized to educate them, nor do we have the resources to do so, in terms of

keeping up to date on every technology that is out there. That gets back to what I consider one of the weaknesses of the system because they have limited resources, too. It is a problem that I don't have a good answer for.

DR. FERREIRA-GONZALEZ: The in-depth knowledge from some of the local advisors vary from place to place, too. There are areas that will have very strong advice from individuals who have a lot of knowledge versus other ones who don't have knowledge in that particular area.

DR. STRAUBE: That is correct. I think that some folks would say that the advice that they are getting, too, is biased and subject to conflicts of interest and other issues present there. We are trying to get around that.

We have undergone contractor reform over the last year or two, as you probably know. Rather than having carriers and fiscal intermediaries in every state -- and my home state of California had two contractors for a while -- we now have a total of 15 Medicare administrative contractors. It is [reducing the number] of contractors, which should lead to a consolidation of resources and expertise because they are bringing

together what used to be separate companies into one big MAC. There are fewer advisory committees, et cetera.

In theory, consolidating like that might help improve matters, but that remains to be seen.

DR. TEUTSCH: Do you have time for one more?

DR. STRAUBE: Yes.

DR. DALE: I wanted to ask about the scope of the problem or the database. As a member of the profession and the public, how could I know what is paid for one place or another? Do you have a handle on that?

DR. STRAUBE: David, that is another good question. Although it is bulky and cumbersome, the Coverage Database does have tons of information. I go there myself to ask the same question when it is brought to my attention that something is going on in a given part of the country.

We need to improve in terms of the workability of that database, but it is a searchable database where you can search pending and final decisions. It looks at national and local coverage decisions. It includes all of the MACs that are out there now and what their policies are.

In addition, if you know for your particular region who the MAC is, you can go to their website, which is probably a little more user-friendly for that particular local area. They will refer you in terms of national coverage decisions to the national CMS website.

I think there is lots of information there. It is searchable on our website. If you want to get even more in-depth locally, you go to your local MAC website.

DR. TEUTSCH: Thank you again, Barry. This has been extremely informative. We look forward to working with you and your colleagues. We get to hear from you again tomorrow, and we look forward to that as well.

DR. STRAUBE: Thank you all again. I appreciate this Committee's work very, very much. Dr. Jeff Roche is going to be sitting in for me the rest of today and tomorrow. Jeff was extremely helpful. I gave him the framework and he helped me with the presentation today, as well as several other staff. Thanks again for all your help.

DR. TEUTSCH: Thank you. A few quick things before we get into hearing from our other ex officios. First, Sheila Walcoff is here. We introduced you

earlier. You have met the Committee before wearing a different hat. Welcome.

Speaking of Email and BlackBerries, if we can keep the BlackBerries off the table it would be really helpful.

Let us begin by hearing from some of the ex officios. We have time today, and then we will pick up on some tomorrow. Unfortunately, we don't have time to hear from each of the organizations in the kind of detail that we heard from Barry. We have asked folks to keep their comments to three to five minutes, and that is really hard to do. Hopefully we will have a chance to revisit some of that and go over some quick updates at least today.

We will start with Kerry Leibig. It is Kerry, right?

DR. LEIBIG: It is Kerry.

DR. TEUTSCH: Let's go ahead and start with Kerry, who is going to be speaking on behalf of the EEOC.

**UPDATES FROM SACGHS EX OFFICIOS**

**Equal Employment Opportunity Commission**

**Kerry Leibig, J.D.**

DR. LEIBIG: Thank you. My name is Kerry Leibig. I'm a senior attorney advisor with the Equal Employment Opportunity Commission. Everyone here probably knows this already, but the EEOC is the federal agency that enforces federal laws prohibiting employment discrimination on the basis of race, color, sex, national origin, religion, age, disability, and in retaliation for protected activity.

In 2008, we got a new responsibility when the President signed the Genetic Information Nondiscrimination Act, or GINA as we call it. GINA has two titles. Title I addresses the use of genetic information in the healthcare industry, and it is administered by HHS, DOL, and the Treasury.

Title II, which becomes effective on November 21st of this year, prohibits the use of genetic information in making employment decisions. It prohibits the deliberate acquisition of genetic information about applicants and employees by employers. It has strict confidentiality requirements for any genetic information that an employer does obtain. Importantly for EEOC's role, it requires us to issue implementing regulations by

May 21st, 2009. We are going to make it pretty close, I think.

For the past year we have been working on drafting a proposed rule. On March 2nd, just a couple weeks ago, we published that rule in the Federal Register. Prior to publishing the rule we actually had a Commission meeting where we discussed what was in the rule. We heard from some invited panelists about the impact of genetic discrimination in the work place. Those were the first comments we received about the rule that was about to be published.

If you are interested in seeing the statements that were made at our Commission meeting as well as a copy of the notice of proposed rulemaking and a question-and-answer document that we drafted that goes over the basics of what is in the proposed rule, you can go to our website, which is EEOC.gov, and click on "Commission Meetings." This meeting was held on February 25th. If you hit that link, it has the notice of proposed rulemaking, the statements that were made at the Commission meeting, and a Q&A document. That is quite helpful.

I will tell you now that we will be accepting comments about our proposed rule until May 1st. If you look at the rule itself, it explains how you can submit comments. I suggest that everybody go check it out and submit comments if you have any.

The notice of proposed rulemaking is about 60 pages, so obviously I don't have time to go into it in much detail. I'm just going to hit the highlights so you have an idea of what we are working with.

First of all, both the statute and the proposed rule include a detailed description of what we mean by genetic information. It includes, for example, information about an individual's genetic tests, information about the genetic tests of family members, and information about the manifestation of a disease or disorder in family members or family medical history.

The basic rules, as I said, are that Title II prohibits the use of genetic information in making employment decisions. It prohibits employers from deliberately acquiring genetic information about applicants or employees, and it has strict disclosure requirements. If an employer does get hold of genetic

information, they have to treat it like confidential medical information.

The prohibition on use of genetic information to make employment decisions is absolute. In other words, a covered entity may never use genetic information in making an employment decision.

The prohibition against acquiring genetic information does have some exceptions. They are described in detail both in the statute and then in even more detail in the proposed rule. There are six and they are pretty narrow. I'm not going to go into all of them, but essentially, the first one we call the "water cooler exception." That is, if a supervisor overhears coworkers, for example, talking and one of them happens to say, "Oh, my mother just had a test for breast cancer," they have acquired genetic information. There is an exception that says if you acquire it unwillingly, you weren't seeking it but it came into your hearing, that is not going to be a violation of GINA.

There are five other exceptions, including things such as if you receive information because someone has asked for leave under the Family and Medical Leave

Act. If, in supporting that request, they provide you with genetic information, that is not going to be a violation, either.

In general, any deliberate acquisition by any employer who is seeking genetic information would violate GINA.

Finally, when employers do obtain genetic information through one of these exceptions, they are required to treat it like any other confidential medical information. They have to keep it in a separate medical file, not mix it with a personnel file. They have limited reasons that they can disclose it that are very similar to those reasons given under the Americans with Disabilities Act. For instance, if a government agency is investigating a violation of GINA, they might need to disclose genetic information to those individuals.

The same remedies apply under Title VII. If an employer, employment agency, or labor union are found to have violated Title II, the individual could be reinstated, promoted, or receive back pay, injunctive relief, compensatory damages, punitive damages, unless it is against a government agency. The remedy and

enforcement provisions were really modeled on Title VII, with the idea that the EEOC already has expertise on how to deal with and enforce Title VII and hopefully that will be of assistance to us in enforcing Title II of GINA.

As I said, the proposed rule is 60 pages. There is a lot of detail. We are accepting comments on it until May 1st. I urge you to check it out and submit comments if you have any.

DR. TEUTSCH: Thanks so much. I'm going to move on because we have a lot to cover. I know this is a topic of great importance that we have been really interested in.

Just so you know, we have asked each of our ex officios to talk about new programs that they have, particularly related to our mission, and anything they can say about things that they are doing in response to the American Recovery and Reinvestment Act that can be publicly disclosed.

Let's move on to Gurvaneet Randhawa from the Agency for Healthcare Research and Quality.

**Update from the Agency for Healthcare Research and**

**Quality**

**Gurvaneet Randhawa, M.D., M.P.H.**

DR. RANDHAWA: Thanks, Steve. Most of you are already familiar with our agency, but our mission is to improve the effectiveness, safety, quality, and efficiency of health care. We do it through looking at the evidence base, improving it as much as we can, evaluating it, and then using that to inform decision-making in a variety of different contexts.

You have in front of you a one-page, double-sided handout that lists all the categories that I thought were conceptually different in which our agency is engaged. The first category is assessing the evidence and evidence evaluation, which is used for making clinical guidelines or recommendations. The two examples that I have highlighted here are the U.S. Preventive Services Task Force that is sponsored by our Agency and also for the EGAPP Working Group that is sponsored by CDC.

I think you will notice the topics for the Task Force are on clinical prevention and the topics for EGAPP are focused more on the treatment and management in the

clinical context.

Apart from guideline development, we also work with different stakeholders to look at the evidence base. There are different kinds of evidence reports that are done by our Evidence-Based Practice Center Program. Going to Andrea's comment about how we can inform decision-makers about things that are rapidly happening, we have done a couple of reports for CMS on scans and emerging genetic tests in cancerous and non-cancerous conditions which take a broader look at the evidence and are not the in-depth review that most of the evidence reports are. That is one way of trying to inform decision-makers about emerging tests.

Another thing I would highlight here is, we have done a fair amount of work on family history. There are two reports that have been put out by the CDC on cancer and family history. One was released in 2007. One will be released in a few days. Some of the information from the report will then be leveraged for another NIH-sponsored project on the state of the science on family history and primary care. There will be a conference scheduled later this year.

All of these projects that I mentioned are on evaluating the evidence base, giving the best information to fill the gaps in evidence, or what the quality of information is.

We have a heterogeneity of programs that are trying to build a better evidence base, which is the third category here. Some of the programs are contract-based programs with the DECIDE Network; some of them are cooperative agreements called the CERTS, the Centers for Education, Research, and Therapeutics; and some are regular RO1 and other grant-funding mechanisms.

The ones that I have highlighted here are projects that are underway. The ones in regular text are the ones that have been finished.

The last three categories are on the other page. One category is on disseminating the knowledge from our evidence assessments from new evidence. These projects have been done by the CERTs. Both of them actually were done by one CERT, the one at the University of Arizona, which is a critical partner to us.

The fifth category is on implementing evidence-based recommendations into practice. We are funding a

project on clinical decision support tools for BRCA testing and for gene expression profiling tests. That is underway.

The last category that Barry had mentioned was about conceptual framework and improving methods so that we can have better consistency in how we evaluate and utilize the evidence. We have four different projects that are underway right now. I will stop there.

DR. TEUTSCH: Before we move on, anything you can say about the stimulus package and what AHRQ is doing?

DR. RANDHAWA: There is a lot of work going on right now.

[Laughter.]

DR. RANDHAWA: We have been fortunate, in one sense, that we have built a track record on comparative effectiveness for the past three years. The three elements that were built were assessing the evidence base, gathering new evidence, and disseminating the information in a usable format. They will all be players, I think, in the upcoming programs. There might be some new ones, but that will remain the main thrust of

what we will be doing in that field.

DR. TEUTSCH: Great. Thank you very much, Gurvaneet. Let's turn to Phyllis, who in three minutes is going to say what all of NIH is doing.

**Update from the National Institutes of Health**

**Phyllis Frosst, Ph.D.**

DR. FROSST: Sarah wisely asked that I speak to you about ARA. From my experience, there is little that interests people more about NIH than opportunities for funding, and certainly ARA has really given us a lot in terms of that.

NIH received a total of $10.4 billion. For those of you not intimately familiar with NIH appropriations, in '08, NIH received about $27 billion, so this is a very substantial proportion of our total budget, which creates both wonderful opportunities and a lot of food for thought about the best way to disburse this money both to achieve the aims of the act itself and to achieve scientific goals.

I suppose I should preface any comments and any questions -- that I'm happy to answer once my three minutes are up, and anytime later -- with the statement

that the goal of the act is to create jobs across the U.S. This is, unfortunately, not exactly in line with NIH's mandate, which is to further scientifically-centered progress and to foster the health of the American public. So some obvious things are, unfortunately, not that obvious.

In terms of how that $10.4 billion breaks down, there is $1 billion for extramural construction, repairs, and alterations. This is going to be administered by NCRR, the National Center for Research Resources, one of the 27 NIH institutes and centers. There is $300 million for shared instrumentation and other capital equipment purchases, again allocated to NCRR for support of NIH activities.

Five hundred million dollars goes to NIH buildings and facilities. This and, actually, a little bit of the instrumentation money are pretty much the only thing that stays on campus. Congress, not surprisingly I suppose, funded money that goes out the door to their own districts. In terms of comparative effectiveness research, there is $400 million that comes to the NIH through AHRQ.

If you have been doing the math, I believe what is left over is $8.2 billion in support of NIH's scientific research priorities. This is divided by the institutes along the lines of how the appropriation is, the same percentages. NHGRI's percentage is about 5 percent of the total NIH budget, or about $127 million, which is about a quarter of our appropriation. That is, again, to be spent over two years.

I should point out that two years in government time is not actually two years in time that we are more familiar with. Because we are already about halfway the year, two years is more like 18 months. That money has to be spent and out the door by the end of Fiscal Year 2010, which is the end of next September.

Of that $8.2 billion, $7.4 billion goes directly to the institutes and centers and to the Common Fund, and $800 million goes to the Office of the Director. That is not including Common Fund money. It is supporting scientifically-related research activities that align with the overall purposes of the act. Again, this is a job and financial stimulus package.

In terms of how NIH anticipates the money, I

can say that there have been no end of discussions about exactly the right way to do so, as my federal colleagues will no doubt understand. Those conversations are broadly ongoing. NIH learned a lot of lessons from the doubling and is really trying to apply them in the way that is best for the scientific community in this exceptional case.

The increase is going to go to funding RO1s -- Congress' main priority is always investigator-initiated research -- primarily those that we were not able to pay due to the previous budget constraints, et cetera, but only for two years of funding. Likely scenarios are a smaller amount of specific aims. Again, a four-year RO1 doesn't compress into two years of research, no matter how many people you have in your lab. That is an editorial comment.

DR. FROSST: Supplements as well. NIH is going to fund both administrative and competitive supplements to existing grants, again over a pseudo-two-year period.

You have probably heard a lot about challenge grants. They are designed to focus on health and science problems where progress can be expected in two years.

I was going to keep this really broad and at the NIH level, but I think there is a lot of really interesting stuff that comes up in these challenge grant topics, including the priorities, and I just wanted to bring out a couple of pieces that I thought this group might find really interesting.

Probably the most useful thing, aside from listening to me and any grilling you might like to do at any point, is to go to the NIH website. Whatever we are allowed to say, which increases by the day, is posted on the NIH website.

Probably of the most use of these high-priority topics for these broad challenge areas is the area of bioethics. Highlights include areas on informed consent and data access, ethical issues in the translation of genetic knowledge to clinical practice, unique ethical issues posed by emerging technologies, electronic sharing of health information, and recontact issues in genotype and genome-wide association studies.

There are topic areas on biomarker discovery and validation, clinical research areas, including integrating cost effectiveness, personalized drug

response and toxicity. There is a broad area on enabling technologies and new computational and statistical methods. There is one on enhancing clinical trials specifically for rare disease genetic patient registries.

There is a whole area on genomics. I have a laundry list of subtopics around genome-wide association studies, genomics of eye disease, and the list goes on and on. I think the value of me going through it is probably decreasing the more I speak about it, so again, I would encourage you to go to the website. This is all there. There is a broad amount of information that speaks to that. Thank you for your attention. I will conclude there.

DR. TEUTSCH: Thanks, Phyllis. I'm sure there is a lot of interest in all of that. The deadline is April 27th, I think.

Let's turn now to Robinsue Frohboese from the Office for Civil Rights.

**Update from the Office for Civil Rights**

**Robinsue Frohboese, J.D., Ph.D.**

DR. FROHBOESE: Good morning, everyone. For those members who are new to the Committee, I just wanted

to tell you a little bit about our office.

We are part of the Office of the Secretary at HHS, and we have two major responsibilities. One is the traditional civil rights responsibilities of ensuring nondiscrimination on the basis of race, color, national origin, disability, age, sex, and federally funded programs through the Department. Our other area of major responsibility is the HIPAA Privacy Rule. You can see that our activities really stretch across the Department.

Like other federal agencies, we have been very involved, as of late, both in the Recovery Act efforts, as well as healthcare reform initiatives.

Since we last met, we have been involved in a number of activities that I just wanted to highlight for the Committee, because I think they are of direct relevance to the work of this Committee. The first area is that the Office for Civil Rights has a particular responsibility under GINA and the rulemaking process to modify the HIPAA Privacy Rule to ensure that health plans do not use or disclose genetic information that is protected health information for underwriting purposes.

That is a more narrow scope than the

responsibilities of EEOC, the Department of Labor, CMS, and Treasury, who have the major responsibilities for the employment nondiscrimination and health plan nondiscrimination aspects, but we have been working very closely with EEOC, Labor, CMS, and Treasury in the rulemaking process to ensure that we are coordinated as we move forward.

As Kerry said, EEOC was first at the bat with the notice of proposed rulemaking last week. The Department of Labor, CMS, and Treasury issued a request for information last October, with a two-month comment period that ended in December. I think we chatted briefly about it at the last Committee meeting. They have been very involved in analyzing those comments.

We at OCR have been working with them as we move forward with issuing a rule under GINA. We are coordinating the timing and expect that certainly by May we both will be issuing regulations that will dovetail with one another.

Another major area in which we have been involved is health information technology. As you may be aware, in mid December the Department issued a Privacy

and Security Toolkit. OCR was very involved in developing that with our Office of the National Coordinator here at HHS. As part of that toolkit, we have very specific guidance that applies the HIPAA Privacy Rule to various principles of protecting the privacy, confidentiality, and security of electronic health exchanges. We have a particular focus in that guidance on access to records as well as personal health records.

We have all of the information posted on our website, which you can easily find at HHS.gov/OCR. We actually have a newly designed website that hopefully people will find very easy to navigate. There is a whole piece on the website on health information technology with this application of HIPAA Privacy Rule principles to electronic health records and electronic information exchanges in the healthcare context.

These activities in December were certainly a foreshadowing of what we are currently facing under the Recovery Act. You may be aware that as part of the Recovery Act there is a whole separate act that is called HITECH for short. HITECH actually stands for Health

Information Technology for Economic and Clinical Healthcare Act. The HITECH Act is devoted to health information technology and use of health information technology in healthcare reform.

There is a particular part of the act that specifically applies to the HIPAA Privacy and Security Rules. That part of the act, Subtitle D entitled "Privacy," requires the Department, and OCR in particular, to take three major activities. The first is to engage in a series of rulemakings both to modify the HIPAA Privacy Rule and to issue a series a guidance.

Of note, the HITECH Act now will cover business associates as a covered entity under the HIPAA Privacy Rule. That is a significant new development. It also now requires the Department to promulgate regulations to cover breach notifications to individuals who are impacted by breaches of unsecured information.

All of these regulations and guidance that are required under the act will happen within the next 11 months. Some of them will occur as soon as next month. We are on a very tight timetable to implement these new regulations and pieces of guidance.

The second major area is increased enforcement under both the HIPAA Privacy and Security Rule. The act actually changes the enforcement of the HIPAA Privacy Rule and increases the amount of penalties that we can collect from violators of the rule.

Also, for the first time, it gives authority outside of the Department to enforce civilly the HIPAA Privacy and Security Act. It gives the authority to state attorneys general to actually bring actions in federal court, in coordination with the Department, where there are alleged violations of the HIPAA Privacy or Security Rule. That is a whole other area, and the Department of course will be very involved in training state attorneys general to ensure consistency and uniformity in this rule.

The third major part OCR's piece and the HITECH Act is in terms of public education. Congress has specifically directed the Office for Civil Rights to engage in a nationwide, multifaceted initiative to educate consumers about uses and disclosures of protected health information, as well as their rights under the HIPAA Privacy Rule. That is something, again, that we

need to develop and maintain within the next 11 months.

You can see that we will be busy, as will our partners in CMS, who are responsible for the HIPAA Security Rule, and the Office of the National Coordinator. The HITECH Act does institutionalize that office and allows grants for promoting the use of electronic health records and standard-setting in health information technology.

By comparison to NIH, this portion of the act is small potatoes, but it is certainly big dollars for the Department. There is a total of $20 billion under the HITECH Act, the majority of which -- $18 billion -- goes to CMS to distribute to providers to promote adoption and use of electronic health records.

There is $2 billion that comes through the Office of the National Coordinator, and there is a departmental working group that has been set up to develop proposals and a spending plan to bring before the leadership of the Department. That has been meeting on a regular basis to look at all of the spending plans that have come out of the Recovery Act to make determinations about the most appropriate use of the funds.

I know we are short of time, so let me just highlight one additional aspect that I think is important for the Committee to know about. In January, as part of the Surgeon General's release of the new Family History Tool, we took the opportunity to also provide information through frequently asked questions about the HIPAA Privacy Rule and how it impacts ability to collect and use information. That also is on our website. I think it is a valuable addendum to the Family History Toolkit.

That and other good things are on our website. Hopefully it will give you insights into other activities in which we have been involved.

DR. TEUTSCH: Thanks, Robinsue. Obviously, those impact rather broadly. Thank you for that. Why don't we turn to Sarah Botha from the Federal Trade Commission.

**Update from the Federal Trade Commission**

**Sarah Botha, J.D.**

DR. BOTHA: Good morning. My name is Sarah Botha, and I'm an attorney in the Division of Advertising Practices at the Federal Trade Commission.

Just to provide a bit of background that

probably most of you are familiar with, FTC is a national consumer protection agency. Our mission is to prevent unfair and deceptive acts and practices in commerce. That would include misleading advertising.

In the Division of Advertising Practices one of our primary areas of focus is on advertising claims for products that promise health benefits. We think that is particularly important because consumers not only can lose money but potentially have health impacts if they are misled by advertising claims in that area. That includes over-the-counter drug products, dietary supplements, and also direct-to-consumer advertising of genetic testing.

My predecessor, Matt Daynard, for whom I'm taking over, worked with FDA and CDC a couple of years ago to put out a consumer-directed educational piece on at-home genetic testing to advise consumers about the current limitations with these tests and what kind of information they can get from those tests. It also recommended that they consult their healthcare practitioner when using and interpreting the results of testing.

Consumer education is a big mechanism for us to help prevent consumer deception. In addition to that, obviously we do law enforcement actions. We also work with industry on self-regulation, where possible.

In this area currently, we are definitely open to considering additional consumer education and whether we can help consumers with any new information about developments in genetic testing. We do have a couple of inquiries right now with some of the companies that are advertising directly to consumers.

The inquiries are not public, so I can't really provide much detail, but we are talking to the companies, reviewing their advertising, and comparing it to the state of the science right now. Hopefully we will be able to have some action this year.

DR. TEUTSCH: Great. Thanks, Sarah. Let's turn now to the Department of the VA and Doug Olsen.

**Update from the Department of Veterans Affairs**

**Douglas Olsen, Ph.D., R.N.**

DR. OLSEN: Hi. I'm Doug Olsen from the VA. I'm a nurse and ethicist, and I'm here for Ellen Fox as her alternate.

What is going on over at the VA in clinical services is that our Patient Care Services is currently in the process of hiring a director of molecular medicine to oversee and coordinate efforts in the clinical genomics and related areas, proteomics and the other -omics. A well qualified person has been identified and a budget has been allocated.

It will be a program to provide education and clinical guidance to physicians, nurses, lab techs, social work, et cetera, as well as education for patients. There are plans to start a central clearinghouse for genetics resources through that office. However, the lead for the program is really just coming on board. It is going to take a couple of years for him to really implement those plans.

The Genomic Medicine Program Advisory Committee, which we have reported on here before, was formed in 2006 and has members with expertise in clinical and research aspects of genomics. There is even some overlap between this Committee and that committee.

Based on their recommendation, focus group surveys were conducted with veterans to assess their

knowledge of genetics and genomics and also their support and expectations for the Genomic Medicine Program. The focus group survey was conducted by the Genetics and Public Policy Center at Johns Hopkins, and the results will be published. I think they are in press and due to be out this spring.

This committee will continue to monitor the Genomic Program at VA and provide suggestions about research and clinical programs.

There are two programs for IT infrastructure that were recently funded and are in the development phase to database genomic, genetic, and clinical information research and planning. One is the Genetic Information System for Integrative Science, GenISIS, and it will integrate data from individual research studies, both genetic and clinical, to repurpose the data, reanalyze, and produce new funding.

The other is called VIICI, Veterans Informatics and Information and Computing Infrastructure. That will integrate existing databases as well as new data to extract information and meaning. It will provide data in a secure, high-performance computing environment.

As far as education, VA is supporting a program with the National Coalition for Health Professional Education in Genetics, NCHPEG, to develop an interactive educational program on familial syndromic colorectal cancer. The content will include pathophysiology, risk assessment based on family and medical history, screening, management, testing, and counseling. It is intended for a wide audience of healthcare professionals. It will be Web-based, and it is scheduled to be ready to pilot-test by the end of the fiscal year.

As for research over at VA, in 2008 there was a funded genome-wide association study on amyo-trophic lateral sclerosis to examine gene-environment interactions in the development of the sporadic form of that disease. There are also planned system-wide studies in Parkinson's disease, PTSD, mental illness, diabetes, breast cancer, and pharmacogenomics, amongst other things.

There are also over 140 investigator-initiated merit-reviewed projects related to genomics on a wide spectrum of conditions prevalent in veterans, including schizophrenia, PTSD, bipolar, Alzheimer's, cardiovascular

disease, diabetes, substance abuse, stroke, chronic viral infections, autoimmune disease, Gulf War illness, and cancers of the prostate, breast, colon, bladder, and lung. Those are the things that are going on over at VA.

DR. TEUTSCH: Terrific. Thanks, Doug. Naomi Goldstein from the Administration for Children and Families.

**Update from the Administration for Children and Families**

**Naomi Goldstein, Ph.D.**

DR. GOLDSTEIN: The Administration for Children and Families is part of the Department of Health and Human Services. It is more or less the "HS" in "HHS." We are a human services agency, and we include the TANF public welfare program, Head Start, Child Support Enforcement, Child Welfare, Child Care, and a large number of smaller programs.

I'm new to the Committee. I have been impressed with the range and number of departments and agencies for which the Committee's work is relevant. It is not yet clear to me the extent to which the work of my own agency is relevant for the Committee, but I certainly stand by to be helpful if I can.

Just for your information, the Recovery and Reinvestment Act does provide funding for eight ACF programs. That includes Early Head Start. The act more than doubles the size of the Early Head Start Program for kids aged zero to three.

There is funding for childcare subsidies for the TANF welfare program, for Child Welfare, for Child Support Enforcement, and for a new initiative to build capacity in nonprofit organizations. I will leave it there.

DR. TEUTSCH: Great. Thanks, Naomi. Peter, do you want to talk a little bit about what is going on at the Department of Energy?

**Update from the Department of Energy**

**Peter Kirchner, M.D.**

DR. KIRCHNER: I'm Peter Kirchner. I represent the Department of Energy, specifically the Office of Science's Office for Biological and Environmental Research. We support research at universities and Department of Energy Laboratories in a variety of areas, including molecular biology directed at DOE missions, currently primarily in bioenergy, waste cleanup, and

carbon sequestration.

We support a small program in radiochemistry research and radionuclide imaging instrumentation that in the past created much of the scientific underpinnings for nuclear medicine. This program is being reoriented toward more focused support of the bioenergy and environmental remediation projects that we are now focusing on.

We also have a small program devoted to low-dose radiation biology research, which hopefully might actually come up with information regarding genetic susceptibility to radiation-induced cancer, which would be, of course, very nice. Apart from this, we have very little else of pertinence that relates to human medicine and genetics.

We have had a program called ELSI, Ethical, Legal and Societal Issues, that has been active since about 1990, two or three years after the initiation of the Human Genome Program. In the past the ELSI Program has focused on genetic privacy, education, and intellectual property protections, but it has not endeavored to support studies in the broad portfolio of

potential issues.

The DOE's ELSI Program is now transitioning to new aims, namely to support bioenergy sustainability issues, synthetic biology, and nanoscience, things that are of great importance to DOE's current mission.

Our office does, however, support the Department of Energy's Human Subjects Protection Program, which is responsible for all human subjects protection in all DOE sites and any research done with DOE funds. It is this program that does intersect somewhat with research that is directed at genetic testing, primarily in two large cohorts that have been studied through DOE.

One of them is the long-term monitoring of atomic bomb survivors in Japan, initially under the Atomic Bomb Casualty Commission and, since the mid '70s, under the renamed Radiation Effects Research Foundation. This looks at health effects both on the survivors as well as the children of survivors. There is genetic research now being done to try to correlate the health outcomes of the radiation effects with potential genetic markers.

Another large cohort that is within DOE deals

with the major and lasting charge for monitoring worker safety. Since the establishment of the Atomic Energy Commission following World War II, DOE has had major responsibility for nuclear materials, nuclear weapons manufacturing, and the related hazards that have been associated with a variety of job-related illnesses. These are being actively monitored through health programs.

A number of universities and outside agencies are mining this information and relating some of the results of these environmental effects to genetic testing in recent times. So we do oversee these areas, but apart from that we do not have a specific program ourselves in this area.

Of course, as you know, there are various preliminary results regarding potential genetic susceptibility to various things such as lung disease. That, I think, summarizes our current activities. Thank you.

DR. TEUTSCH: Thanks, Peter. We obviously have only gone through some of the agencies, and it is great to see both the breadth and depth of all the things that

are going on. Having cut Marc off earlier and knowing we only have a few moments, if there are some specific questions for today's speakers, let's take advantage of the few moments we have to raise them. Marc Williams.

**Question-and-Answer Session**

DR. WILLIAMS: This is directed to Kerry and to Robinsue, and it relates to what I perceive as an overlap between Title I and Title II of GINA. That relates to self-insured employers, who have not only the traditional role of the employer but also insure their workers through a self-insurance. There are a variety of mechanisms under which that insurance is administered, but they do have rights to certain aspects of protected health information. I'm just curious how your two groups are interacting around that area.

DR. LEIBIG: There is what we call a firewall between Title I and Title II which says that for any remedy that you get under Title I for a health insurance-related violation, you cannot make a claim under Title II. One of the things we have asked for comments about in our notice of proposed rulemaking is thoughts about how the firewall might be further explained.

There certainly was an understanding in Congress that they wanted to avoid double liability, I guess you would say, and there are methods being developed to do so.

If an employer makes an employment decision that involves health benefits, that would be covered under Title II because Title II prevents discrimination in any employment-making decision, which could include health benefits. For example, if they decided not to hire someone because genetic information that they had made them believe that they would have to pay more for their health insurance, that would be a Title II violation.

In the situation you described where they also would be making decisions as a health insurer, any decisions that they made in that role would be covered by Title I.

DR. FROHBOESE: I think you summed it up very well. It has been a topic of ongoing conversation, as Kerry said, and one which they are looking for comments on in terms of the EEOC proposed rule.

DR. LEIBIG: When we were writing the proposed

rule, we engaged in many, many months of interaction with the Title I agencies and OMB to make sure that we addressed that problem. In fact, that is why we haven't published it until now.

DR. FITZGERALD: Just a quick question for Sarah. When you talk about the FTC regulating health claims, what definition of "health" do you use? Do you talk to the other agencies about what that might be?

DR. BOTHA: Yes, we talk to other agencies. I don't know if I'm going to answer your question about our definition of "health" very well.

We have memoranda of understanding with FDA, for instance, on drug advertising, where FDA regulates prescription drug advertising and we regulate over-the-counter drug advertising. Generally, we would defer to FDA's interpretation of the scientific standards because we are really not a scientific agency. We might retain experts on particular issues, but we certainly consult with other agencies on those issues.

DR. TEUTSCH: One last question or comment?

[No response.]

DR. TEUTSCH: We also, obviously, can invite

some of these folks back because there are lots of meaty topics here for future meetings.

Thank you to all of you. It is very gratifying to see all the work that is going on throughout your organizations. We will hear from several of the others tomorrow in the few moments that we have.

We will break for lunch. For those of you who ordered box lunches, they are available outside. For those of you who didn't, the cafeteria is just down the hall. We will reconvene at quarter of one.

[Lunch recess taken at 11:58 a.m.]

+ + +

AFTERNOON SESSION

[Reconvened at 12:49 p.m.]

DR. TEUTSCH: As we move into this afternoon's session, I just want to ask the folks who are around the table to please speak loudly. I understand that we are not projecting all that well to the back of the room. When you speak, please make sure your mics are on but try and project as best you can.

One of the critical functions that SACGHS has is to serve as a public forum for deliberations on many of the human health and societal issues raised by the development of genetic technologies. One of the principal ways we get that information is through the comments we receive from the public.

As you all know, we set aside time at each of our meetings to hear from those who would really like to bring issues to our attention. We welcome and appreciate the views they share with us.

We have, I believe, one speaker who has asked to come before us today, and that is Theresa Lee, the vice president of payment and healthcare delivery policy for AdvaMed, the Advanced Medical Technology Association.

We welcome your comments. Please go ahead.

**PUBLIC COMMENTS**

**Comments by Theresa Lee**

**Advanced Medical Technology Association (AdvaMed)**

MS. LEE: Thank you. Good afternoon. My name is Theresa Lee, and I'm here on behalf of AdvaMed, the Advanced Medical Technology Association. AdvaMed represents the medical device and diagnostics products industry.

AdvaMed's members constitute nearly 90 percent of the healthcare technology purchased annually in the United States and more than 50 percent purchased annually around the world. Our members range from the largest to the smallest medical technology innovators and companies and include a significant number of in vitro diagnostics firms that are hard at work developing and refining tests that are used in all settings -- physician offices, hospitals, clinical laboratories, at the bedside, and at home -- to provide the information health professionals need to prevent, diagnose, treat, and manage disease.

Over the years, AdvaMed has followed and supported the work of this Advisory Committee, especially

your work on the issues surrounding patient access to genetic tests, your interest in the way tests are evaluated, and your attention to the methods insurers use to make coverage and payment determinations. We have offered our support by providing comments to your staff on draft reports, by sharing analyses we have commissioned, and by supporting you and your mission.

I have several points to make today. First, I'm here to let you know that AdvaMed supports reform of the U.S. healthcare system in order to achieve expanded patient access to quality care at an affordable price. Because healthcare providers rely on clinical diagnostic laboratory tests to inform and guide much of the care that they deliver, these tests play a critical role in determining whether we will achieve a more efficient and affordable healthcare system, whether we will achieve better quality outcomes, and whether we will meet patient needs.

We ask the members of this Advisory Committee to work closely with the White House and HHS officials to develop a reform plan that builds on the promise that diagnostic tests offer.

In particular, we urge you to continue to point out the need for health care that is both personalized and preventive. We are convinced that diagnostic tests, which currently account for only 2.3 percent of U.S. healthcare expenditures and about 2 percent of Medicare expenditures, can play a central role in heading off and preventing disease. As you know, prevention is regularly included as an essential component of a reformed healthcare system.

We think up-front spending for promising prevention and screening services, services not typically covered by insurers due to their focus on reactive care, will pay dividends over time.

This group understands fully how new advanced diagnostic tests that harness molecular, genomic, and proteomic technologies can help predict an individual's response to therapy, how they can lead to a better assessment of patient risk for developing diseases like cancer or diabetes, and how they can identify the biological mutations that are the markers of disease. We need to take steps to ensure that the proper incentives exist to encourage their development and use.

This leads me to my second point, the need for a modernization of the Medicare clinical laboratory fee schedule. We are pleased that you have identified coverage and reimbursement as a high-priority issue for the Advisory Committee. We believe that reform of the current Medicare payment system for clinical diagnostic tests is long overdue. Its shortcomings have been documented in numerous blue ribbon reports and studies, including your 2006 report. Because it serves as a benchmark for private payers, the Medicare fee schedule impacts the entire healthcare system.

What is most troubling to us is that the promise we see for advanced diagnostic tests in advancing personalized and preventive medicine will not be realized unless we put into place proper mechanisms to cover and set rates for new molecular tests.

Medicare needs to find ways to draw on the expertise of the laboratory community to factor in the value of these new tests and to set payment rates that spur continued innovation.

Third, we commend you for identifying the evidentiary issues associated with assessing the utility

of diagnostic tests as a priority matter for the Advisory Committee. Diagnostic tests pose difficult challenges for technology assessors, and we believe that current evidentiary standards used to evaluate therapeutic products and procedures may not be appropriate for diagnostics. We hope that your attention to this matter will lead to more appropriate standards.

I would like to conclude my remarks by reminding this Advisory Committee that it has been nine years since the Institute of Medicine completed its assessment of Medicare laboratory payment policies. The report the IOM published on this effort called for a series of fundamental reforms of Medicare's clinical laboratory fee schedule, most of which have gone unaddressed.

The report also warned that problems with the outdated payment system could threaten beneficiary access to care and the use of enhanced testing methodologies in the future.

AdvaMed believes that the current Medicare payment system for tests is a poor foundation for new molecular tests, including genetic tests. The enhanced

testing methodologies referenced in the IOM report are here today, and both device innovation and patient access are threatened if we do not correct the way new tests are valued and priced. Thank you for your time today.

DR. TEUTSCH: Great. Thank you very much. Obviously, we share many of those concerns. We had a long discussion this morning with Dr. Straube. We talked a fair bit about this is going to move forward. I wanted to ask you one question that relates to all of this, particularly since you emphasized the prevention component. That is the one area, of course, where CMS can use cost effectiveness analysis.

As we move to an era where clinical utility is going to be the sine qua non of what gets done and we see all the comparative effectiveness legislation that hopefully will help inform us and will also provide some direction to industry as to the kind of information that is going to be needed, I wonder if you could reflect upon what the industry can do to help us get the cost effectiveness information that is going to be needed to make the compelling case to move that field along.

MS. LEE: At AdvaMed, we are very strong

proponents of trying to show the value of technology. We have an entire Value of Technology campaign. One of the ways we do try to show value of certain technologies is to look at cost effectiveness. We do not think that cost effectiveness should be used as a general matter in making coverage decision-making, but we are aware that under the MIPPA provision that Dr. Straube referenced this morning that outcomes and expenditures are a consideration and that it may be appropriate in that context under MIPPA to look at cost effectiveness.

In the context of diagnostics, we are actually in the process of working with ACOA on commissioning a white paper specifically to look at the value of screening. It gets at this issue of trying to make sure that we are looking at prevention and integrating in vitro tests into that picture so that we maximize the value of many tests that are simply under-used today.

In terms of delivering the kind of information that you are talking about, Dr. Teutsch, I think that we will be touching upon cost effectiveness of certain key tests. I think that we are going to be featuring four specific case examples of screening tests, and cost

effectiveness will be one of the considerations. So we are going to try to deliver that information to you.

DR. TEUTSCH: Any other comments?

[No response.]

DR. TEUTSCH: Thank you very much. We look forward to continuing to work on these challenging issues.

MS. LEE: Absolutely. Thank you very much.

DR. TEUTSCH: Any other public comments that I'm not aware of?

[No response.]

DR. TEUTSCH: Then we will move forward. At our last meeting, in December, we discussed one of the new priority topics, which was the consumer-initiated use of genomic services. We decided we should review some of the recent activities and developments in the field and see how the Committee can contribute to the current debate and discussion.

We have invited several speakers to update us on their activities in this area. Sylvia Au, who led the Committee on this priority, will lead this discussion this afternoon. Sylvia, it is all yours.

## CONSUMER-INITIATED USE OF GENOMIC SERVICES

### Session Overview and Purpose

### Sylvia Au, M.S., CGC

MS. AU: Thank you, Steve. You know how important direct-to-consumer is when our esteemed colleague Jim Evans is quoted in a magazine on direct-to-consumer genetic testing. He is quoted in an article titled "Tempted by At-Home Gene Tests." He says, "Without guidance testing results are, arguably, worthless," which is a typical Jim statement, for those of you who know Jim.

[Laughter.]

MS. AU: The purpose of this session is to provide an update on government and private sector activities related to direct-to-consumer genomic services since the session on personal genome services that we had in July 2008. After the speakers, we are going to be looking at some short-term action steps that the Committee might like to consider to help address some of the issues around direct-to-consumer genomic testing.

Our first speaker is familiar to all of us. it is Greg Feero. He comes to us from the NIH National

Human Genome Research Institute, and he is the chief of the Genomic Healthcare Branch.

**Outcomes of an NIH-CDC Workshop on Personal Genomics (December 2008)**

**William (Greg) Feero, M.D., Ph.D.**

[PowerPoint presentation.]

DR. FEERO: Good afternoon. Thank you for having me before you. I'm actually a substitute for Muin Khoury, who could not be here today to present this. I think that most would agree that probably this meeting that I am about to report on was largely his brainchild.

I am going to talk to you briefly about a meeting that was held on December 17th and 18th at the NIH, sponsored in part by the CDC as well, to look at the scientific foundation for the most recent wave of direct-to-consumer testing vis-a-vis the genome scan type of technologies.

To give you a little bit of context for the meeting, personal genome-wide scans have become quite inexpensive. The cost is going down, it seems, on a quarterly basis. They are directly available to the public.

The research discoveries that are coming from genome-wide association studies that relate to the genetics of common complex disorders are very rapidly being moved from the research setting directly to a place where they can be marketed to the public and also to healthcare professionals. Sometimes this isn't even within days of publication, it is the same day of publication, as was the case for some recent prostate cancer discoveries.

Obviously, there is vigorous debate about how and when to translate these types of research discoveries from genome-wide association studies to healthcare applications to make them available to the public. This Committee has talked about many of these issues in great detail over time.

The particular meeting that occurred on December the 17th and 18th really focused largely on the issues of clinical validity, clinical utility, and education, I would say. Some of the other issues, although recognized as being very important, were not really a central focus of the meeting.

I think for everyone that was present at the

meeting the goal was to take the complex scans, who are in this far realm of potentially dubious use in clinical care and for healthcare purposes, and really migrate them back, through developing an evidence base, to a position here on this scale where they actually become a part of preventive services.

As I mentioned, the meeting was sponsored by the NIH and the CDC. A really major co-sponsor was the National Cancer Institute. The National Heart, Lung, and Blood Institute also participated, as well as the NHGRI.

The meeting itself was a two-day event. There were approximately 100 attendees. It was a jam-packed agenda. There were 40 speakers and panelists. I'm afraid some of the speakers were quite frustrated because they were given a very short time period to get very complicated stuff across, but there was ample time, I think, for discussion in many of the sections. That was part of the reason the speakers had such a short time to actually speak.

Diverse perspectives were presented, including government, academic, and industry perspectives. There was a blend of both didactic presentations and mediated

discussion panels on the topics at hand.

It was broken down into several sessions. I will just go quickly over those and the people that chaired them. The first was getting people on a level playing field with regard to the basics of genetic and genomic profiles and risk assessment in personalized health. That session was mediated by Greg Downing.

The next really dealt with the scientific foundation for which the variants could be included in genome profiles and essentially dealt largely with the issues surrounding clinical validity of the markers.

I think most people at this meeting felt that, at least for the major purveyors of the genome-wide scans, the analytic validity was not so much in question for the markers. The clinical validity is really where the discussion started.

Then there was a large discussion about how you go about establishing the clinical validity and utility of genome profiles.

The following day there was further discussion around case studies for clinical validity and utility, a discussion of models that could be used that go beyond

the randomized control trial to demonstrate clinical utility, and then, finally, a discussion of next steps.

The most immediate next step from the meeting was the development of a manuscript based on the content of the meeting. That is currently in preparation. I believe it is slated already for one of the major genetics journals. I thought I would go briefly over the five main points that came out of the meeting.

The first, and you will hear more about this this afternoon from Amy Miller from the PMC, is that there was a general consensus -- and there was already movement in this direction prior to the meeting -- that the industry itself that is offering these types of tests should work to develop industry-wide scientific standards for personal genomics. That really has to occur in partnership with other groups besides industry because a lot of the information that the industry relies on to make their risk assessments is generated from studies that are well beyond their means to conduct on their own.

The next is to develop and implement a multidisciplinary research agenda. It was recognized at the meeting that no one organization or one bin of

science would be sufficient to move the ball forward in terms of understanding the utility of genome-wide profiles. Novel public-private partnerships would have to be developed that encompass folks from multiple disciplines and perspectives to move this forward. To some extent, the GaapNet proposal brought forth by Muin Khoury as a potential architecture for public-private partnerships, was also discussed.

Another is, enhance credible knowledge synthesis and dissemination of information to providers and consumers. This is really to reinforce a lot of the work that AHRQ, EGAPP, and others have been trying to do. It was discussed extensively that providers, policymakers, the public, and public health officials all need unbiased sources of information that are truly accessible for this type of testing. That accessibility means not only from a literacy standpoint but also accessible from a cost standpoint.

There was also a feeling that not only do you need to have the information but that there needs to be somebody that is familiar with the ins and the outs of this type of testing that could actually make

recommendations based on the information. That would take the public and the providers out of having to be the absolute experts on the information and allow them to be at the 10,000-foot level when trying to make an assessment with regard to the utility of this type of testing.

Finally, there was a substantial discussion about the definition of clinical utility and what all that means. I think there is a growing understanding that these tests may have value beyond the immediate clinical setting but extends into the individual's own perceptions and behaviors that isn't directly clinical. There was a feeling that this is almost certainly true but right now there aren't very good objective measures that can be used to determine the absolute value of this personal utility. Therefore, it is very hard to study and make recommendations about its magnitude of value in healthcare systems or society in general.

I would like to conclude just by saying that the slides from the meeting are all available. In your handout you should have this slide showing the .gov website. I think you will find a wealth of information

there. It really was quite a rich conference.

I would be happy to take questions, if that is permitted. I will try to answer them. Since I'm not Muin, it may not be possible.

**Question-and-Answer Session**

DR. EVANS: Greg, would you go into this a little more? I'm frustrated by this notion of personal utility.

My analogy with that is that many people in the U.S. would claim that their horoscope has personal utility. The problem with that concept of personal utility is that by its very nature it is a way to get around objective standards. While people may find horoscopes personally useful for a variety of reasons, I don't think in the absence of objective data it holds any water. I hate to see the discussion about personal genomics derailed and diverted by what I think is an intentionally obscured notion.

DR. FEERO: Obviously, I can't fully address your question. I would state that there are competent folks out there even in the academic realm that make arguments that if in fact even slightly erroneous

information results in an individual improving behavior and improving outlook on their health that that is of intrinsic value. I think that is an interesting and potentially perilous argument. I think the idea that you need to come up with some metrics to measure this will clean things out in the wash, if you will.

MS. AU: I think Marc is next.

DR. WILLIAMS: I'm a little bit concerned about the other end of the spectrum, which was the idea that the analytic validity is assured. This may represent ignorance of the actual testing on my part, but the information that was in our packet from PMC regarding the accuracy of the tests was saying that they are delivering the tests at a 99.9 percent accuracy. On the surface that seems good, but if you are doing a one million SNP, that is a thousand wrong calls.

Some of these relate to where you are aggregating 50 or 100 SNPs, and you could argue that maybe the incremental harm there is less, but some of the things that are incorporated into these relate to specific mutations in genes like BRCA and CF. If you make a wrong call there, then I think there is a very

different impact. I'm a little bit concerned that we may just say these things are valid and we don't need to worry about them.

DR. FEERO: I think that the meeting attendees would agree with you, but the focus of the meeting was really on the clinical validity issue because it looms in most folks' minds right at the moment, with these types of multiple-gene scans, higher on the profile of potential problems.

I don't know if there were other attendees at the meeting who are on the Committee. Feel free to also comment on that.

DR. FERREIRA-GONZALEZ: I understand what you are trying to say with the major need to look at the clinical validity of this, but we cannot forget the analytical validity. We have here the potential to maybe start developing the clinical validity, but we cannot disregard the analytical validity.

DR. FEERO: Correct. The point, though, is that let's say 99.9 percent of the time you are giving the correct genotype but only 15 percent of the time is that genotype actually reflective of actual risk. The

major problem doesn't lie in the analytic validity, it lies in the clinical validity. That was the major focus for the scientific discussion at this particular meeting. It wasn't the nuts and bolts of the CHPs.

MS. AU: We will take Kevin and then we will move on. There will be time for other questions after everyone has spoken.

DR. FITZGERALD: I wanted to just get a better sense of the personal utility. I understand, Greg, this wasn't your idea or anything like that, but you were there.

My concern is, as we look ahead and we are trying to figure out exactly how to take this landscape of personalized medicine and understand it in realistic even economic ways, it may be true that with the technologies and techniques we have now, there are certain people that could make Jim look like this if he so desires.

[Laughter.]

DR. FITZGERALD: I want to know, is that going to be considered health? This is the issue. If we are going to get personal utility merging with clinical

utility in any way, we are really going to be taking that landscape and making it extremely amorphous.

DR. FEERO: Obviously, that is a boundary issue that I think goes well beyond personal genome-wide scans. That is across the playing field of preconception counseling. Where are the boundaries.

MS. AU: I think Paul wants to speak.

DR. BILLINGS: While I may have a lot of ideas about the issue of personal utility, I will point out to this Committee that this is not an issue that is new to genetics. For instance, there was a long argument in genetics around the notion that any test that didn't have a specific treatment was not worth providing because there was no action to be taken upon it.

The determination of what that action was, was generally made by the provider, while patients, for instance, might have chosen to change their will as a personal response to the information that might have been contained in the genetic test.

DR. FEERO: I think that was articulated very well at the meeting with the Reveal Study with Alzheimer's.

DR. BILLINGS: Exactly. What I would just point out is that personal utility is an evolving concept. While I can understand some of our friends' objections to some of it, I don't think it is to be trashed altogether.

MS. AU: Thank you, Greg. Cathy reminded me that at the end, after all the speakers finish speaking, we will have them come back to the front and answer questions.

Our next speaker is on the telephone, actually. Christy White is the founder and principal of Cogent Research. They have a longitudinal study of American awareness, acceptance, and preferences for genomic-based benefits, products, and solutions. She is going to be presenting on some of their work today.

Your slides are up, Christy.

**Genomic Attitudes and Trends**

**Christy White**

[PowerPoint presentation.]

MS. WHITE: Thank you. I will just briefly talk a little bit about the study.

As was mentioned, it is a longitudinal study.

In this report we will be reporting on three years' worth of data. The goal of the study, on the Objectives slide, slide no. 3, is really for us to have this comprehensive, actionable assessment of where Americans' attitudes are and to monitor those over time.

Our goals are to look at awareness, attitudes, and preferences for using genetic information and to really understand what their views are. Are they similar or divergent. What are their views in general as it relates to both nutrigenomics and pharmacogenomics, or personalized medicine. We also look at that through a variety of different types of consumer models.

The objectives that we cover are on slide no. 4. There is a lot of data in this study. I have about 10 minutes and I'm going to focus on some of the critical issues specifically as they relate to DTC testing, but I have a couple of overview slides as well. There is a lot more in the research. If there are specific questions that the Committee has or there are things they would like to know, I would be more than happy to share specific pieces of this data with you. This just helps you understand more holistically what we cover.

The survey itself is about 120 questions. It takes about 15 minutes for consumers to do. We cover a lot of awareness, interest, and usage areas. Are they aware of the role of genes, are they aware of genomics in particular, are they interested in that. What specific health benefits are they looking for. We do actually delve into the whole issue that was being talked about earlier in terms of are they only interested if there is a specific benefit or treatment on the back end. Also, what have they actually done surrounding genetic testing.

We also look at perceptions and barriers. What do they think is good about genomics. What are they concerned about. We have a lot of information on discrimination. I know we have covered that in previous meetings. That continues to be an issue for consumers.

One of the things I won't cover today but can just tell you is there is very low awareness of GINA and no change, really, in consumer confidence that their information will not be used in a discriminatory fashion. I have that data and can share it with the Committee very easily if you are interested.

Then we get into more of the stuff we do on the

for-profit side around what do consumers want, who will they share with the information with, how do you best communicate with them. Then, as I mentioned, we do look at some policy-related information.

The methodology of this study is on slide no. 5. This is a representative sampling of the U.S. population. It is a Web-based survey and has been throughout its history. We are very careful in setting up quotas based upon U.S. census data to make sure that we get the right representation of age, income, ethnicity, region, and gender. We look at those numbers very carefully on the back end as well and, if necessary, do any weighting, which is usually minimal, to ensure that we can project this to the U.S. population.

We talked to a total of a thousand consumers. The sampling error for looking at this data is about plus or minus three. As I mentioned, we will be comparing this and looking at trending data to other years. We are looking at a sampling error of plus or minus four.

Slide no. 7. One of the first things we do in the survey is look at overall awareness. As you can see, awareness has basically been hovering around 75 percent.

Although we did see a statistically significant lift, it really isn't much in terms of total numbers. We started out with about 75 percent of the U.S. population saying they were aware of using genetic information to understand and optimize health. We don't actually ask them if they have heard of genomics, but we explain it to them in basic terms. You can see that that number at this point is at about 79 percent, which is a slight lift over what we have seen in previous years.

So they have heard of this general idea. We wanted to delve more deeply this year into direct-to-consumer testing and the availability of Web-based tests. In fact, we had talked with a couple of people at HHS. Scott Boyle and Greg Downing had given me some feedback on these questions when we were developing them.

They read a brief description of what we meant by personalized genetic profiles, which I will read to you.

Over the past year or so, a number of Web-based companies have started to offer personalized genetic profiles directly to individuals. These profiles are based on a DNA sample collected

using an in-home kit and provide you with information about your risk for approximately 30 diseases, such as arthritis, diabetes, and various cancers. Have you seen or heard anything about these personal genome services?

As you can see, about 12 percent of the population we surveyed said that they had in fact heard of some of these, which, frankly, was a bit higher than I had expected but still is only about one in 10.

We followed that up with a question asking what exactly do you think it means when these companies say they provide information about your risk. This was actually a multiple-response question because, as you know, it is not always the same. Interestingly, consumers chose pretty much only one response.

There is a lot of confusion. As you can see, there is very little agreement on exactly what it is that they would be getting for their money if they did choose to have such a test. About a third said that it would identify the chance of getting a specific disease, so that it would in fact give them some kind of a figure, like a 67 percent chance.

The next-greatest proportion said that it would tell them if they were at greater risk but it really wouldn't give any information about to what extent or exactly what the level of risk was.

Around one in five thought that it would just say that their genes look similar to those associated with the disease but not whether they had any increased risk level.

Only about 7 percent said it would determine whether they definitely will or will not get a specific disease. So only a few consumers are saying that it really cannot tell with any definitive answer whether they will get a disease or not.

Four percent said it would tell only if they already had a specific disease. Interestingly, only 8 percent weren't willing to wage a guess here in terms of what they thought it meant.

I think the key here is that consumers are willing to make an assessment of what they think they are getting, and what they think they are getting is really very variable.

On slide no. 9 we look at how interested people

are. We know that about one in 10 are aware specifically of DTC, but just in general we wanted to know how interested they were. You can see, again, it hovers around 50 percent. We haven't really seen much of a change over the past few years. Just about one in two consumers are saying that they are interested in using their genetic information for the purpose of understanding and optimizing their health.

We do see that there are specific subsets of the population that are disproportionately interested, and those are those with household incomes over $100,000 and those whose health profile has them on three or more prescriptions.

On slide no. 10 we look at what they actually want from these tests. Are they looking to just test for an individual condition or issue or do they want to know everything, all issues at once. You can see that there is a huge preference for that. Consumers are three times more likely to say that they want to test once and they want to get as much information as possible about what their genetic profile says about their health status.

One of the other interesting pieces of

information on this slide is the fact that you really only have about 20 percent of the population, and now 13 percent of the population, saying that they would never have a genetic test, they are not open to having a genetic test.

On slide no. 11, we actually asked consumers about very specific diseases and said what diseases would you be most interested in knowing about. I think one of the interesting things here is that when you roll up all the information and you look across all of the answers that Americans provide, actually 91 percent of them would want to test for at least one condition. So that 13 percent that said they would never have a test is probably really more like 9 percent. That is not too far off, but you do get a little bit more interest when consumers start to think about the specific things that they might be able to test for. So, large numbers of Americans are very interested and can think of something that they would want to test for.

You can see some of the things that they are most interested in. Cancer definitely shows up in the top 10 quite a bit. Also Alzheimer's, and of course

heart disease, not surprisingly, is right up there at the top.

On slide no. 12, one of the things that we noticed in this research this year is that consumers are feeling empowered. Across a lot of the questions that we asked we saw a lot more willingness to act on their own and not necessarily share the information with their doctor unless there was a problem, which we will talk about in a minute.

We have a question where we ask people would they actually involve their doctor in the decision of whether to have a test or not. We have seen a drop in that number. What we also see on this slide here is there is an additional drop in the number that are saying that they would share the information or they would want the results of that information to be shared with their doctor.

I think that obviously has a lot of implications, if you think about the fact that consumers are very interested in these tests. They can think of areas they would like to have the test. They don't necessarily what the information means when they get it,

and only one in two are saying that they would involve their doctor in the discussion of that information. This increased empowerment on the part of consumers is something that I think is really important for the Committee to keep in mind.

Slide no. 13. If they were to get the results and it were to indicate that they were at risk of a disease, now there is a slightly different story that emerges. You do see that the majority of people are saying yes, I would go and bring this information to my doctor or I would talk to my doctor about it.

We also wanted to look at some other actions. You can see about half are saying that they would want to see their physician more often to have some type of screening done. A little bit less than half are willing to make some lifestyle changes, either diet or exercise. I think that feeds into what we do know is an increasing belief on the part of Americans that diet and exercise are factors that can heavily influence their health status.

Only a third said that they would tell their family. We do know that consumers are very worried about

the emotional burden of having a test and they are not willing, as you can see, to share that burden with their families.

One in four are saying they would take prescription medication on a preventive basis. Thirteen percent are saying they would consider preventive surgery. Only about 4 percent say that they would not do anything as a result of that information.

Those are some of the highlights that I thought would be of most interest to the Committee. As I discussed, there is a lot of data and information in the study. I would be happy to talk with any of you individually or to provide information to the group as a whole if there is any other additional information that you think would be beneficial.

**Question-and-Answer Session**

MS. AU: Any comments or questions for Christy right now? Dr. Dale.

DR. DALE: I have first a comment and then a question. It looks to me like this panel you showed us about the difference in sharing information between '06 and '08 shows a general trend downward. I don't share it

with anybody. I interpret that as distrust.

The other comment that I would like you to respond to is, did you ask if people would want their samples saved for future discoveries or in some way get at the concept of a bank or storage?

MS. WHITE: We do actually cover that information in the study. I would have to look it up to be sure, and I know we are going to get back to questions later on so I will make sure I have that data. It was my understanding that that has also declined. Very few people want the information to be saved, but I will get those actual numbers for the later discussion.

DR. DALE: I'm thinking about saving the DNA.

MS. WHITE: Yes, absolutely. That question is covered.

MS. AU: I think we will move on, in the interest of time. Our next speaker is Larry Thompson. He is going to be telling us about the NIH Website for Consumer-Level Information about Direct-to-Consumer Genomic Services. Larry comes to us from the National Human Genome Research Institute, and he is the chief of the Communications and Public Liaison Branch.

## NIH Website for Consumer-Level Information About DTC Genomic Services

### Larry Thompson

MR. THOMPSON: Which may make you wonder, why is a communications guy up here talking about this? That is probably mostly because I have to do with websites.

Let me talk to you about three parts of this and give you a little bit of history of why NIH is moving towards trying to create a resource. We just did our own consumer research study as preparation for this so we wouldn't just completely make this up. Then let me tell you a little bit about what it is that we are thinking.

Of course, you all know that these direct-to-consumer tests started about two years ago. Out of that came some concerns by NIH leadership because they are outside of the medical model. These are complicated tests. The answers are not always particularly clear as to what they mean.

They were being marketed as entertainment or the new pet rock or something. People were worried that this would become viewed as genetic snake oil by the public so that when this stuff really did work people

would be skeptical about it.

Plus, we were hearing things like from one writer who has a book coming out. He was tested. One company told him that his heart disease risk was low, another said it was medium, another said it was high. That gives you a sense of how reliable this is.

We also learned of a physician in Philadelphia who was told his risk was really low, don't worry about a thing, but he of course had already had a major heart attack before the test was done. So the anecdotes were not reassuring and raised a lot of serious questions.

Dr. Zerhouni, back when he was the director of the institutes of NIH still, charged a bunch of IC directors with coming up with some plan to communicate to the public very authoritative stuff so that they would have a place where they could go when they wanted to understand that.

A trans-NIH committee was created. Dr. Guttmacher, who was the deputy director at NHGRI at the time and is now the acting director, and John Burklow, who is the associate director for NIH, were the co-chairs. Alan Stepped down when he took over as acting

director at Genome, and I replaced him.

We started moving very quickly to start making a bunch of sites and do things. We also started looking around in the world out there. It looked like we were creating much of the same information that was already out there, and so we began to wonder what we were doing. We ran out of momentum and started to slow down.

Then our friends at the Cancer Institute offered to actually do some market research for us. I'm a former journalist. We just go out and tell stories and make stuff up. Instead we thought we would actually do something different and get some information first, and so we decided to go ahead with this study, which was done last fall.

The report was just presented to the trans-NIH committee last week, so this is very good timing. I can tell you a little bit about what we found. It sounds very much consistent with what we just heard from Cogent, which is always encouraging, because ours was done as focus groups.

Let me tell you about the research and how that is affecting us. We did 10 focus groups in Chicago, New

York, and Washington. Eighty-four consumers participated. We also did in-depth interviews with nine physicians who were in primary care practice.

On the consumer side, demographically we had 61 percent women, 39 percent men. Not surprising, since women tend to focus on health more than guys. Seventy-seven percent were white, 18 percent were African American or black, 5 percent were other. Only 13 percent were ethnically Hispanic. I think we have to keep this in mind because of how this skews the population.

Also, this was a very educated group, which in some ways also skews it. All of them had high school diplomas. Many of them had been to college and a substantial number had college degrees. Half had children, so they were worried about inheritance if there were diseases running in the family.

We tried to stratify the consumers into three different groups: people who were not thinking about genetic testing at all, people who were thinking about doing it, and then people who did it. The last group we called doers, the ones who had actually had a genetic test.

We asked the recruiters to specifically go try to find people who had had direct-to-consumer tests like 23andMe or Navigenics, and they couldn't find any. Now, this is just a sample, and it is a very small sample, so it is not too surprising that we couldn't get any in who had done it. But they looked for them specifically, and that really made us all wonder. I don't know what to make of it. Again, it is a very small sample, but it was very interesting.

Let me tell you about the results from the consumers and then we will go to the doctors. Again, these are not quantitative. These are focus groups. We are trying to get impressions about what is going on.

Most consumers, at least in the focus groups, were broadly aware of genetic testing. That is probably why they agreed to be in them. They knew very little about the details of them, and when they were pressed for details they got stuff wrong all over the place. There really is not very deep knowledge among the public.

Many did not want to know their risk of getting certain diseases if there was no treatment or cure. If they couldn't do anything about it medically, they didn't

really care. Some said they did want to know, especially if they had a family history of a disease running in the family. They wanted to know if they were at risk themselves.

Most consumers were still very concerned about privacy and confidentiality. I'm not surprised to hear from Cogent that most people don't know about GINA. There is certainly a lot of work to be done about that. The consumers were particularly concerned about insurance companies and employers.

Most thought that a trained health professional should be involved in interpreting the test. They recognized that their own ability was not so good to really understand this stuff.

All the doers who had taken a genetic test had done so specifically because of a family history. They wanted to know what their risk was. Again, that is not too surprising.

In general, the consumers wanted us, the government, to provide lots of reliable, unbiased information. That is actually good news for the effort that we are looking at.

The results from the physician interviews were pretty interesting, not particularly surprising. Just to give you a little context on the practice setting for the docs, six were in small private practice, two in large private practice, one in a hospital practice, but they skewed older. I was a little disappointed at that when I saw their results. Two had practiced one to 10 years, two had practiced 11 to 20 years, and five had practiced 21 or more years. Genetics has changed a whole lot in that period of time and they didn't have a lot of that in medical school.

It is consistent with NHGRI's fundamental concern. When all this information starts pouring into the medical system that physicians are going to be deluged with it, we are worried about whether they will know what to do with it, frankly.

Again, these were interviews. The findings were that genetic testing really doesn't come up much in their practice. It just doesn't come up.

Few have had patients ask for help interpreting a genetic testing, including the DTCs. They are just not seeing it in their practice. The doctors really felt

that patients don't understand probability and really had no idea how to interpret the results of a genetic test.

The doctors also felt that patient information about genetic testing that we might be providing needs to be really practical and not technical at all. I guess I'm going to have to drop that wonderful graphic I made about how many angstroms there are in a single turn of DNA. We'll just forget that.

[Laughter.]

MR. THOMPSON: Many of the doctors said that they did not know enough about the kinds of genetic tests that were out there. They didn't have classes in medical school on it and, really, they wanted us, the government, to provide a list of approved tests. Of course, NIH is probably not likely to do that.

It certainly raises the question of vetting and endorsement issues and many other complicated things. They may be more appropriate roles for FDA or CMS or somebody like that, but I don't see us particularly doing that at this time.

The doctors were just generally skeptical about the value of genetic testing. They did feel, mostly,

that NIH should play an important role in providing information. There were some that thought we should just stay the heck out of it, that this is really an issue between the doctors and their patients and we should just be quiet. We will see how that goes.

Here is how we are not going to be quiet. Here is what we are thinking about doing. There were some recommendations that came out of the study, and then here are some ideas that we are developing right now to see how this could actually go.

The recommendations from the NCI study were that the information clearly had to be basic and practical, it had to be all about genetic testing, and it had to be very straightforward. We needed to develop it for different audiences. Certainly the public, but we really needed to be generating information for our professional audiences because they need a place that they can go for good stuff, too.

We needed to explain direct-to-consumer testing clearly. We should probably include genetic testing on the website, and we need to do basic, good standards for utility testing and stuff like that.

The assumption that we are going in with, or maybe I should say the assumption I'm going in with, since I'm charged with basically building this thing, is that consumers don't care. They are really disinterested in this subject, until they are interested. For the most part, we Americans are bombarded with messages, thousands of messages a day, and we filter them all out and ignore them until we get converted into information-seeking behavior. There are lots of studies about that around health information.

I think what we need to be doing is creating an authoritative, reliable, unbiased resource that people can go to when they get converted into that information-seeking mode. What we probably need to do is market the availability of that information when they want it.

If something comes up, like my kid gets sick or my parent is sick, or my sister, I want to know whether this is going to run in the family. I remember, "Oh yeah, those government guys, they have something out there that I can go find this."

The good thing about the way search engines are working these days is that government sites are

preferentially listed above commercial sites. We will bubble up to the top pretty quick, and people shouldn't have too much difficulty finding information that we put on the Web.

We are focusing on the Web because the people who are using this and seeking this information are very Web-savvy. Things are being marketed on the Web. This tends to be a more affluent group. We are not worried at this time, although we may get to that, about reaching further out into the world where people aren't using the Web and trying to reach those audiences as well.

The other thing that we are thinking about doing in this Web 2.0 world, which is overused and much hyped, is the social marketing of it all. We think this site needs to be engaging. The government, from my point of view, does lots of Web blogs. We create all kinds of content and put it on the Web. That is what a Web blogger does. They write something and put it on the Web.

What the government really doesn't do well is listen. We don't listen to the users and we don't want to take the time to try to sort it out and have a

conversation with our audience. We want to try to do that with this site. That is what we are thinking of doing.

We might want to take that even a step further. What I'm going to try to push, besides blogging this whole subsite, is to do a video blog on it. A video blog is basically just, instead of writing something, we bring somebody into a room, sit them down, do an interview with them, put a webcast up on the site, and the information becomes quickly available.

It is easy for us to do those. We can do that fairly quickly. My institute right now is trying to create a small interview studio so that we can test this idea and push this along.

It is easier in some ways for the audience to take this information in because all they have to do is sit there and watch TV, basically, on the Web. I have worked in broadcasting as a journalist. Television is automatically less dense. You just can't get as much information in television as you can in print. We will have to supplement with some text, but generally, it is a stream of consciousness way of getting information

across. It will be done in a Q&A kind of format.

There are challenges. We have to be 508-compliant. Closed captioning costs money. It has to be done quickly. We will definitely be working to put those resources in place. The other challenge, of course, will be finding experts across NIH, and wherever else we draw them in from, who can speak in a way that my mother can understand. She yells at me for not being understandable, but we will have to try to get there so that the information is accessible.

There are some other challenges. There is no budget for this. Like so many trans-NIH efforts, we are dependent on the kindness of colleagues. Right now people have been volunteering like crazy and it has been really great.

There is no dedicated staff for this. All the people that are working on this, including myself, are volunteers for it, and we are all hyper-busy, but there is a strong sense that this is important and it should be done.

This is a rapidly changing field, so we are going to need a group that monitors and keeps up as this

goes along. I am almost certain that I'm going to make mistakes as we are doing this, but I think that it will be an interesting exploratory process. If there is a conversation with our audience about it, I'm not as worried about making mistakes because we will talk about it. We will sort it out with that community of people who are interested in all of this. Overall, I'm optimistic that this will actually be helpful.

I will tell you one more thing in closing. An interesting note is, we were using a shorthand to refer to this and we were calling it Gene Scan. We were thinking about calling the site GeneScan.NIH.gov. We tested that when we had the consumers in the group, and they said, don't do that. They said it sounds like "scam." That was a New Yorker, so that is not too surprising.

[Laughter.]

MR. THOMPSON: The general sense was that this was something that was going to be cursory. It was not going to be in-depth and we would just gloss over it.

So we are still working on a name. If you have any good ideas, I'm all ears. I would be happy to take

questions.

MS. AU: I think Marc has a question or comment. Maybe Lyla can start moving up to the podium.

**Question-and-Answer Session**

DR. WILLIAMS: I like the idea of the videos. One thing that you might consider, given all the constraints that you previously mentioned, is that Dartmouth has published on shared medical decision-making using videos where you basically have patients relating stories to patients about a choice. I think the one that they studied most extensively was on benign prostate hypertrophy and the different interventions.

I think that this would be a great opportunity to have people tell stories about why they chose to be tested, why they chose not to be tested, why they chose to tell or not to tell their doctor.

I think, as you well know, being a journalist, we relate to stories much better than we relate to anything else. This might be a really cool opportunity to test how that would work in this setting.

MR. THOMPSON: We have been thinking about how do you have the dialogue on a government site and who do

you let in. You can't just let people post whatever they want to. It has to be vetted. There are some HHS policies already about that.

I do like the idea. I'm a little bit of a geek and I go on websites where there are technical discussions all the time, and people tell each other stuff all the time. I want to figure out how to enable that in this site as well. I think that is really important. Thank you.

MS. AU: We will have more time to ask Larry questions at the end. Our next speaker comes to us from the Institute of Medicine, where she is a senior program officer. She is going to be telling us about the plans for the National Academies Direct-To-Consumer Genetic Testing Workshop.

**Plans for the National Academies DTC Workshop**

**Lyla Hernandez, M.P.H.**

[PowerPoint presentation.]

DR. HERNANDEZ: You all know how important direct-to-consumer genetic testing is an issue. It is consuming a lot of our time and effort these days. Several different segments of the National Academies felt

it was important enough that, unlike when we are all trying to get our own projects going in our little areas, we thought it was very important to take an Academy-wide look at direct-to-consumer genetic testing.

Several of us got together, including the NAS Committee on Science, Technology, and Law, the National Academy of Science Board on Life Sciences, the Institute of Medicine Roundtable on Translating Genomics, the Drug Forum, the National Cancer Policy Forum, and we went to the presidents of the Academies and the Institute of Medicine and asked them for money to put together an Academy-wide workshop that would look at the kinds of issues that are of concern to various segments of the Academies in this whole area.

We have a Workshop Planning Committee that is composed of representatives that come from each of the segments of the Academies that is participating with the Genomics Roundtable, which is what I direct. We have Kathy Hudson and Muin Khoury, and I know you all know them. These are the rest of our members.

The goal of the project is actually to bring together numerous stakeholders -- something we all try to

do these days -- including scientific, medical, legal, and policy communities, and the public, to look at issues, opportunities, and challenges in this whole area.

We have four areas of emphasis. We are going to briefly try to get a handle on the current state of the knowledge and a future research trajectory in this area; shared genes and emerging issues in privacy, which you talked about this morning; the regulatory framework in DTC genetic testing; and then education, or communication and understanding I guess one would say, of the public and the medical community.

We were asking certain questions in the knowledge and research trajectory area, including the current status, of course. What do we know about the analytical validity and the clinical utility of these tests. Can we learn anything from these tests; if so, what. What will not be learned from these kinds of tests. What can we anticipate the future is going to look like in terms of the genetic tests that come online that will be available in the next five to 10 years. What is the market going to look like. Those are the kinds of questions we are exploring in the first session.

Our second session will look at shared genes and the emerging issues in privacy. One of the things that the planning group was particularly interested in is can we balance this consumer -- and now we know it is a small percentage of consumers -- desire to know with the need to protect and the need to guide. What are the risks and benefits for family members who use these tests; for public figures, if they choose to use them; for the legal system.

A big question is, who owns the individual's genomic data. There is the issue of discrimination and effectiveness of GINA. There is an emerging online social networking system that is based on these direct-to-consumer genetic testing results, and we want to explore that a bit.

There are many regulatory framework issues. I'm going to let you read the slide rather than reading it to you. Perhaps that will help speed us along so we aren't as far behind. I'm sure you have a copy.

A big area is what do we know about what the public knows and what the provider community knows, and what kind of providers are we talking about. Primary

care is very different than pediatrics, which is very different than obstetrics and gynecology in terms of the level of knowledge about certain kinds of genetic tests.

How do we ensure that those who take these DTC tests get proper interpretation. Are there mechanisms or innovative models that could be used to help that. What is the minimum knowledge required. What kind of lessons have we learned from other diagnostic tests and procedures.

We have not scheduled a date. We have had two planning committee conference calls. We hope to have another one in the near future and finalize the agenda, but we hope to hold the workshop in the late summer or early fall. You can contact either Anne-Marie Mazza or myself for more information. Thank you.

MS. AU: Do we have any questions or comments for Lyla?

[No response.]

MS. AU: Thank you, Lyla. Our next speaker is Amy Miller. She is the public policy director for the Personalized Medicine Coalition. She will be talking to us about Standards for Analytical Validity and Clinical

Validity of Genomic Scans.

**Standards for Analytical Validity and Clinical Validity of Genomic Scans**

**Amy Miller, Ph.D.**

[PowerPoint presentation.]

DR. MILLER: Thank you for inviting me to speak today. I would like to run through some Personalized Medicine Coalition efforts in this space.

First of all, who are we. We are interested in personalized medicine as a large concept in the future of health care. We represent all the different stakeholder groups in personalized medicine. That includes pharmaceutical companies, diagnostic companies, lab service companies, the academics who do the initial research, and the medical centers who put it into practice.

Here is a handy little diagram about we see ourselves. As you can see here, healthcare providers and patient groups are members of our organization.

You heard a little bit about the HHS, NIH, and CDC efforts in consumer genomics, and through those conversations there were some concerns that maybe the

results weren't similar when people got the three different scans. The companies, before this became a very public concern, hadn't really talked with each other.

During the HHS and SACGHS efforts over the summer of 2008, three gene scan companies: 23andMe; deCODE; and Navigenics; along with DNA Direct, came together and said it would be a good idea if we got together, talked about our products, and talked about how to get them a little more aligned.

DNA Direct, for those of you who don't know, is a longstanding direct-to-consumer genetic testing organization that does tests that usually you get through your physician. There is a physician who orders the tests, and the results are transmitted through a genetic counselor. DNA Direct has long been a member of the PMC and a leader in this field, and that is why Ryan Phelan in particular was involved in this conversation, but they don't do gene scans.

These three companies that do gene scans came together and said let's try to get our tests aligned so that when a journalist gets them all done they do get the

same results. Through that effort they came to adjust their algorithms in some ways so that the results are more similar. They also recognized that transparency would be very helpful to the community.

This is actually a link to the CDC's website, but it is also on the PMC webpage. This link, and what is in your book, is a four-page overview of the workgroup's efforts. The companies have recognized how important transparency is, and in the fourth page you will see links to the transparency pages of the three companies, where they go through how they calculate risk.

They have also pointed out some areas where it would be helpful to have the government say what would be useful. So, where is the consensus on how to calculate risk, or where is the consensus on when to include a SNP in results communication. These are some open questions that the companies themselves recognize.

Now, PMC is partly an educational organization, educating whomever about personalized medicine. Since these organizations have gotten so much attention publicly in the media, we thought it would be very useful if some organization came up with some educational

materials. To do that, we hired, frankly, Scott Boyle, who used to work at HHS and has since returned to academe, to help us write a consumer guide.

We also wanted patients and providers to have some input into this consumer guide, so we drafted a document and sent it to our Public Policy Committee at PMC. Some of you in this room actually took part in editing the guide there. We also sent it through our Science Committee. Some of you are also there. We shipped it around to some federal friends and received feedback there.

Then we sent it to the community and asked for feedback, and hosted a roundtable, where we asked patients and providers to read the document, to listen to companies present their products, and to give open and honest feedback about what kinds of information they want, how they would like it to be presented, what are some cautions they see in the products, and what are some benefits they see in the products.

PMC went into this event blindfolded. We didn't really have any expectations for outcomes. What was most surprising to me is that when we presented the

guide -- which is in your books, and for the rest of you is available in its entirety on this website -- the consumer groups represented in the room said we would like this guide to be redone for our needs. So I said, take it. If you want to take the content in this and expand on certain aspects and contract certain other aspects and remodel it for your own use, please do.

I was listening with rapt attention to the NIH gentleman who before me. There is a need for that. There is a need for an educational, government-wide effort. It should be focused on different kinds of groups as well. We heard it loud and clear from our consumer effort.

Now, in terms of going forward, as PMC received feedback on that very large guide we incorporated that feedback. The guide just grew and grew. We do hope to do a small educational brochure. We have some history of doing that before, and we hope to get one out soon. There is still, I think, a thirst for knowledge in this space.

MS. AU: Do we have any questions or comments for Amy?

[No response.]

MS. AU: Thank you, Amy.

DR. MILLER: Thank you.

MS. AU: Our next speaker is well known to the Committee because we keep inviting her back over and over again to give us great feedback. Anne Willey comes to us from the New York State Department of Health, where she is the director of the Office of Laboratory Policy and Planning. She is going to be telling us what is going on in that great State of New York.

**New York State Laboratory Requirements Relevant to Genomic Services**

**Anne Willey, Ph.D., J.D.**

[PowerPoint presentation.]

DR. WILLEY: Thanks for having me back again. I understand there are some new members of the Committee, and so very briefly I am going to just review the New York State oversight of clinical laboratories. I will emphasize again, as I have repeatedly before, this system operates for all laboratory testing in New York. It is not unique to genetics, but all genetic testing is subject to this system.

The statute in New York State preexists all federal statutes regarding oversight of clinical labs, having been passed in 1964. It requires all laboratories testing any specimen derived from the human body collected within the geographic jurisdiction of New York to have a permit from the New York State Department of Health, regardless of any other permit, regardless of any other accreditation.

The criteria for issuance of a permit requires that the lab director be qualified, that they submit an application and they pay us money, that the facility be inspected, that every assay they offer is either generally accepted -- that generally means FDA-cleared -- and approved by the New York State Department of Health, which means we have a rigorous review with assay validation, and they have to comply with any other state statutes.

Directors have to have a doctoral degree and four years post-doctoral experience. Two of those four years must be in the specialty, in this case genetics, and that experience must be within the last six years.

The lab submits an application in which we

review their ownership and financial interests, the physical facility layout and equipment, who is working in the lab, and what tests they intend to offer. Their initial fee is $1,100. It is then a percentage of their revenue. For some large major labs, this means they pay us over $1 million a year.

There is an on-site physical inspection of every facility. We go internationally to Hong Kong, the United Kingdom, and Iceland.

Every assay that they offer must be reviewed for its validity. That includes a specific assay description, a suitable guide that will be used by the person ordering the test, and an explanation of their consent process. New York State is a state that believes in genetic exceptionalism and has a specific statute in the civil rights law that explicitly requires written informed consent for all genetic tests. That is DNA, RNA, chromosomes, gene product, and/or product of gene product, for inherited traits. We are looking at germ-line mutation defined as genetic. It includes specifically DNA profiling.

We review analytical validity, and I will

generally agree with some comments made earlier that this is probably the easiest element for the laboratories to document. That doesn't mean we don't review it. We look at their actual data and their claims, their cutoff values and their error rates, and their precision, accuracy, and reproducibility, but it is their ability to detect and/or measure whatever that target is, be it the DNA sequence, the enzyme activity, whatever it is they are claiming.

We also review clinical validity, but this is generally documented by literature references. It is the documented association of the analytical target with some clinical condition or outcome or component of the biological specimen. New York State includes under its laboratory licensure program things beyond the CLIA definition of a clinical lab so that genetic profiling, paternity, forensics identity, and hobby genetics, if you will, are subject to oversight because it is a specimen and it is the measure of a component in that specimen.

We also review their reporting format. In genetics we require that that be in a format suitable for a non-geneticist.

Some of the other statutes become of issue, particularly when we are talking about the kind of direct-to-consumer marketing of genomic profiles. New York State is not a direct-access state. Individuals cannot order their own lab tests, with some very, very specific exceptions.

Therefore, every test, if it is performed by a permitted lab, is only performed at the request of a person authorized by law to make use of those test results. In the case of most genetic tests, that would be the clinician, generally a physician. Genetic counselors are not licensed healthcare practitioners and cannot order lab tests in New York State. It may be a lawyer in certain legal circumstances, such as paternity, identity, forensics.

Laboratories must report the results only to the person who orders the test, and they may communicate those results, which must be an exact copy of what was reported to the authorized person, to the patient or person tested only with written authorization of the ordering person.

We also have lots of business practice rules

for laboratories, including direct billing laws. Laboratories must bill the person tested or their insurance, with authorization. This to avoid middle men who mark up charges or add on services that may or may not be appropriately attached to the lab test.

There is a provider-to-provider exception between permitted labs. When a specimen goes off to one lab, that lab doesn't do the test, they refer it to another lab. The first lab can bill for it and pay the second lab.

Facilitators, intermediate marketers, and Internet facilitators cannot receive funds on behalf of a person tested to pay for the lab test. If they are arranging tests, which we have mentioned DNA Direct does, then the lab that does the test has to bill the person who is tested. DNA Direct can bill the person for the medical services they provide but they cannot be the pass-through for the money.

There are some very rigid anti-kickback statutes in New York State. There may be no fiscal or other incentives provided by a licensed laboratory or other entity to the ordering practitioner. You can't pay

them a fee, you can't employ them, you can't put them under contract, and perhaps more specifically, the laboratory cannot provide services to the person tested that would otherwise be provided by the practitioner.

Laboratories cannot provide genetic counseling for the persons they test. They can provide genetic counseling education to the physician who orders the test, and they can provide a copy of the test result if the physician authorizes them to do so, but the laboratory cannot practice medicine. Genetic counseling is considered the practice of medicine.

Under state education law, the license of a physician prohibits that physician from being an employee of a corporation. Corporations cannot practice medicine. Laboratories can't practice medicine, laboratories can't employ physicians who practice medicine, and physician groups have to be careful as to how they incorporate under New York State law.

Now, I'm asked how this works for the entities that are offering direct-to-consumer testing. I tried to be creative. I have learned a great deal. I can now draw arrows in PowerPoint.

[Laughter.]

DR. WILLEY: Education and information flows relatively freely. The one place we need to be careful is between the laboratory and the tested person. The tested person can provide information to the laboratory, but the laboratory can only communicate with the tested person in anything other than generic webpages or information or educational materials at the authorization of a physician.

There is an arrow missing on the slide between the laboratory and the authorized person or the physician. We want the labs to educate the practitioners about the tests that are available.

Within the different components of a laboratory, those who collect the specimen, those who perform the analysis, those who interpret the data, we expect appropriate exchanges of information.

There are these facilitators or marketing firms out there who can share information with physicians, share information with patients, and get information from the laboratory. That is another arrow missing from the slide. You will see it gets complicated enough.

We want a free education. We want free information, with one caution, that being between the lab and the person.

You will also note down here under the laboratory I have indicated three different components. We believe that it is consistent to say that these entities that will obtain raw data from the analytical testing facility and generate a report that would go to the ordering practitioner are laboratories. Making them laboratories creates the provider-to-provider exception regarding financial arrangements. It creates an appropriate provider-to-provider exception for exchange of patient information. It facilitates the kinds of activities that corporations like, if we will, the big four wish to engage in.

Making them laboratories does subject them to an inspection, the naming of a director, paying of a fee, and participating in whatever oversight and submission of data we require, but we believe it is also consistent with the CLIA requirement that says that the pathologist who receives the slides or the images from the analytical facility and issues an interpretive diagnosis on a Pap

smear must be licensed as a lab. We consider these data management facilities no different than that entity in pathology. So we are making these data management companies laboratories.

Information flows freely. There must be a written informed consent, and the statute specifies eight elements. Four of those elements can only be described by the lab: what test are you going to do, what is the predictive value of the test, what are you going to do with the specimen, and those kinds of things. The lab has to provide to the physician half of the information for the consent.

The physician is the only one who knows why they are doing the test, what it is going to mean for the patient, and they are the ones who have access to the signature of the patient. The actual execution of the consent, the turquoise line on the slide, occurs between the ordering physician and the patient.

The laboratory can get a copy of that consent. They are not required to have a copy. The physician who orders the test must retain the written informed consent.

Money. The tested person must pay the lab.

The tested person presumably pays the authorizing physician for their medical consult. The authorizing physician could pay a facilitator in exchange for information. That is that educational piece, that CME piece.

The laboratory could contract with that marketing entity for the distribution of educational materials. As between the components of the lab, they can exchange money. One entity gets all the money, they pay all the parts. The laboratory can give no money and no incentive to the authorizing physician.

The report is the white lines on the slide. The laboratory reports to the ordering physician. The ordering physician interprets and provides some results to the tested person. If the physician authorizes the laboratory to give a copy of that report to the patient, that can happen.

Adding in the two arrows I left out, when we try to explain the business practice criteria that we use to review these, we are looking at all of those various components in agreeing to approve one of these entities.

We monitor the Internet for marketers of lab

tests. Genetic tests are just one of the types of tests we monitor. We have sent to approximately 40 entities, since 2004, letters that say not in New York unless you have a permit.

I was asked to report on what the responses to those letters have been. I have copies of all the letters that went and copies of all the responses that came back. There are approximately 40 because the companies morph. They change from one into three and then they combine.

Anyway, we have had no response from eight. They tend to be small entities. They come and go on the Internet. There were eight that did not respond.

There were 12 that responded, we understand, we know you have rules, we won't do it in New York, and they put disclaimers on their websites that say not in New York.

We have five that said, we know you have rules, we think we are going to apply for a permit, but we won't take specimens from New York until we get our permit.

We have five that we still need to follow up. They are in that category. They do need a permit and we

need to get them into the system.

We have three that we have determined do not fall under our jurisdiction because you have to travel to that facility in order to have the specimen collected and that facility is not in New York. Therefore they are not in our jurisdiction, or they are not a laboratory. They are the practice of medicine, they are not performing any tests. That is three of them.

We have the biggies. Three have applied. One we have determined is not a lab. The remaining one is still in negotiations regarding the requirement for a physician's order and whether there are any options under the New York State statute.

I would be happy to take questions.

MS. AU: While we are asking Anne questions, if I can have the other speakers start moving to the front so we can do the panel. Yes, Jim.

**Question-and-Answer Session**

DR. EVANS: I will ask the obvious question, Anne. You left us with the three biggies and you had determined that one was not a lab.

DR. WILLEY: DNA Direct is the practice of

medical genetics. They facilitate the testing, but they do not do any testing. They have accommodated the New York State direct billing law. The Department of Education has cautioned them regarding the corporate structure under which the New York-licensed physicians provide the medical services, but that is not a laboratory issue.

DR. EVANS: Where do things stand with the large labs like 23andMe in getting this? At least one of them says, we have a physician that orders the tests, but that would seem to be in conflict with your rules.

DR. WILLEY: It is.

DR. EVANS: So they would not be eligible to do this on specimens collected in New York.

DR. WILLEY: Not if there is any financial arrangement with that physician.

DR. WILLEY: Julio.

DR. LICINIO: My question was, I have been reading about how people have these DNA parties where everybody goes and collects samples.

DR. WILLEY: Those specimens were destroyed.

DR. LICINIO: Yes, but let's say I am not a

resident of New York and I go to such a party, and the test is sent outside of New York. So I don't reside in New York, the test does not happen in New York, but I happen to be in New York for the collection, is that legal or illegal to you?

DR. WILLEY: If the specimen is collected in the geographic boundaries of New York State, then the laboratory that performs the test is subject to the jurisdiction of the State of New York. It is not that far to Connecticut.

MS. AU: We won't tell the governor, Anne. Any other questions for Anne right now?

DR. WILLEY: The answer to your question is no, no labs are approved in New York State to offer whole genome scans. Some of you may know that in the last two weeks we have approved three laboratories to do array-based genome scans, but those are for specific genetic conditions which are confirmed by cytogenetic fish.

MS. AU: Why don't we have all the speakers come up to the front. Do any of the Committee have questions or comments for any of the speakers today? Jim, do you have a question?

DR. EVANS: This would really be for all of you. As I was listening, one thing that I was struck by was a fair amount of discussion about analytical validity and a fair amount of discussion about clinical validity. I think, as a practitioner and as a patient, is that what is most important is what those two concepts are subservient to ultimately, which is clinical utility. I'm just wondering what your thoughts are about clinical utility because I didn't hear much about that.

Anne, you are the only one who I think was clear on that. It doesn't fall under your jurisdiction, really.

DR. WILLEY: To make it clear, if a laboratory includes in their report something which verges on claims or patient-specific recommendations. It's one thing to have educational material on the website that says if you have this test and we find these markers, people with those markers may have these increased risks. That is educational material.

After the test has been done and you are saying to the patient, "You have these markers. These markers are found in individuals at increased risk of," the

laboratory cannot then say, "Therefore you should take this drug or have this test." Laboratories can't do that. The utility, what you do with this information, is left to the practitioner who ordered the test.

**Committee Discussion of Issues and Next Steps**

DR. EVANS: I'm interested in where that concept falls for the rest of you.

DR. FEERO: I will first comment from the standpoint of the meeting that I talked about. I think that utility was definitely part of the discussion at the meeting. It is obviously a very difficult thing to define. It is very, very hard to define. It is quite hard to measure. It takes a lot of time and effort.

I think a lot of the meeting actually focused on the need for adequate clinical validity before you can get to really addressing in big studies the clinical utility issue. If the SNPs aren't predictive of risk in all the populations you want to include in a large utility study, you can't do the study.

As anybody knows who has heard me speak before, utility is near and dear to my heart as an issue. I think you cannot neglect that lens for these

applications.

DR. MILLER: I was about to answer very similarly. Just to add on to that, because clinical utility is so hard to define one unintended consequence of these companies coming forward is that consumers know a whole lot more now about what genes mean to their health. I think they are also starting to learn a bit more about probability. That is an unintended but perhaps positive consequence. It is adding to what consumers understand.

DR. EVANS: I'm actually skeptical that there is an increased understanding of any real appreciation for probability and utility.

DR. MILLER: I don't have any data to back up what I said.

DR. EVANS: Right. That is my next question. I don't think there are data to suggest that.

DR. FEERO: I would say that a definite benefit has been an increase in the dialogue and also the sense of urgency to address the issue. These companies I think have done a service in that respect to propel the discussions that need to happen as these technologies are

becoming more and more viable for healthcare applications.

MS. AU: Christy, are you still on the phone?

MS. WHITE: I'm here.

MS. AU: Do you have that information for Dr. Dale?

MS. WHITE: I do. I know there was some discussion with the last speaker about that in terms of the ability for people to retain information.

The way it was worded actually is, "What should happen to your DNA sample after the test is complete?" and 46 percent said, "Retain the DNA sample for future tests of my choosing." When we asked them who they would want to keep the DNA, the vast majority of them, two-thirds, said that they would want it to be kept by the company that conducted the test. Very few said a private medical storage company. Less than one in 10 said that a government agency should have that information. No offense to anyone in the room.

MS. AU: Andrea.

DR. FERREIRA-GONZALEZ: Did you also ask them about not only retaining the specimen but if we can use

it for further testing or for other purposes?

MS. WHITE: We did have another attitudinal question at some point that didn't ask them if they would want it but were they concerned that that would happen. I think something like two-thirds of people said they were very concerned that their test may be used without their permission. While we didn't ask that exact question, from a lot of the qualitative research we have done I would say absolutely they do not want that information to be used except by their own choosing and for a specific test that they would indicate.

MS. AU: Gwen.

MS. DARIEN: Hi, Christy. It is Gwen Darien. I have a question. You asked it one way, but one of my colleagues, who is an OB/GYN and bioethicist, did a survey and asked the question in a different way. The question was how people would feel about having their embryos used for research if it would help forward medicine.

Overwhelmingly, the families that were asked said that they would be happy to have their embryos used for research and that they weren't using their discarded

embryos.

It seems to me, that the way the question was posed would lead people to answer the way that you answered it. In my mind, there would be some suspicion in the way the question was posed.

MS. WHITE: Right. Obviously, if you are giving people an altruistic reason to use the DNA you might see a different response. In this case it was really more the likely scenario, which is I have had a genetic test for my own purposes, I have had my DNA taken to tell me about a specific test I want, and I'm housing my DNA there for my own purposes in the future.

Certainly, if it is more mom and apple pie and it is served up in an altruistic manner, particularly among women as it relates to children or disease prevention in the future, I would imagine you would see an inflated response. Absolutely, the context is critical.

MS. DARIEN: I don't even think it is inflated. I think it is just flipped.

MS. WHITE: I don't mean erroneously inflated. I mean truly. Certainly you would have people

responding differently depending upon what you were going to do with it.

Actually, in '06 we asked a couple of questions about consumers' willingness to be part of a larger database that the government would have for very similar purposes, more for the greater good of the American public. We did see that there was definitely interest for consumers, but it wasn't as widespread as we would like to see, potentially.

MS. DARIEN: Was this done before or after the passage of GINA?

MS. WHITE: It was done a month after, which I found very interesting. If there had been any publicity, or to the extent to which there was media coverage about it, it was probably happening right around or, frankly, right before a flurry of communication, if you could call it that, about the passage. We probably were in the field where we would have expected to see the highest levels of awareness, and we basically saw absolutely no lift in awareness of protections from '06 to '08.

MS. AU: Paul.

DR. BILLINGS: I would like to ask for a couple

of points of clarification about the New York State situation, which is complicated for my untutored mind. For instance, several of the national labs, who I believe practice in New York State, employ genetic counselors. From what I think you said about the relationship between labs and counselors, does that mean that for samples collected in New York State the labs have not been using those counselors as part of the process?

DR. WILLEY: No, those counselors either provide education to the ordering physician or provide guidance to the ordering physician in interpreting the results.

DR. BILLINGS: They don't provide services direct to the consumer?

DR. WILLEY: With the written authorization of the ordering physician they can provide the service, which would repeat the result and explain what it means. By our criteria, that is probably not genetic counseling in its fullest extent.

Now, are those genetic counselors talking to patients who are tested in New York? Yes.

DR. BILLINGS: Yes, I know they are.

DR. WILLEY: But they are not supposed to be providing genetic counseling.

DR. BILLINGS: Second of all, as I understand your diagram, the result of a lab test cannot be provided to the patient directly.

DR. WILLEY: No, with the written authorization of the physician it can.

DR. BILLINGS: Right. So, if a doctor orders a test and then goes out of town or on vacation and the person is waiting for their cancer test result, they have to wait until the doctor comes back?

DR. WILLEY: I believe it would be considered negligent medical practice if the physician did not make arrangements for that.

DR. BILLINGS: This leads to my question, then. It is a remarkably intricate and important regulatory network that you have set up. From New York's point of view, what is working well and what needs reform?

DR. WILLEY: From New York's point of view, to the extent that laboratories apply for permits, have their assays reviewed, get permission to offer the assay because its analytical validity and clinical validity

have been documented to our satisfaction and we are happy -- and we look to other national organizations for what criteria should be used -- and we generate a list of not only the approved labs but the approved tests, that works well.

We also do have a mechanism by which a physician can make a request to use a lab that is not permitted for a particular patient for a particular clinical need, and we have never said no, so long as it is unique to that patient and a justifiable medical need. So you can use labs that don't have permits and you can use permitted labs that aren't approved to do a particular test if the clinician feels that is necessary.

That system works. What doesn't work, from our perspective, is that a patient can go to Connecticut and get the test. Unfortunately, that is true, and it argues that we are providing overkill.

Our program costs us $20 million to run. We do regulate 1,600 labs. We believe we regulate over 75 percent of all the genetic testing done in the country because all of the major labs are New York State-licensed. The courts look with great disfavor when it

turns out the lab did not meet New York standards on a specimen from Connecticut because, after all, New York standards are more stringent and more rigorous than CLIA.

For the residents of New York State, our system is working. For New York State residents who choose to avoid the system, there may be problems. I do believe there is really a problem for the rest of the country.

Just relevant to retention of specimens, because it has come up in terms of the genome profiles, New York State civil rights law requires a specimen be destroyed at 60 days unless the tested individual explicitly consents to its retention. It can be retained deidentified for unspecified research. If it is retained in an identified format or used for any genetics research, it must be an explicit genetics research consent.

The issue regarding genome scans has come up. What about the data? It is more efficient to run the full genome SNP profile using however many you can do at once. You have the DNA. You can get all the data now. You don't need to keep the specimen. That data is not yet clinically valid because we don't know what it means.

Can we keep the data and mine the data later? We have said yes, if the new analytical purpose of mining the data has been validated and if the patient's physician explicitly orders the new test. It gets very complicated.

DR. BILLINGS: It seems insurmountable.

MS. AU: I have Dr. Dale, Kevin, and Mike, and then I think we need to move on.

DR. DALE: Go ahead.

MS. AU: Go ahead, Kevin.

DR. FITZGERALD: Of course, the questions are always too brief. Getting back to the personal utility issue, which I don't want to become too confused, obviously, one would hope, anything involving health care would have personal utility. My question is going to be, how are we going to try to put parameters around what we are doing and to what end. So, where does clinical utility come in as a bottom line, or is it the bottom line? If it isn't the bottom line, what kind of utility will be?

There is not only the possibility of personal utility, there is also public utility. If we are

collecting this data and we are putting it in public databases, obviously government institutions can come in and claim the utility on their own to pursue their own ends.

DR. FEERO: I will try to tackle that. I think it depends a lot on what the desired end product is. I would think if you were a payer for health insurance, clinical utility would be largely what you were thinking about. If you were a regulatory authority trying to decide whether you should be able to offer these tests, period, you would probably have to look at some sort of aggregate measure of its overall worth rather than simply saying clinical utility.

Let's just say some state decided to say no, you can't offer genome-wide scans. To make that decision I would think they would have to look not only at clinical utility but at personal utility or some other, more nebulous measure of whether or not for an individual consumer this has value beyond the way the doctor, the P.A., or the nurse practitioner is going to use the information in a clinical setting.

I think it very much matters in what window.

To me, it would make sense to explore moving to a broader definition and a very narrow view of clinical utility for the majority of these discussions when we are talking about it from a societal perspective.

DR. MILLER: I think some individuals would argue that they can themselves decide if there is some utility. Some people without a family health history, for example, may find they have a personal utility for this information that otherwise may not be.

DR. FITZGERALD: Right. I guess that then gets back to what we see as the ultimate utility of this information. Is this just another commodity for people to buy, like a car, or is this in some way different because it has to do with health care. Again, it is this intersection of things. That is why I'm curious to see where you see things going and where you see the line.

DR. FEERO: I would tell you to look around at other healthcare applications for models of what you can access and what you can't access. Don't use a genetic exceptionalist perspective on this. You can go out and buy a lot of things that don't make a lot of sense in our healthcare system right now.

I think a big question that all of us should be asking is, is genetics so different that we should be holding it to a higher standard. I would argue that we should at least entertain that because its applications are so broad and potentially costly to healthcare systems.

MS. AU: I think Mike and Jim are dying to jump in on this.

DR. AMOS: As far as the process, the next part of the agenda is to get into next steps and action items. Considering the fact that our panelists have thought about this a lot, before they go sit down and we lose them would it be appropriate to ask you what you think we should recommend to the Secretary as to what the next steps should be with regard to direct-to-consumer testing?

We might be learning something from the research that is being done by these companies, but maybe not. I'm still unclear. Is there the potential for things to be learned, or would we be throwing the baby out with the bath water if we shut everything down?

DR. TEUTSCH: Let me recast that. You can

advise us on things that we might want to take up rather than specific recommendations. What are the areas that we should be looking at that would add to the utility for the Secretary?

DR. MILLER: When PMC was doing our work, we just had the same conversation time and again. This is early. We are talking about SNP technology and CHIP technology. Soon, meaning five years from now at the most, the technology is going to be completely different. There is a baby-and-bath-water issue. There is also a horse-out-of-the-barn issue, and I'm sure I could come up with some more picturesque speech if I thought about it. So I would suggest that this Committee look forward no matter what you do.

MS. AU: Jim.

DR. EVANS: I just wanted to try to put in perspective this issue of utility. I think that one of the things that we all have to recognize is that robust genomic analysis is definitely going to exist, probably predominantly outside of the traditional medical model and outside of the Academy. Therefore, I think when we get to issues of utility, Mike's admonition -- or maybe,

Paul, it was your comment -- about personal utility perhaps having some merit is well taken.

I think what we have to do in that context is reconcile claims that are made with utility. In other words, if laboratories are going to, either de facto or explicitly, make medical claims, then they have to be held to traditional models of clinical utility. If they choose to market their products as entertainment or as hobbies, fine. Then people are free to interpret their own personal utility, but they then cannot make medical claims.

I think what is really important is that we have some reconciliation between the claims that are made and what is actually being offered.

DR. MILLER: Greg could probably answer this even better than I can, but I will take a stab at it. At the CDC-NIH event, one of the roundtable participants said the big three -- 23andMe, Navigenics, and deCODE Genetics -- are talking to federal regulators, SACGHS, and federal researchers and regulators, and there are some companies who aren't. So I think these companies are cautious about making medical claims.

DR. EVANS: Actually, they are making medical claims. I think that is obvious in their websites and their advertising. That is where I think we need some reconciliation.

DR. FEERO: I think that is the real challenge, the explicit versus the implicit claim of clinical usefulness. I don't have a solid sense as to how you can deal with that in the current environment beyond being fairly draconian about what SNPs you are using.

MR. THOMPSON: Can I just respond to that really briefly? I think that the answer ultimately is that however you define the policy side of clinical utility, it is really wise to keep a close eye on the science side of it. NIH sponsored a conference about a month and a half ago called the Dark Matter of the Genome. Basically, we were trying to figure out where all the inheritance is. There is all this SNP stuff being done and these genome-wide studies being done, and we are not seeing the amount of inheritance that would be expected.

There are a lot of unanswered questions out there. For companies to be making claims about anything,

it is making the people around me go, "What the?" I think that is an important, ground-based reality question. Stay close to the science.

DR. FEERO: I would like to go to the question about what some of the next steps are. I think that one of the things that this Committee could help to do is to focus HHS's attention on the need for a very considered and thoughtful approach to the issue of translational research in this area.

I think that it is clear that the prime mission of most of the research is in the early discovery phase. That is probably very justified. It is exceedingly justified. Just as we had a focus on ELSI early on in this topic area, I think we are moving to a stage where maybe there should be an increased emphasis, similar to ELSI, on making sure that the movement to clinical application is done in a careful and considered way.

DR. BILLINGS: I just wanted to point out, to Jim's comment, that blood groups have been measured and have an important clinical utility in transfusion and transplantation. Yet there are cultures that use blood group information for all sorts of things.

DR. EVANS: That doesn't mean that they are correct.

DR. BILLINGS: They are what they are.

DR. EVANS: What I'm saying is we should not be in the business of promulgating myths.

DR. WILLIAMS: I wanted to respond to Jim's point. I'm not sure that I actually heard him right, but this was also true in the information from PMC, if I'm not mistaken. It seems to me that there is an attempt to create an island of sorts by using terms like "informational." In other words, there is recreational testing, there is medical testing, and then there is informational testing, which seems to relate to some of this issue about personal utility.

I recognize that some of this reflects the rugged individualism of the American people, but I would be reluctant to let the company define where it wants to sit. I think we would then be in the same sort of situation we are currently in with nutriceuticals and alternative medicine, which is if you claim "I'm nutritional and I'm not a drug," you are exempted from a tremendous amount of regulation. Yet we have very good

examples that in fact the harm may be quite more substantial than what we have in the pharmaceutical industry.

I think we have to be cautious about creating safe harbors by using some of the language imprecisely.

DR. TEUTSCH: Let me thank all the panelists. You have obviously sparked an interesting discussion that we need to grapple with. So, many thanks. Chances are, we will get back to you.

MS. AU: Thank you. Thank you, Christy.

[Applause.]

DR. TEUTSCH: Having heard all of this, do you have some suggestions for how we proceed?

**Proposal for Short-Term Action**

**Sylvia Au, M.S., CGC**

[PowerPoint presentation.]

MS. AU: The next section is going to be a proposal for short-term action for the Committee. The proposal for the short-term action is that we develop a brief document that reviews the concerns about direct-to-consumer testing, such as limited data on clinical validity and utility of tests, consumer and provider

understanding of test results, privacy protection, companies that skirt oversight regulations, and false and misleading claims.

The reason we picked those right now is because we have recommendations from SACGHS on them. Instead of making new recommendations, this would be taking recommendations we already have to address these issues and then recommending other action steps for maybe a more in-depth report or other action. Keep in mind this is a short-term action step.

When we went through the recommendations, which all Committee members should have memorized and tattooed on your body -- new members should have that done as soon as possible -- we found that there were two recommendations that would deal with the clinical validity and utility data recommendation, three recommendations that dealt with consumer and provider education, one recommendation that dealt with privacy protection, and one recommendation that dealt with false and misleading claims.

I'm not going to read all these recommendations to you, but as I was reading them again, I realized we

are a very wordy bunch.

The first recommendation that Cathy and I think has to do with some of these direct-to-consumer issues is the FDA evaluation of lab tests. I'm sure our FDA colleague is very happy to hear that we are bringing that up again, since they were so happy to hear that the last time.

Continuing on with the clinical validity recommendation, we have recommendations for creating the public-private workgroup, developing criteria for risk stratification and how to apply the criteria, and also that lovely mandatory test registry.

Following that, we have another recommendation about a public-private group of stakeholders to assess clinical utility, which we have been discussing today. That is a very long recommendation that goes on for three slides.

We also have recommendations on funding clinical utility research and how to disseminate that information to the public so they can use it.

Education recommendations that we have are that public and private entities should address knowledge

deficiencies and the need to train and educate healthcare providers with appropriate funding, resources, et cetera. That recommendation continues with having additional funding for education and training.

We also have a recommendation that education resources are made available on websites to help consumers make informed decisions about their health care.

We had that regulation that CMS loves about CLIA oversight and privacy protections. Then we have the regulation, again, that we had put up to address false and misleading claims and to regulate marketing of direct-to-consumer genetic testing.

Those were the seven recommendations that Cathy and I could come up with. Of course, there could be other ones that we could come up with. All of them are actually at the back of the progress report that is included in your briefing book if you want to start memorizing them now.

Our next step, if the Committee decides that we want to take this action step, is to form a small short-term task force -- "short-term" meaning less than three

years long -- to develop a really fast report. This area seems to be in the news a lot, so we can highlight some of these existing recommendations that we have had for so long. Then we can also have the short-term task force look at what issues have not been addressed by our prior recommendations, and what further work might need to be done.

**Committee Discussion**

DR. TEUTSCH: Great. Andrea, did you want to comment?

DR. FERREIRA-GONZALEZ: The idea is that we will develop a brief report where we are specifically addressing direct-to-consumer issues and then pulling from the previous reports' issues. So we will be highlighting that we are concerned about direct-to-consumer testing.

MS. AU: Yes. Then we can also put what issues we need further study on, because we are not going to do this in-depth four-year report that we do all the time.

DR. FERREIRA-GONZALEZ: I think I like the idea. I think it needs to be separately addressed, even though we have addressed it in other reports.

DR. TEUTSCH: I would be curious about whether we are monitoring the relative success of these enterprises. The fact that they get a lot of coverage in the media doesn't indicate that they are necessarily flying off the shelf in terms of their popularity. I wonder whether that data might frame some of the issues or the amount of money being spent.

One of the things we saw in this panel is that here in Washington a lot of money is being spent on DTC genetic testing. I'm not sure it deserves it.

MS. AU: I think that is one of the issues the small, short-term task force needs to look at, whether it is actually happening. I don't know what we can do to evaluate that unless they give us their financial information, which would be interesting.

DR. TEUTSCH: We had some information today when we heard that someone conducted a survey of a thousand people and apparently zero, or close to it, had used the testing.

DR. FERREIRA-GONZALEZ: As these start showing up in these magazines, and with our esteemed colleague representing us, I would expect that to rise.

DR. AMOS: I just think that Amy's recommendation for looking forward is really critical. At NIST we have looked at the GWAS studies and we have made a decision not to worry about standards for this because we don't think that the technology is going to last that long. We are the government. It takes us a while to do anything. In four or five years the technology is going to be sequencing.

Maybe the kits are not flying off the shelf right now, but when it is possible for $1,000 to get your entire genome sequenced, a lot of people are going to go after that.

MS. ASPINALL: I would actually agree with Mike. I think the relative financial or business performance after a certain hurdle, if these are relevant and being talked about, is not a key issue. We could spend a lot of time saying what is successful and what isn't successful. I think it is a broader policy issue. We will deal with it a little bit in the Futures Panel tomorrow, but it needs to be something that, from a policy point of view, we think has the potential of being relevant and therefore is high-priority, not literally

what is happening today.

DR. LICINIO: I think that, actually, the current economic situation, if anything, is going to pressure the companies to make these products cheaper. 23andMe went from close to $1,000 to $399 a few months ago. The cost of doing this for them decreases, and then, because of the financial pressure, they are probably going to lower the cost, which may increase the outreach. I think that we really have to continue to do this.

DR. TELFAIR: I heard this earlier but I wanted to echo it so it doesn't get lost in the morass. It is going to be very, very critical to have some kind of strong recommendation for monitoring and assessment, whatever else we come up with. We should consider that, particularly around this. If we are going to put forward the policy issues, we also need to consider what is going to be the mechanism to be able to do that. That is going to be very critical in the long term.

DR. FROSST: I will start by widely agreeing with Amy that the technology is going to be changing very rapidly. I think by the time we really fully understand

what we think about this issue we are going to be looking at sequencing rather than a scan.

Then I'm going to agree with Paul and say that I think the volume of tests right now is small. I think the amount of people that are signing up to do 23andMe or Navigenics is small. If you look at it from a public health perspective, does it merit all our time? Probably not.

I think that if we consider the implications of DTC for a gene scan versus the implications of DTC for a whole genome scan, the main issues that we are going to look at are very comparable. It is the broader issue of people buying or getting information for which the validity and utility are unknown and rapidly changing that makes it an important point for us to look at.

DR. TEUTSCH: Barbara and then Marc.

DR. McGRATH: I was just going to say what you said, so I will just second that. I think the price is going down, but still, even at $1,000 or $400 in these economic times, a certain segment of the population is going to do it. As we think about the public health of the nation, we should be cognizant of who we are talking

about. If we look at the larger issue of not specifically the people who are having the DTC tests but some of the principles about it, then I think it makes good sense.

DR. TEUTSCH: Marc and then David.

DR. WILLIAMS: This also relates to the issue of sequencing and cost. I think the point that is going to be different is that the price point is not going to affect consumer uptake. The price point is going to affect the purchasers of services, like the government and the payers. In other words, if payers can get the whole genome at $1,000, they are not going to pay somebody else $4,000 to get one gene.

I think it could completely change the paradigm. Then the push is going to be very different because we are going to have much more information than what was specifically asked for. I think it will be a changing paradigm, but a lot of the same issues relating to validity and utility will still attend.

The small point I wanted to make was just to emphasize something that I heard in the Cogent presentation. Actually, they were all cogent

presentations, but specifically the named Cogent presentation.

Physicians want a repository. Actually, the physicians want a Good Housekeeping Seal of Approval, which the government may not in fact be able to provide. I think nine out of nine said, we want a registry where we can go and see these things. I think that is a strong external endorsement for what this Committee felt very strongly about relating to having a centralized repository for genetic testing. I would definitely want to move that up the prioritization.

DR. TEUTSCH: David and Mara. Then Robinsue. David, go ahead.

DR. DALE: I was just going to comment that I appreciate Jim being willing to speak up about unsubstantiated claims. On the other hand, the technology has a real promise in terms of its medical application. We need to push the research agenda to define where that application is most appropriate.

DR. TEUTSCH: Mara.

MS. ASPINALL: I have two things. One piece is, I very much agree with Phyllis's comment. I think in

general we have to be technology agnostic because we cannot anticipate what technologies are and deal with the information.

I guess, Sylvia, I go back to the comment about the time frame and whether it is one year or three years or four years. My concern on putting a priority on this is when will the regulation likely be promulgated? If it is a result of perceived or actual risk, there is going to be a lot of activity on putting regulations on this. That happens in the next year. Our report that takes three years will not be relevant.

I think the prioritization in terms of timing is our key issue. Coordinating with other bodies that may be taking actions during this period of time is the most important piece to ensure what we do is actually relevant and helps the argument.

DR. TEUTSCH: I think we are talking about something fairly short-term here, too. Robinsue, and then I would like to see if I can pull some of this together.

DR. FROHBOESE: Thanks. I just wanted to add a very brief and technical point. To the extent that this

document is going to be reviewing main concerns, on slide no. 2 one of the concerns listed is privacy protections. I think it is going to be very important to ensure that we are distinguishing between is this an inadequacy with current privacy protections or is it, as I heard the reports coming in, a lack of awareness or perhaps misunderstanding of protections that already exist.

I just want to make that point because you will see in the next session, when we get to research and the HIPAA Privacy Rule, that that is another issue that we are going to be raising.

DR. TEUTSCH: Let me see if I can pull this together a little bit. The initial proposal was that we look at our current recommendations and put together a short report that could be looked at probably at our June meeting and then promulgated.

I also heard some core issues being raised here of things that are beyond what we have done, particularly the discussion of clinical utility, as well as personal or public utility, and how that should inform our discussion. That seems to me to be a large and rather core issue and certainly a lightning rod for our

discussion today.

I heard some issues on translational research -- some of which I think were embodied in the clinical utility recommendation -- for privacy, equity, and how should these technologies go on being monitored.

I heard we should probably be technology agnostic at this point because we can never get ahead of that curve.

What I would suggest is that we get a small group together to focus on the short term and give us something to look at in June. They will look at our recommendations and also tell us which of this constellation of other things really rise to the level of things that we should address in what time frame and in what way.

DR. AMOS: I just had one other suggestion. Writing and thinking about this should be fluid. Maybe you could almost put in acceptance gates for the future, to the point where you need a great deal of restriction until the clinical utility and analytical validity is understood. Then maybe you need additional restriction until the standards are in place for the technology

utilization.

DR. TEUTSCH: Looking at the overall process of dissemination.

DR. AMOS: Yes.

DR. TEUTSCH: Mara?

MS. ASPINALL: I would agree with your recommendation, with one addition. That is, understand what the other relevant bodies might be doing. I think that would be a key piece to include in the June report so we are not overlapping with what other groups are doing.

DR. TEUTSCH: Does that seem like a reasonable proposal, as amended? Is there anybody who disagrees and wants us to do something different? If not, could I get some volunteers who will work with our dear colleague Sylvia Au?

MS. AU: All the people that I have helped.

[Laughter.]

DR. TEUTSCH: I have Jim Evans, David Dale, Julio Licinio, and Andrea. I think that is great. Others who want to, you can let the staff know.

DR. WILLIAMS: I would think Sarah, if she is

not on the list.

DR. TEUTSCH: I think that is a terrific suggestion. Sarah, can we draft you?

DR. BOTHA: Sure. I will do my best.

DR. TEUTSCH: I think these are really critical issues that go beyond our traditional FDA-oriented clinical thinking about these issues.

Having reached this point and actually gotten to a decision, we have earned a short break. Thank you, Sylvia. Thanks to all the panelists. We will return at quarter past to continue. Thank you.

[Break.]

DR. TEUTSCH: We are going to move on to the next session. Welcome back. This session is on another very topical issue, Informed Consent, Privacy, and Discrimination Issues Related to Genomic Data Sharing. This is very timely, as we are hearing all of the discussions regarding HIPAA. We are going to take advantage of Kevin, as he always talks about these issues.

Kevin, let me turn it over to you to introduce the speakers and the discussion.

**INFORMED CONSENT ON GENOMIC DATA SHARING**

**Session Purpose and Overview**

**Kevin FitzGerald, S.J., Ph.D., Ph.D.**

DR. FITZGERALD: Thank you, Steve. Actually, it is great when you get to go a little later in the day because there are normally many references to the topic and the spectrum that you wish to address.

I have a lot of people to thank. I want to thank Greg Feero for leading us right into this, asking for next ELSI steps. I wanted to thank Robinsue, but I think she disappeared on me, for talking about the need to focus in on privacy. Is it a problem with the law; is it a problem with public understanding; is it more than all that; and if so, how do we describe that terrain. Also, we heard from Christy White about the lack of public awareness of legislation like GINA.

Finally, I would like to point out what Phyllis was talking about briefly. If you do go to those challenge grants and look in the bioethics area, every topic that is listed has some connection to this area that we are going to discuss now. You have informed consent and data access policies, unique ethical issues

posed by emerging technologies, ethical issues in health disparities and access to participation in research, ethical issues associated with electronic sharing of health information, ethical issues in the translation of genetic knowledge to clinical practice, ethical issues raised by blurring between treatment and research, and recontact issues in GWAS-like studies.

All of these things are going to impinge upon informed consent, privacy, confidentiality, potential discrimination, all in the sharing of data.

What we would like to do today to dive in the deep end, since we don't have enough time to wade from the shallow, is take a look at two areas that have already had some work done in them by other organizations that work in parallel to SACGHS.

Our first presentation will be by another person who is well known by this Committee, Rod Howell, who is with us representing the one group in government that has a worse acronym than we do for trying to pronounce as a word.

[Laughter.]

DR. FITZGERALD: I'm not even going to try to

pronounce it, but it is the Advisory Committee for Heritable Disorders in Newborns and Children. Rod is at the University of Miami. He is the professor of pediatrics and chair emeritus in the Department of Pediatrics in the Leonard Miller School of Medicine. He is going to enlighten us as to the efforts of our sister group. Thanks, Rod.

**Informed Consent Issues of Concern to the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC)**

**R. Rodney Howell, M.D.**

[PowerPoint presentation.]

DR. HOWELL: Kevin, thank you very much. I'm delighted to be here. Actually, our name has improved with the revision of our charter, which was just signed this February. Our name used to be the Secretary's Advisory Committee on Genetic Diseases and Heritable Disorders in Newborns and Children. Apparently, the folks that think about high things decided that "heritable" and "genetic" were redundant, and so they dropped one in the new charter.

I'm delighted to be here this afternoon. I'm

going to spend a fair amount of time actually talking about this Committee. I'm going to talk a little bit about what we have been trying to do. I'm going to spend quite a lot of time talking about the discussions of the Committee about how conditions are actually recommended for the newborn screening panel, which is one of the things you have been talking today about, the value and utility and so forth of various and sundry genetic testing.

Let me comment at the beginning of this that our Committee, although it has a fairly broad charter, has spent much of our time on newborn screening. There are several very interesting things about newborn screening that I think this Committee is very aware of but that I would like to remind you of again.

Each year we test 4.1 million babies in this country. At the current time, the average number of tests done on the baby is about 30. So we are doing about 120 million straightforward genetic tests using genetic technology.

The other thing that is very interesting is that all of this testing is done under the aegis of the

state health departments. These are public health programs. Although we focus and try to recommend national standards and national policies, the ultimate decisions about how they are implemented and what the take-up is reside with the states.

Let me just comment briefly. The Committee was authorized under the Children's Health Act of 2000. That is the same act, as a matter of interest, that also required the establishment of the Children's Health Study that is currently going on under NICHD. The Committee first met in June of 2004 and has basically been functioning for about five years.

At the time the Committee was founded, one of the driving forces that was going on, and a problem, was the fact that, as I mentioned, newborn screening is a state program. There had been extraordinary variability. This was becoming a tremendous problem, with some states screening for handful of conditions, others screening for many.

As people moved around, this created very real problems. If you had a child that was born in Connecticut and identified with a given condition, and

you moved to Virginia, which was one of the slowest states to move along, they were not screening for it. You had a new baby, so what did you do. It was a very big issue.

Let me show you what has happened since we started work in the summer of 2004. This is just a snapshot showing that at that time about 28 of the states in the country were screening for under 10 to 20 conditions. As you see, in December of 2008 those fewer than 10 and fewer than 20 have fundamentally disappeared. Virtually all the states in the country are currently screening for what has been recommended as a core set of conditions.

Fundamentally, this statute has said that we are supposed to come up with ideas and recommendations for a state screening program that would meet "federal guidelines." The Committee also was required to establish a grant program, which I might point out never had any money in it until last week. That will be an interesting thing.

Now, when we first started working on this, one of the discussions that came up in this august group that

I have the privilege of working with is that we were making all these recommendations but, since newborn screening is a state program, we could make all the recommendations we wanted but nothing was ever going to happen. The first slide I showed you has shown that not to be true. Basically, what has happened is that once national standards and so forth are recommended by a group that thinks them through carefully, the states tend to pick them up with their review committees. Also, I will not get into it, but parental work at the state level has been very important in moving this along.

A bill was recently passed in 2008 to reauthorize this Committee. It is reauthorized under a very large bill called the Newborn Screening Saves Lives Act of 2008. It was passed, unanimously I might point out, by a voice vote in both the House and the Senate and signed by President Bush in late 2008.

It has requirements for the Secretary of HHS to ensure quality of laboratories involved in newborn screening and to develop a national contingency plan for newborn screening. This became a very big issue during Katrina, when the state laboratory of Louisiana was

completely wiped out in the hurricane. You had all of the operations of the state, et cetera.

It also had specific discussions about the National Institutes of Health carrying out research in newborn screening, including new technologies. NIH has already been doing that, but it has a lot of language that directs the NIH and also names the program at the NIH the Hunter Kelly Newborn Screening Research Program after one of the big advocates for this bill.

The Committee has spent a great deal of time considering how conditions should be added to the panel. The nomination process has been worked on and approved by the Committee. It was felt that there should be broad access to the process, that anybody should be able to nominate a condition. The process should be very transparent. There should be consistent criteria, and there should be a structured evidence review group.

This is one of the more exciting things, I think, that the Committee has done. That is, there have been never been traditional evidence reviews of rare conditions because they are rare, and the traditional patterns of review don't work terribly well. The

Committee has contracted with Dr. Perrin at Harvard to organize and do evidence reviews in a systematic way of anything that comes to the Committee. The three areas of consideration are the condition itself, the test, and the treatment.

This is the nomination form. It is in your briefing book here. I won't spend a lot of time going into it, but it has a section discussing the incidence of the condition, the timing of the onset, and the severity. It has a lot of information about the test itself, as you have been discussing today, as well as how the test is to be used, the validity, the laboratory performance, confirmation, the risk, and the treatment. That includes modality, urgency, efficiency, availability, et cetera. It has a core set of references.

This is very similar to the nomination form that was used by the American College of Medical Genetics, but it has been polished and so forth. The very big thing is the evidence review committee.

The condition is nominated. The Advisory Committee looks at a nomination form like you just saw. The Committee and a subgroup of the Committee will look

at that and decide based on the information there whether it looks like a reasonable nomination and is sufficiently meritorious that it will be sent for an evidence review.

The evidence review is a big deal. It is expensive. Everything that comes along is not deemed worthy of an evidence review because of the money and time that it costs. Fundamentally, the Committee has approved that.

This is just a very simple thing. The nomination form comes in, and it goes through a federal administrative review at HRSA. Dr. Puryear is executive secretary of the Committee, and she resides at HRSA. Her staff looks at the nomination just to be sure it is complete. They do not make decisions, but all the stuff has to be there and so forth.

The Advisory Committee looks at it and then sends it for an evidence review. It goes through the evidence review and then comes back to the Committee, and they send a recommendation to the Secretary.

These are the questions that are in the evidence review. They basically are taken heavily off the nomination form, and I won't go into that. They

include the benefits of the treatment, the harms or risks, and the cost.

The evidence review has a decision model and evidence questions. The search methods that are to be used are defined. Dr. Perrin's group reviews peer-reviewed literature only, English only. They, however, do look at gray literature from pharmaceutical companies and so forth. They exclude case reports, which is a problem with rare diseases, but they do exclude those. They review consensus statements as guides but not to abstract those.

They do standard quality assessment methods. I might point out it is a traditional evidence-based system. They analyze any raw data that they can acquire from unpublished sources. They also routinely have focus groups of experts. They have investigators and families. Then they synthesize the data and provide it to the Committee.

They look at any rationales in treatments. Fundamentally, it is to provide timely information for the Committee so that the Committee can make specific recommendations.

The results come back to the Committee. We have had a chance now to have several of these reviews come back to the Committee. They summarize the key findings and they indicate, which is extremely helpful, where evidence is absent, what evidence would be most critical, what we don't know, the level of certainty, and new information.

The expert review group is independent and does not make decisions. It provides detailed information that comes back to the Committee.

The decisions by the Advisory Committee, I might point out, will be published. They are all on the website, but they will be published in journals as they come along.

Here are the recommendations that the Committee might make. Once it goes to the evidence review group, it comes back to the Advisory Committee. The Committee can review all that and make the following recommendations.

We can recommend adding to the core panel. That means that all the information is there, the data is there, it is convincing, it works, the treatment is

there, et cetera, and we should recommend that it be added. We have not yet had a condition come to the Committee that has met that level, I might point out.

The second is, we can recommend not adding to the panel but doing additional studies. The kind of information you would get back is that this is an important condition, the treatment really looks good, the test looks like it works, but there hasn't been a test done in a public health laboratory in a large group and so we really don't have sufficient information to recommend going to a core panel.

The third is recommending not adding to the panel but additional evidence is needed. That is very different because there just doesn't seem to be enough information there to make a decision. In other words, we don't know enough about the condition. Basically, you need to get this together and come back.

Finally is recommending not adding to the panel. That last recommendation is a level of certainty. In other words, the data are there. It does not seem to justify being added to the panel with certainty. That is a level of certainty. The first and fourth would be

certainty.

Now, at our meeting very recently we had two major discussions that I would like to describe to you. It is very much what you are dealing with here. The first was translational research policy, with introduction and discussion of institutional review boards and informed decisions. An extraordinarily important area that we discussed was residual blood spots and their policies and use.

The institutional review board discussion was moderated by Jeff Botkin. We had presentations and discussions by Ed Bartlett from the Office of Human Research Protection and from Alan Fleischman, who serves as ethicist on the National Children's Study. He is medical director of the March of Dimes.

Jeff Botkin provided an overview of the regulation and oversight of research with children. Dr. Bartlett discussed the regulatory options for multi-center research, meetings on alternative IRB models, and proposals to hold the IRBs directly accountable. Then Alan discussed the translational research and how we can make it work. He also provided an overview of the

California and Massachusetts models for obtaining informed consent.

Let me comment just briefly about the California and Massachusetts models of obtaining informed consent. When California was introducing tandem mass spectroscopy, it was deemed, since this was an experimental technology, that they would need to acquire informed consent in a large pilot project. That turned out to be extremely complicated, and they got only a very small portion of the people that they asked to participate. That has obviously been discussed a great deal, but about 25 percent participated.

On the other hand, Massachusetts had a similar type of program in that they had what I will call their usual pattern of screening tests that they were doing. As they decided to expand the panel, they did that with permission. Interestingly enough, they did this for a number of years, and it turned out that nobody was turning them down. In other words, they were getting permission from virtually everybody. Obviously, the method of getting permission was different, but that is a very interesting area.

Now, one of the reasons we are particularly interested in institutional review boards and research is that at the current time, as we move into new conditions that might be used in newborn screening nationally, we will be doing multi-center research programs. In other words, our Committee will not be, but the group that we work with will be. Obviously, these become very, very important issues to discuss.

Now, our final discussion was residual blood spot policies and usage. Harry Hannon, whom many of you know, has been responsible for the operation of the quality assurance program at the CDC for newborn screening for decades. Harry reviewed with the Committee the current patterns of storage retention and use of residual dried blood spots in the country.

I think that this group is aware of the tremendous interest in the dried blood spot at the current time. Obviously, it is used in newborn screening for looking for certain metabolyse enzymes, but it is obviously used for genome-wide studies in certain conditions.

Some states do not retain these spots at all.

In other words, they will discard them promptly. The major reason they discard them promptly is they don't want to deal with the question of how to store and use them. The safest way to get around that is to throw them away.

At the other end of the spectrum, there are states that preserve them in perpetuity in very careful conditions. California is certainly a good example of that. With 500,000 deliveries a year, they have literally millions of spots on hand.

I might point out, states will keep them for a huge variation, either weeks, months, or years. How the states use them was addressed by Jeff Botkin. They have commonly been used by state laboratories in establishing a new test. For example, if you want to set up tandem mass spectroscopy, it has been traditional that those spots would be anonymized and brought into the laboratory to see if your test is working and are you getting results. They have been used for that.

They have been used in an anonymized fashion by many, many states. Obviously, for them to be used with their name attached has historically always required

parental permission.

In talking about dried blood spots, it would be a travesty not to mention Denmark. Denmark has been retaining their samples for over 25 years. They have one of the most well organized and well monitored repositories in the world at the State Serum Institute there, operated by Dr. Bent Petersen. They have federal legislation dealing with those spots.

Those spots have proved invaluable in Denmark for a variety of studies. Number one, they can find all their people. People tend to stay in Denmark, and so they can find people for a long time. If they find a given condition in someone who is 20 years old, they can go back and retrieve that spot and identify things. It has really been a valuable repository.

For example, one of the things that they are considering doing at the current time, which we don't do in this country, is looking at the cytomegalovirus and how important it is for hearing difficulties. Denmark has an incredibly well organized hearing program. They know everybody in the country who has hard-of-hearing situations and how hard of hearing they are. They are

preparing to go back now and look at their dried blood spots to see how many of those might be related to CMV. They use those in a very efficient way. I might point out they have very discrete and well-defined federal regulations about what they can do.

Our Committee in the coming weeks is going to be drafting a white paper that will discuss some of the issues about institutional review boards. After considerable discussions, we obviously are going to make some recommendations to the Secretary about policies for retaining blood spots and informed consent for stored samples. I think these will be very key issues as we move forward in the coming weeks and years. Thank you very much.

DR. FITZGERALD: Thank you, Rod. Thank you again for a marvelous presentation, which I'm sure is going to raise a lot of questions. We are going to hold the questions for now. We will go to our second group, which is being led by Larry Gostin, who was the chair of the Institute of Medicine Committee on Health Research and the Privacy of Health. That then led to a report which is Beyond the HIPAA Privacy Rule: Enhancing

Privacy, Improving Health Through Research. Larry is also one of the editors of that report.

I have to tell you that Larry is a faculty member of a peerless academic institution here in Washington, D.C., often known as Georgetown University. With you, if I'm not mistaken, are a couple of others. Stanley Crosley is an attorney and chief privacy officer at Eli Lilly. Dr. Tom Croghan is senior fellow at Mathematica Policy Research here in Washington, D.C. Andrew Nelson is the executive director of Health Partners Research Foundation.

**Institute of Medicine Report: Beyond the HIPAA Privacy Rule**

**Larry Gostin, J.D.**

[PowerPoint presentation.]

DR. GOSTIN: We decided, since we have a relatively short amount of time, that we would dispense with all of us giving the remarks. My colleagues, who will come up and stand in the back, will hopefully be able to answer any of your questions.

I will take about 10 minutes or so to familiarize you with the report and then we will take

questions. I have to ask your forgiveness before I even begin because I do have to leave a little bit early. I have another appointment.

The Institute of Medicine had the following charge. We were asked to make an assessment as to whether the HIPAA Privacy Rule undermined or interfered with health research. If so, what recommendations might we make for the reform of the HIPAA Privacy Rule.

Clearly, this rule is of very great importance at the moment. The stimulus package gave a good deal of money for health information technology and also tried to firm up some of the provisions in the HIPAA rule. Similarly, it sent the rule back to HHS asking for some reformations, so we believe that our report is timely and important.

In answer to our charge, we found that the HIPAA rule did in fact undermine important and valuable health research. We therefore made a number of recommendations about privacy relating both to the HIPAA rule and to the Common Rule.

We took the view that there were two exceedingly and equally compelling values in society.

One of those values of course is privacy and security, so that patients must have strong expectations that their personal information will be kept in a private and secure way. At the same time, we thought there was an equally compelling individual and societal value in research because, without good quality research, the public is less safe and less healthy. It thwarts important scientific discoveries. We as a society have equally powerful interests in both.

The IOM Committee therefore made recommendations which we think will do both, which is to improve privacy and also to maintain and indeed facilitate important and valuable research in our society. We took the view that the HIPAA Privacy Rule and the Common Rule were actually intended to protect privacy, but in fact don't protect privacy very well at all. At the same time, they have the adverse effect of really impeding important research that we need to do in the country.

We therefore made two sets of recommendations. One is a bold, innovative approach to changing the entire framework or paradigm of how we think about

privacy, consent, and research in the United States today. It is something that doesn't follow the same model of autonomy, control, and ownership of information which has been very much a part of bioethics and law for a long time and, frankly, what the public expects. We are very clear that we face an expectation of the public that doesn't conform with our views of how this should be protected.

At the same time, as we have delivered our report and as we have talked to bioethicists, lawyers, and policymakers in the country, while not everyone agrees with it, everyone thinks that we need to have a new, fresh, careful approach to privacy and research.

The second part of our report was under the recognition that not everyone will agree with our innovative strategy. Even if they do agree, and we believe that many will agree, the political obstacles of doing that are extremely difficult. We therefore made a number of very careful, detailed, and, I believe, thoughtful recommendations for reform of the HIPAA Privacy Rule and the Common Rule which would have the effect both of improving privacy and facilitating

research.

Let me very briefly give you an account of these two approaches. First, the bold approach. Why do we say that the current model of authorization and each individual's control of information is not protective of privacy. There are several reasons. One is the fact that the Privacy Rule and the Common Rule are what lawyers call under-inclusive. That is, they only apply to a certain number of patients and transactions, leaving many other patients, research participants, and other transactions who are not covered under the rule virtually unprotected. So you have a rule that protects some and doesn't protect others.

The second reason is that we found that the Privacy Rule and the Common Rule are highly inconsistent and have extreme lack of uniformity. In any given situation, depending upon which rule applies or how the rule is interpreted by an IRB or a privacy board, what will happen is that you will have opposite or inconsistent results.

The under-inclusiveness -- that is, who should be protected and who shouldn't -- and the inconsistency -

- that is, two different people or two different circumstances of like circumstances being treated differently -- we found had no ethical, legal, or other principle that justified them. It was simply a question of happenstance in how these rules evolved over time, but there was no even colorable ethical reason why you would treat these situations so differently.

Finally, we find that the current model doesn't protect privacy because it is mostly formalistic and not meaningful. When a patient goes to a doctor's office, for example, and is given a privacy notice, most of us don't read it. I'm a law professor, and I barely understand it. It really wouldn't matter if I did understand it because if I didn't sign it I wouldn't be treated anyway. That is really only a formalistic way, the accounting for disclosures, the privacy notices. It is really substituting form for substance.

We wanted to go to a model that really was not something that was form but substance. We made a lot of proposals for essentially two things. One is to have very strong privacy safeguards to make sure that institutions that hold data for research purposes are

certified and are trustworthy. Secondly, that they have privacy practices as to who they would authorize getting that information which are consistent and strict. Third, that there are very detailed and careful security provisions.

If you think about what patients or research subjects should be worried about, it is really those things, not having absolute command and control over every bit of their information.

At the same time, we found that having this idea of consent doing all the work in this area thwarts research in very significant ways. We discuss many of them in the report, but one that I want to point out is the problem of selection bias. If each and every individual controls all of their information and some of them would be more likely to opt in and some more likely to opt out, it means that the results may be wrong or skewed in the wrong direction.

There are other reasons. For example, researchers may not need to have names and so forth, but they may need to be able to follow individual research participants over time. To do that, they have to have a

means of linking. We suggest that in our report in a way that we believe would be very helpful.

Finally, if you have any individual patient, or 10 patients, or 100 patients or subjects, or 1,000, or, in genome association studies, tens of thousands, if every single one of them could say, "I agree to this piece of information but not to that," or "You can use it for prostate cancer but not for breast cancer, or for heart disease but not AIDS and STD," to me, that doesn't make common sense. It really isn't protective of what we are trying to protect, which is to make sure that insurers, employers, family, and friends don't get this information in ways that harm or embarrass.

We make a number of very bold proposals to change the paradigm, but we also recognize the political problems and that not everyone will agree that we ought to change the model. We understand there are genuine differences of perception. We therefore make very detailed proposals about how we could change the Common Rule and the Privacy Rule either by more clarification in interpretation and guidance by HHS and OCR or by changes in the HIPAA rule. We notice in the stimulus package, as

I mentioned, HHS is being asked to reopen that, so we think it is timely. Finally, only if it is necessary, we will ask Congress to make some changes.

We tried to have a gradualist approach and make it as easy as possible for policymakers, if they agree with our approach, to be able to adopt it in ways that make sense.

We thank you very much for allowing us the opportunity to present our report to you. We will have a paper in JAMA summarizing our conclusions and adding additional observations in the first week in April. We will invite our staff and committee members to come up and answer any of your questions. Thank you very much for having us.

DR. FITZGERALD: Thank you, Larry. That was excellent. We would like to invite the staff members to come up, please. Rod, if you would please come back up, that would be great.

I think the presentations will probably engender a good deal of comment or question from this normally shy and retiring group, so I will throw the floor open at the moment. Sylvia, you get to go first.

**Question-and-Answer Session**

MS. AU: I just want to clarify something that Rod said. For the California program, actually what happened was it wasn't 25 percent of the participants gave consent to go for the pilot project for tandem mass spec. What happened in their state is that they decided they needed to go through the IRB of every single medical facility that was going to be in the pilot project. They didn't have the time or the manpower to actually do that with every medical facility, so only 25 percent of the newborns that were born in the state actually could participate because the other 75 percent were born in institutions that they didn't complete the IRB for. So it wasn't that it was 25 percent of all of the families that were asked to participate.

The only reason I know this is we were trying to do a comparison study with them. In Hawaii, we actually did active informed consent for our pilot, and we had people actually talk to parents for 20 to 40 minutes about tandem mass spec and newborn screening before they consented. We were going to compare it with the California program, who handed them a brochure and

had the nurse say, "Are you informed? Do you want to participate?"

We couldn't do that in the end because the California people realized that some of the nursing staff were sticking the "yes" sticker on without asking the patients if they really meant yes.

DR. HOWELL: I think Sylvia's comment brings up the issue of when you are trying to do informed consent for something that is national or state-wide and you have to deal with so many IRBs. It is a deadly problem. That is obviously a significant thing.

I think the other thing that Michelle reminded me of is that in Massachusetts they use an informed dissent program, which is a little bit different side of events. Again, many of us in the field feel that probably the best way to look at the informed consent in newborn screening is basically to have a very good information program and then have people dissent who do not want to.

DR. TEUTSCH: Before we leave the newborn screening, I have a quick one, Rod. It is great to see that this is getting on a much firmer evidence-based

footing. Going forward that should strengthen things. Are you going to have a chance to go back and look at the ones that were already recommended and reassess those to see how strong the evidence base is for those? I know that becomes a challenge.

DR. HOWELL: That has been discussed. At this point in time I don't think any decision has been made about that, period. It has not been made.

DR. FITZGERALD: Just following up on that issue, actually I'm intrigued by the body language here. Were any of you involved with working with Larry before?

[Laughter.]

DR. FITZGERALD: Anyway, in the report one of the issues I'm sure which is going to be huge to wrestle with is the database issue. The VA has a huge database. So does DOD. The Indian Health Service has a very interesting database in newborn screening. How are you addressing that particular issue with this idea of restructuring our way of looking at privacy?

DR. CROGHAN: The Committee has discussed the issue of linking databases, which is really a main part of what you just mentioned. It is very important to

health services researchers and will be increasingly important to all of us, particularly with genetic information.

There were several recommendations. The one I want to mention is to have some sort of certification of organizations that had met all of the criteria that Larry mentioned, such as security, privacy practices, and so on, who would then be trusted to take data from various data sets, link them in sensible ways that made them research-usable, and then make them available in a deidentified manner or in a limited data set manner, depending on what was most appropriate for the research question.

DR. FITZGERALD: Just as a follow-up to that, one of the issues that has come up before this Committee is this idea of how to define "deidentified" anymore. If we do start sequencing genomes for $1,000 and it only takes 70 SNPs to identify somebody, is there a set of criteria that you have for that particular issue? What are you going to use as a standard for deidentification?

MR. CROSLEY: The Committee looked at a lot of different resources when we did this. One of them was to

look outside of the U.S. as well. As you may know, the 27 member states of the European Union have an organizing body around data protection called the Article 29 Working Group, referencing the article of the European Directive that created the group. They have written a paper, WP139, which references in fact genetic information.

Their assessment was at this point sequencing of data and genetic information in general is still not identifiable without the reference.

That doesn't directly answer your question. Your question is, five to 10 years from now, 50 SNPs, 70 SNPs, whatever the number, how will that be created. I think that one of the recommendations from the Committee, apart from the Privacy Rule having its own model, enables you to be more nimble and to be more flexible in your assessments without all of the other entanglements of the rest of health care which the Privacy Rule has to consider as it makes changes.

I think Tom was explaining there are protective mechanisms around reidentification that the Committee focused on some, versus what is truly deidentified. We are setting up the model to prevent the harm rather than

trying to pursue an elusive concept of continually updating the deidentification criteria.

DR. FITZGERALD: Thank you. I just wanted to point out to everybody, in spite of our efforts to deidentify Tom, he is still identifiable because he is the only one left on the list who has not been identified.

Any other questions from the group? Yes, please.

DR. CAROME: I had a question for Larry and your colleagues. Separate from the issue of lack of coverage of the Common Rule, that it doesn't cover all human subject research involving data, and separate from the inconsistencies between the Common Rule and the Privacy Rule, were there specific provisions of the Common Rule that you identified as being problematic? That didn't come across clearly to me in looking at the Committee's recommendations.

MR. NELSON: The Common Rule is an HHS-wide adopted Common Rule. At the same time, trying to harmonize that with the Privacy Rule sometimes confuses IRBs. Oftentimes when confusion happens at a local

level, then more conservative decisions are made. So you have less organizations, less individuals, and less IRBs who are willing to do multi-site studies. Therein lies the complication.

DR. CAROME: So you really are focusing on the lack of harmony between two rules. If the Privacy Rule didn't exist and you only had the Common Rule, which applies to multiple federal agencies in addition to HHS, would there still be a problem? That is what I'm getting at.

MR. CROSLEY: Yes. There are a couple of things. One is a more comprehensive privacy regime to accompany the Common Rule and the acknowledgement that privacy and research are equally critical and equally important. The Common Rule isn't specific enough and doesn't go far enough in its privacy protective regime. So it is a marriage of the privacy regulations under HIPAA with the Common Rule.

Then there were some very specific security recommendations, regardless of which paradigm was used. I think that is probably the most significant.

Also, there were areas like secondary use.

There is a potential overreliance on the Common Rule having figured out how the IRB should advise on whether the consent form was sufficient to apply to some secondary use. Certainly, there was an understanding that expertise would exist within the IRB to solve some of the issues that we already have with the Common Rule, I think.

MR. NELSON: The final thing is that the Common Rule only covers what is funded by the federal government. We feel very strongly that this should apply to all research, no matter what funding source.

DR. FITZGERALD: Using a very complex and powerful algorithm, we have now identified Tom. We just wanted you to know that.

[Laughter.]

DR. FITZGERALD: Sue, did you have any comments?

MS. McANDREWS: Yes. In terms of full disclosure to complete the Georgetown control of this whole conversation, I did get my law degree from Georgetown Law. We now have all sides of the triangle there, and we rule.

On behalf of the Office for Civil Rights, I did want to thank the IOM for their report and their recommendations on how to improve privacy and security in the research context. We do appreciate their efforts in struggling with the very difficult balancing that we have dealt with in trying to design the HIPAA Privacy Rule in terms of individual interests versus societal interests. It is a matter of balancing the need for the data and the need of the individual for privacy and confidentiality when exposing their data and being willing to share their data in order to get the treatment that they need and deserve. We do not want fear of secondary uses to interfere with their ability to get care in the first place.

I want to just say that we have, since the beginning of the HIPAA Privacy Rule, endeavored to work with the research community in aligning the provisions and that we did make substantial realignments back in 2002 which did go to two of the areas that still showed up in the IOM report as needing further reconciliation. Those are the accounting for disclosures as well as the simplification of how you can go about waiving the

authorization requirements, largely for access to information as opposed to clinical trial interactions with the patients themselves.

In part, I would ask to what extent the report and the recommendations in those two areas really took into account the steps that were made back in 2002 and focused on the practices and problems that may have continued to reside in those two areas, as opposed to simply being a reaction to people's opinions back in 2000 when the rule was first issued.

DR. CROGHAN: I will start. First of all, in the interest of disclosure, I'm also a faculty member at Georgetown.

Secondly, I want to point out, we recognize the challenges that OCR faces. The Committee was of the strong opinion that privacy and health research are both private and public goods and that neither one occurs adequately without the other one. We really were trying to improve or enhance both in all of our recommendations.

With regard to the specific comments on notice about disclosure and so on, we did hear from OCR. In fact, they was very helpful in our discussions. We were

aware of the changes prior to the 2003 implementation.

We also heard from the research community. They are still barriers. Not as much as they would have been had the changes not been made, but they were still getting in the way of achieving our goals of enhancing privacy. We did hear from organizations who, because they didn't understand or correctly interpret, would not release records. Researchers had these experiences.

In fact, in our last meeting we also heard that the accounting for disclosure rules actually have a cut point of 50 records or something. There are in fact many research projects, including one that I recently had, where we were getting two or three records from a hundred physicians. Something like a third of the physicians just didn't understand the rules and therefore didn't give us the records.

So we did take the changes into account. There continue to be barriers. We think that they could be improved upon.

DR. FITZGERALD: Following up on that, when we look at some of these research programs that are going to use databases and the information that is there or can be

gathered, he newborn screening database actually might be one which is somewhat representative. Much of what we have right now as data is not truly representative of the diversity within this country.

The groups that have been marginalized up to this point may have good reasons within their groups for suspicion of benefits coming from any major research projects, but it is still my understanding that in order to get their information into these research programs in a way that will take into account their lack of representation, they actually need now to be overrepresented in the research programs that go ahead.

It seems you have a potential issue there that could really gridlock the system as we move ahead. Any thoughts on how to address that particular challenge?

DR. CROGHAN: I will start. We did some public surveys through the Harris Public Poll, and we had members on the Committee who represented patient groups. The most vulnerable groups, those with AIDS for example, those with mental health problems, those who had the most reason to be concerned about their privacy because of the potential for harm, were actually the ones who were most

likely to endorse releasing their data without prior consent and to endorse participating in research. Now, remember this is a public poll, so that comes with its own problems.

The members of our Committee who were engaged with these patient groups with chronic diseases, actually said, if you think about it, they also have the most potential for gain. They are the people who are seeking our help the most. In fact, they were the ones who were making this important decision. I think that was telling.

Andy has something to offer.

MR. NELSON: I really enjoyed your presentation about the potential for multi-site studies when you are looking at newborns. This capacity is a new capacity. When we look at intervention studies versus database studies and being able to aggregate large sets of data without bias, it is an extremely important societal benefit. We were very cognizant of wanting organizations to participate in that process. Right now there is fear among organizations for collaborating because they worry about any disclosure that those researchers might

produce, even if it is just the data-driven pieces.

I think we are looking for some supportive guidance from HHS to help organizations that are locally based to more clearly understand and more clearly give permission to contribute to the societal good.

DR. CROGHAN: We didn't absolve the researchers, by the way, of their responsibility. Part of this, we also found out in our polling, is that the public does not really understand research.

In focus groups, we understood that often people who had participated research did not hear back from the researchers. They didn't know what the results were. We make the recommendation that no matter which course is taken to improve on privacy that in fact researchers and others have the obligation to educate the public about research processes and the results of research.

DR. FITZGERALD: I have Gurvaneet next.

DR. RANDHAWA: In the discussions of the Committee I don't know to what extent you considered different models of data aggregation from the centralized, deidentified aggregate databases. The other

moral would be small federated databases where the data is all identified and controlled locally but there can be distributed queries specific to a research question or project so you don't have to aggregate data in any one centralized place.

I wasn't sure if the Committee had gone into the privacy issues for these two models and if one was better than the other one.

MR. NELSON: Yes, there is an increasing ability to conduct research through these federated data. In the example of the HMO Research Network, for instance, the identifiable data never leaves the firewalls of those care-providing organizations, but a query might be sent in from the outside and analysis would then be done inside with a large population. Only the aggregated deidentified results then transfer to the researchers outside. That is an increasing capacity, and it is very much encouraging in terms of protection and safety issues.

The second is, there are organizations that don't have that capacity because it takes quite a large effort to map and configure data that way. There has to

be the ability to be doing both the federated data consolidation approach as well as working with organizations that don't have that capacity.

MR. CROSLEY: The other thing I would add is that one of the models that we discussed and included in our report was having a certification agent, modeling it somewhat on the Ontario privacy law that has qualified entities who can hold reidentification keys. Certainly you can have that encryption key exist at the data level. You could also have a federated query authority as a trusted agent or an authentication agent that could then do the same thing.

I think the model certainly anticipated distributed data sets and having trusted agents or third parties who would in some manner be certificated to enable the research across those data sets.

DR. FITZGERALD: Joseph.

DR. TELFAIR: Thank you. I appreciate the presentation. I have probably contingency questions. This issue comes up a lot. I appreciate the presentation by Dr. Howell on newborn screening.

The one thing that is there as an example of

the others is the actual question of follow-up and longitudinality. You talked about maintaining longitudinal databases, but you also talked about working with the public and with vulnerable populations. I think one of the last things was the issue of scientists reporting back to the population itself.

Taken as a whole, the implications for that have a lot to do with the willingness to have these long-term databases and the ability to refresh those and go back. For example, you have someone who was picked up on newborn screening but then you had to go back to them at some point. The question really is, you did a lot of work on their sample early on but now you have to go back to them to reconsent. Were there any recommendations in a very practical way of how you would really do that?

I haven't heard a lot about it. It is a very tough problem. Given the recommendations you already have made, that seems to be something in line with what you have been thinking about. I was just wondering whether anything concrete may have come out of that recommendation-wise. Do you understand what I'm asking?

DR. CROGHAN: Let's take a little bit simpler

case first, which is an adult who can actually give consent. Here the Committee found a real discrepancy between what is in the Privacy Rule and what is in the Common Rule. People, under the Common Rule, can give consent to future research. Now, there are some boundaries around that, and the Committee did not get into the details about where to draw the line.

In the Privacy Rule, you cannot do that. That is one area of harmonization.

Now, we did not discuss at all the special issue of children and newborns, where the model is more you can assent children. I don't know what age is the bottom rung there, but that is something that we will kick back to you all as a Committee, and to others, to have that important discussion. I would imagine at some point there would be some talk about the need for consent.

DR. HOWELL: Let me make a brief comment. We did not discuss it at all today, but it is an important thing. The National Institutes of Health have just funded a major newborn screening translational research network.

The background is that when children are detected with rare conditions, be they in North Dakota or South Carolina, right now they basically are identified and their treatment is begun and then they are out of the system. The plan for this would be to identify and follow these children in a systematic way all over the country so that you would have all of the children with some rare condition. There would be plans to follow them, and there would be protocols.

One of the issues that has come up in a big way early in this is of course the data system. Early thoughts would be that the data would be retained locally but there would be an infrastructure, working with caBIG from the Cancer Institute as a model for doing that.

Anyway, this would be a very interesting thing. Steve asked if we are going to go back and so forth. We will have the prospective data on these conditions and we will know what happens to them and how they are treated, but the translational research network will be an exciting new program.

Again, a child will be detected. The parents will then be asked. They will go back to the child, but

the state, of course, always goes back to the affected person and asks, would you like to participate in the program, protocol, et cetera. They will be invited at that time to participate in the follow-up treatment protocol.

DR. TELFAIR: That is similar to the multi-site study models from multiple places. My other question may be even more difficult. I was thinking of the whole spectrum for the young person from birth on. They are very young, so of course their consent is given by their parent. Children and adolescents can assent, but they still have to have consent by the parent.

The other question is the vulnerable adults, those who cannot sign for themselves. You get a sample from them, and then you try to get a sample 20 years later but the person who signed for them is no longer there, for example. That is an adult-related problem. To me, those are real questions that are being asked.

I know you spoke about the European model, but I have looked at a lot of what they have and I didn't see that come up. I'm wondering is that, again, something you would kick back to us or do you actually deal with

it?

DR. CROGHAN: The Committee drew a distinction, and I think it is an important one, between interventional research and information-based research. Interventional research is the types of things that Rodney may have been referring to, where the research subject actually has something done to them, often in a randomized way, but there is some intervention that occurs. Our way in America of looking at those types of research is in fact consent.

The Committee drew a distinction between that and information-based research. If you have a sample about a child and you know something else about them from their administrative healthcare records over time, can a researcher access that information without ever needing to talk to or intervene with the research subject, even when they are an adult.

Now, we thought that with the appropriate controls, as Larry outlined, that could happen. We made the recommendation that that could occur within some boundaries.

MR. CROSLEY: With IRB oversight.

DR. CROGHAN: IRB oversight, appropriate security, and all the types of things we have been talking about.

DR. FITZGERALD: David.

DR. DALE: I really appreciate this discussion. The HIPAA rules are national rules, but the IRBs are locally controlled. Did you take a position on national IRBs, particularly related to rare diseases, where if you do a study you have to do it in multiple places?

MR. NELSON: We didn't go into that specifically. We did want to see, and made the recommendation on the Committee's behalf, to harmonize so local sites could have an easier way of interpreting things. Though this multiple-site IRB problem is not going to go away by the recommendations of this report, we think that better harmonization of rules so that local sites can interpret, and developing some templates that IRBs could follow, would be very helpful. Right now they are on their own.

MR. CROSLEY: We also made a recommendation that, regardless of whether it was the new model of research being pulled out of the rule or whether it is

changes to the rule itself, IRBs be given some layer of indemnification protection and liability protection. We saw from the research that came in that there was a vastly different interpretation of the Privacy Rule based on the constituency in the IRB and from one place to another. Those caused significant issues.

We tried to resolve that, as Andrew mentioned, by getting better guidance and some best practices that would be eventually blessed or sanctioned by HHS to give them freedom to operate within that sphere. The liability protection we thought was also a very important layer to give them the freedom to make good judgment and rely on their judgment in the circumstances.

DR. FITZGERALD: Michael, Sue, any further comment or questions from your end? No? Thank you.

One last question, then, for all of you. Going ahead, this Committee is going to continue to look at these issues of informed consent, privacy, discrimination, and all that. We have already touched on some of the areas that you have mentioned that you didn't particularly focus on, like children, newborns, adults that don't give their own consent. Are there any other

areas that you would like to see this Committee address from the perspective of the IOM report but also from the perspective of our sister committee? I will just throw it open to you.

DR. CROGHAN: The Committee's charge didn't include recommendations about genetics, so I'm now only speaking for myself. I think the issues that were raised here today, particularly with regard to integration of genetic information, how those data are maintained and how they are integrated with other protective health information and made available to the research community, are going to be an important part of any deliberation and something we need to think about.

We didn't consider genetics because they are not currently part of the HIPAA Privacy Rule.

DR. HOWELL: I think the thing that would be most helpful would be looking at the mechanisms of informed consent. When you have multi-site studies and the whole background that surrounds that as far as harmonization, a central IRB absolving the local IRBs of risk so that they might more readily do that I think is going to be very important. As Sylvia pointed out, even

in the State of California, you try to go to multiple IRBs and it just doesn't work. Solving that will be important.

I gather that the big issue with a central IRB is the fact that the local IRBs are still holding the bag, so they are really not willing to hear what a group of talking heads in Washington has to say on the issue because they have to deal with things back home. I think solving that and figuring out a way to do that in an ethical and legal way will be very important for genetic studies in general but particularly for newborn screening, where we are, again, looking at 120 million genetic tests a year and not 1,000 BRCA genes.

MR. NELSON: One other comment that the Committee did make is on this issue of transparency in the field of genetics, the use of phenotypic and genotypic data together, and the transparency of the discussion on the trust that has to come from the public. We really need to engage the public and figure out a way to engage them in a way that has their support. We need to communicate clearly the intent of what we are doing. We need to come up with a community-supported approach to

this privacy issue.

I think those discussions are extremely important and [constitute] a new science area where we have tools that are dramatically different than we have had in the past that expose privacy and security issues beyond what we have had to take care of in the past.

MR. CROSLEY: My final comment is not necessarily a recommendation on an area but some learning that we had in the composition of the IOM Committee. We had privacy advocates, patient advocates, people who suffered from chronic illness, and public and private researchers, and that constituency was incredibly powerful in sifting through the issues and making sure all the voices were heard.

I'm sure you are taking those things into consideration as you deliberate on these incredible topics because privacy and ethics, personalized medicine, it is an incredibly important and critical area. I think that we can't go very far unless we really start talking about it.

DR. FITZGERALD: Gentlemen, thank you very much. That was wonderfully interesting and informative.

I thank you for your participation.

[Applause.]

**Committee Discussion of Issues and Next Steps Related to Informed Consent on Genomic Data Sharing**

DR. FITZGERALD: I have my charge from the boss. He wants to know where you want to go next on these issues.

As we heard, there are areas that were just mentioned, some of which we have begun to address in some of our earlier reports. Certainly, public engagement has something that we have continually been bringing up, including the large population studies, the pharmacogenomics, and the genetic testing and screening.

There is also the question of, how will informed consent be reconceptualized, redescribed, and redefined. That does seem to be an area that is going to be rather neuralgic as we continue to go forward.

Would people feel it would be best that we get more information on a particular specific area? Do you feel ready to become a task force focusing on something? Where are people leaning at this point?

Just to let you know, Charmaine Royal, who will

be coming on the Committee as I'm being voted off the island, has agreed to do anything and everything.

DR. TEUTSCH: You know you can never leave.

DR. FITZGERALD: You never get to leave, right.

DR. TEUTSCH: That is what we need to hear. We have a lot of priority areas, and this was one of the ones that was important. Are there things that we can do now, long-term, short-term?

DR. BILLINGS: Maybe I missed it in the discussion, but do we know what the Institute is going to do with their work? Obviously, with all these people with these Georgetown connections, there is a certain institutional bias in the information that we got. I suspect that the other august institutions of law and ethics out there may have slight variances on the model.

DR. FITZGERALD: There are others?

DR. BILLINGS: Yes, yes, there are. Before I can say what I think should happen, I would like to know a little bit more about what is happening and how broad the range of difference of opinion is.

DR. TEUTSCH: Perhaps what is proceeding on the federal side with these issues, too. I don't know if

either of you can speak to that.

MS. McANDREWS: I certainly can't speak globally on that. I will say that last week the IOM did present the same report to the Secretary's Advisory Committee on Human Research Protections. That entity, SACHRP, has made recommendations on privacy and the intersection of the HIPAA Privacy Rule and research in the past. I suspect that they will be looking at their prior recommendations in light of this new report and will be propounding additional recommendations to the Secretary based on that.

Within OCR itself, as was mentioned and as you may otherwise know, we have a fairly full and ambitious regulatory agenda that has been handed to us courtesy of the HITECH Act which will be occupying our time and resources for the next year to 18 months, both in terms of regulatory changes and studies.

There is good news and bad news in that. None of the legislative changes in fact go to research at all. It wasn't really touched on in the HITECH Act.

In addition to those mandated statutory changes, and I would throw GINA into that mandatory

statutory work that we are engaged in, there may be some synergy in certain areas. A study of deidentification is one of the mandated areas that may allow consideration of what that term may mean in a research as well as a healthcare setting. There may be other things in the way of accounting for disclosures, although it is tending in an opposite direction from the recommendations of the IOM. That is broadening the areas for the accounting rather than taking items off the accounting.

Authorizations and other things may be areas that we will have an opportunity to work on in conjunction with our statutory mandates.

DR. FITZGERALD: Thank you, Sue. David.

DR. DALE: Is the full report available?

DR. FITZGERALD: Yes, it is.

DR. CAROME: The Secretary's Advisory Committee on Human Research Protections, SACHRP, met last week. They received a similar briefing on the IOM report. SACHRP previously made a series of recommendations about the Privacy Rule several years ago that are still undergoing deliberation and consideration by the Department. Those recommendations fairly well align with

many of the recommendations, or at least the general framework of the recommendations, that the IOM made. They tend to reinforce one another in terms of the concerns and issues that have been raised.

All of the recommendations of SACHRP to date are directed at the Privacy Rule and would require action by OCR, with input and consultation with others in the Department.

They mentioned today that they have concerns about the Common Rule. They focus on a lack of harmony between the Common Rule and the Privacy Rule, and that has been obvious to many for years, and a lack of coverage for all research involving human subjects that involves private information. When I pressed them on that, it is still unclear to me, if you didn't have the Privacy Rule and if the Common Rule covered all research, what problems the Common Rule poses to the type of research they are involved in. I'm still unclear on that.

They talk about not wanting to have the Department or the government go forward with prescriptive solutions, but by their very nature regulations are

prescriptive.

The current regulations we believe offer a lot of flexibility in this arena. There is a lot of research activity that isn't covered by the regulations either because the way it is done doesn't involve human subjects or the way it is done is exempt. For research that is not exempt and is covered, there are procedures for waiving informed consent, which have always existed. I believe those allow a lot of this research to go forward if the waiver is appropriate.

With regard to the provisions on privacy, there is one basic provision, and that is that when the IRB reviews and approves research it must ensure that there are appropriate provisions to protect the privacy of the data collected. That is a fairly simple provision which gives the IRB and investigators great discretion to design appropriate privacy protections. That can be along the lines of the privacy protections the IOM talks about, such as stronger protection and control and restrictions over release, but you can do all that now within the framework of the current regulation.

DR. FITZGERALD: Gurvaneet.

DR. RANDHAWA: Since we are at the information-gathering stage, one community we haven't heard about is the health information technology community. I'm sure they have wrestled with some of these issues from their perspective. It may be useful to engage with the successor of AHIC or somebody similar to give you some information on what is going on there.

DR. TEUTSCH: What I'm hearing is there is already some action being taken to flesh these things out. Just as a reminder, we had this session because we knew this report was going to be issued. That is why we wanted to defer the decision. It sounds like a fair bit is going on. There are a few loose ends but not major ones. There are some that relate specifically to the use of genetic information and privacy, as well as some data-sharing issues with the electronic medical records and information sharing there.

The question then becomes, do we monitor all of this at the moment or do we form a little workgroup to sort out whether there is something here that we can actually begin to do that will help inform this discussion? That is what I would like to hear.

DR. WILLIAMS: Joe.

DR. TELFAIR: Thank you. I appreciate the information because it narrows the gap a little bit. I guess my outstanding question in terms of a direction to go is, what can we make in terms of a contribution. I would recommend looking at the question related to the last item they discussed, which is vulnerable populations. How does this work within those groups.

I think much of what is being discussed is general population issues, but one of the things we do have a charge for is also looking at whether there is discrimination in working with vulnerable populations and then the permutations that have to do with that.

I don't know if there is a grant area around the whole thing. It seemed to me that we can focus on this one area. Maybe we can look at some of the other ones, but this seems to be a reasonable one that we can put on the table given that so much else is being covered. That is just a recommendation.

DR. BILLINGS: In response to your comments, Steve, I think it was fortuitous that you had Rod Howell there, too. The point about what can be done with the

Guthrie cards, that issue has been out there for a long time. I can remember an article by Phil Riley about this 15 or 20 years ago. That seems to me to be a practical genetics issue for this Committee, in conjunction with the activities that Rod is leading up, however they might proceed.

It is an important issue. We were talking about all these new technologies that can be applied. You can sequence the whole genome off these cards, maybe. What would that look like. What would the opt-in/opt-out rules look like for that, if any. How would it be used. As you said, it is a really nice non-biased population as well because it is broad. There are some positives and negatives to it. It seems to me that is a really interesting, specific issue which has been out there. It doesn't seem to be answered in policy yet, so we may actually have something useful to say.

DR. FITZGERALD: The question there would be how much of that is going to be addressed by that NIH grant that went out for the translational work in the newborn screening. I don't know that. We could ask Rod or we could ask ACMG.

The other would be taking that and saying, in a sense, that too is vulnerable population. Getting back to what Joe just said, depending on how we define or delineate vulnerability, that could be an issue that would be important to look at. That does raise in particularly emphatic ways some of these issues that, when you look at it more generically, don't necessarily get highlighted as strongly. I would say that would be something that would be a possibility.

DR. WILLIAMS: This goes off of what Gurvaneet mentioned about the AHIC successor. The other thing is that there was just an announcement that came out about another Secretary's Advisory Committee on Health Information Technology that is going to report to the Secretary of HHS. Now we have, by my count, four Secretary's advisory committees that have some piece of this pie.

It seems to me that one tangible suggestion would be to create a formal liaison group between the different committees that can assess where there is overlap and then perhaps in some ways divvy up the work so we don't all end up doing the same thing. It might be

good to have that group have the responsibility to say we are going to charge SACGHS with this and the Newborn group with this and Human Subjects with this. It might be a possible way to move forward.

DR. TEUTSCH: I agree. The Guthrie test issue and what we do with it longer term sounds like something that your Committee, Rod, is grappling with and falls naturally in that sphere. If you had something that could inform that, I think it would be good for us to know.

Would you have a concrete recommendation for next steps?

DR. FITZGERALD: I think the idea of coordinating with the other advisory committees is key. I think that is going to be important. I don't know if the other committees have the same charge as we do with regard to a group like vulnerable populations. We are genetics, health, and society, and that would seemingly be within our purview. Depending upon how that gets delineated, maybe that is the next step. If there is going to be some information gathering in this area, the step between now and the next would be how are you going

to delineate vulnerability and what is that going to mean.

As was mentioned here, certainly you have populations that are vulnerable because of particular medical conditions they may have. You have populations that are vulnerable because of historical or socioeconomic situations, like Native Americans or the poor. It is going to be important to figure out first how to delineate that and then see where you want to run with it.

DR. TEUTSCH: We also have the whole topic of vulnerable populations under our population health component. The issue here is that of privacy, research, and consent for those populations, which is a discrete subset. The question is, do we look at that more broadly in some other way.

DR. BILLINGS: I was just going to point out that the Common Rule has provisions for vulnerable populations as well. It is consistent in that sense as well.

DR. WILLIAMS: In terms of trying to make our work efficient and not to necessarily transition us into

the next topic, one of the groups under education and training has a focus on educating the public. I think we heard loud and clear from all the folks up here that we need to be engaged with the public and we need to have some role there.

It seems to me that there could potentially be some overlap with what we are going to hear about from Barb in a couple of minutes regarding what that task force is up to and how we could add in perhaps a piece of that and work together.

DR. TEUTSCH: I'm fine with that. I also think that I'm hearing a lot of concrete suggestions but nothing I think we are ready to quite talk about in a major way. We may ask you, Kevin, and maybe a couple of other folks, like Charmaine, to come back to us in June with something more concrete. We can learn about whether there is interest in having this consortium of the other agencies or the other committees. I'm not sure we are ready to proceed with those at the moment.

DR. FITZGERALD: I would certainly be happy to come back tomorrow, but June, I don't know.

DR. TEUTSCH: You have June and you have

October.

DR. FITZGERALD: I would be happy to work with Charmaine.

DR. TEUTSCH: Then we can explore some of those other issues.

DR. FROSST: I would like to follow up with a point relevant to what he said, which is that I have been mulling over since you said it the idea of these other Secretary's advisory committees and the vast amount of effort it takes to put together one of the reports that we do. I wonder if perhaps the other committees don't feel the same way about the herculean task that they take on.

There may be a way to merge a few of the committees together on a topic that is of relevance to more than one. I think to hit all four would probably be overly optimistic, but fantastic if we could. So this committee takes this view of it, and this view of it, and this view of it, and we come together at the end with something that really benefits the Secretary or whoever it is that is really looking at our products.

I have to say that in terms of process of doing

this, I'm not sure exactly what the best way is to do it.

DR. TEUTSCH: We can certainly put feelers out and have discussions with them before we actually recommend doing something to see what the receptivity is to that. Yes, David.

DR. DALE: I think this is a really important issue. I'm an active researcher. Almost every day this issue is in the way of the research, particularly for multi-institutional studies.

In my work, I have a compartment of isolated computers for clinical data and isolated computers for genetic data, and I have difficulty in linking them. I have another filing cabinet full of paper records which I can't look at between the people working in the space. This is multiplied by the multiple institutions. We have trouble cooperating with Canada because of our HIPAA regulations. It is just a mess.

I think it is a very constructive thing they have done. I don't quite know what to do because I haven't read the report yet, but I think that at our next meeting we should talk about this substantially.

DR. TEUTSCH: I do think we need to have some

of these discussions offline. Kevin, if we can wrap you at least into some of that with a twist. Charmaine is obviously going to be interested in some of that as well. We need to get her up to speed. People need to have a chance to review this report and tie it to either work of these other committees, the vulnerable populations, and some of the data sharing issues.

I think there is plenty on the table here. It is just what we can bite off that is not going to add to the noise and be constructive.

We are going to move on, then. Barbara, who has been leading the Education Task Force, is going to give us an update. I understand we have some data.

DR. McGRATH: Yes, we do.

## GENETICS EDUCATION AND TRAINING TASK FORCE

### Update on Data Gathering

**Barbara Burns McGrath, R.N., Ph.D.**

[PowerPoint presentation.]

DR. McGRATH: What I'm going to do today is give an update on the Task Force and provide some preliminary data. I actually thought that we were going to win the wow factor with this because we have a little

bit of data. I know in a lot of these meetings we have no data, just ideas. This afternoon there has been so much data coming your way that it is not such a big deal anymore.

The purpose of the session is to update you. We are about halfway through on this task force, I would say. We are finishing our data gathering, so it is a good time to see if anybody in the room has suggestions for whether you think we are heading in the right direction. We are not going to completely change direction, but we welcome suggestions for new areas to look at and emphasizes.

A little bit of background, particularly for the new members. This issue of genetics education and training has been high on the priority list of SACGHS since its inception. In 2004, there was a similar task force that was formed. They had a roundtable. Rather than a large report, they got away with just a letter and a series of recommendations to the Secretary of HHS.

We looked at those again around 2007 and, as a group, decided that it was time to look at it again. Things had changed enough. We decided that the issues

merited forming another task force to look at this. So we have been around for a couple of years.

In the meantime, we had a Cathy/Kathy switch. Cathy Fomous was the staff person initially, and Kathy Camp now is the staff person assigned to this, so there have been some changes.

The Committee talked about what should the scope of this task force be. Like a lot of things with SACGHS, it is really a hydra. There are so many different ways you could look at genetics education and training.

We talked about K-12 education. We talked about emerging groups that haven't been addressed who have needs, like laboratorians, hospital administrators, or speech pathologists. There is no end to the boundaries of where you could think about who might benefit from greater genetics education and training, if that is your ilk.

We did decide to limit our scope to three groups. We were guided by the principle of point of care, trying to think of limiting it along those lines. We decided to focus on healthcare professionals and

practitioners and their needs, public health providers, and then consumers and patients, including the public.

Underlying all of this is a hope that the results of this report will be recommendations to the Secretary of HHS and that our recommendations will be measurable and actionable. We are trying to focus on that angle. They are actually under the purview of HHS, trying to keep a focus on what is the role of the federal government in this area and trying to avoid getting too broad.

We are hoping to have a forward-looking document, not just looking at education tools that are in place now or education needs that are current but also look forward a little bit to what might be coming down the pike. Those are our hopes.

Those three scope areas were formed into workgroups, and I'm going to be reporting the data from those workgroups on their behalf. I think there are representatives of each workgroup still in the room, so we will lean on them.

The first one is the Healthcare Professionals Group, led by Greg Feero. He has a nice group of people

there that he works with. They are approaching their goal of trying to assess the training needs of health professionals by using a survey-based design. They are using two surveys. The first one is looking at professional organizations. They have done some survey on that. The next one is to use the same survey that was used in 2004 and try to compare some data with that. I will talk about that in a second.

Before I go further on that, all of the groups are doing review of literature of the areas that they are dealing with, with the goal of not to replicate existing efforts. We are trying to move forward rather than replicate what others are doing.

We have some of the results of those surveys. The first one, which is the one with professional organizations, identified 57 in those kinds of categories. Twenty-nine were general professional -- and these are professional organizations like AMA or American Academy of Family Physicians -- some of the genetics specialty ones, ones devoted to professional education with an eye toward certification, and then looking at three advisory committees.

The return rate today is 58 percent, but one survey came in this morning. We expect that there might be more coming in, so that response rate of 58 percent is likely to go up. Not surprisingly, from genetics specialty groups there was 100 percent response. The general professional ones were pretty good. The educational committees had a pretty low response rate. I won't go into why.

Preliminary data. Of those groups that you saw, half of them actually have something dedicated to genetics, which means half don't.

The question was, what do you identify as your organizational barriers to providing education to your constituents, and those are the ones that they identified. [Indicates slide.]

This slide shows in broad relief the ones that stand out as competing priorities. These are priorities that the organizations have for providing it. You can imagine what some of those might be.

One thought we have is that if there was increased clinical utility demonstrated for genetics and genetics testing that the numbers of competing priorities

might go down a little bit and it would rise as a priority issue. There are lots of other reasons to explain that one.

The second survey is the one looking at federal activities. Again, we are trying to compare has anything changed since the report of 2004. This is a smaller sample, for many reasons. One would be able to compare five of the agencies to that. The data analysis is just underway on that. We don't have a lot to say on that, but again, we are trying to see if there is any way to measure change over time with this.

Their next steps are to, of course, encourage the return of samples and do that comparative analysis and the complete data analysis. There are other reports coming out looking at genetics education and training from federal groups. We want to synthesize those reports so that they fit together nicely rather than duplicating or being really disparate. There are efforts to talk about synthesis.

Another goal is to have their report articulate personalized medicine initiatives. We want to ensure that some of the things that come out with that make

sense in terms of this report. That is that group.

The second group is the Public Health Providers Group, led by Joseph Telfair and his group of nice people. Their goals are similar. Their approach is to start with the notion of competencies. They have had the herculean task of gathering public health competencies around genetics and genomics from the various organizations. I think they started off with something like 100. They are working to whittle those down to a concrete set of 12 that at this point seem to be the core ones.

That set of 12 will inform the development of a survey to then be administered to the right people to see if they are achieving the competencies. If so, we want to know where they get the education. If not, we want to know where they wish they would.

These are examples of the kind of competencies they are talking about. These are four of the twelve -- I will just let you read them for a second -- looking at up-to-date scientific knowledge and behavior, opportunities to integrate into healthcare practice, of course the ELSI issues, and then how to implement

research. It clearly covers the whole public health arena.

That part is finished. The next part will be developing the survey. It will be an online survey to be distributed. They are at that point, so the survey should go out pretty soon. Then there will be data analysis of that.

The last group is the Consumer and Patient Workgroup, led by Vince Bonham, who is not here right now. He is in Africa. Sarah Harding will be here tomorrow, and she is filling in for him.

This is their group. We are proud to add a new member. Gwen Darien has agreed to join us, so that will be an excellent group of nice people.

Their goal is to provide recommendations that address the needs of consumers and patients. Their approach is to start with qualitative interviews. They conducted five paired semi-structured interviews with professionals in the following areas to get the landscape of identified areas of genetic needs for patients and consumers.

The data is just being analyzed, but some early

thoughts are that, not surprisingly, consumers get information from providers and the media. Interestingly, they feel government does have a role to play in this in terms of guidance.

Those interviewed people suggested that the need that they see for consumers coming up the pike is greater understanding of multiple risk factors and how genetics plays with that. Obviously, that is important, along with the role of the environment.

Other needs are for some discernment about the expertise among healthcare providers, who you go to for what sorts of issues, and some helpful tools. We talked about that with DTC this afternoon. We need some tools to evaluate this.

Some of the barriers that those professionals and advocacy groups identified for consumers were just general poor health literacy, a notion of genetic determinism or fatalism -- why learn about this when there is nothing you can do about it? -- and then fear of discrimination continuing even past the GINA era.

What they will do with those themes is to turn this into a survey, which is happening right now, and

then to distribute these to larger community-based organizations. The hope is for an N of about 100 of these, so a pretty good size for this kind of project.

Our group met this morning before orientation for this meeting, and one thing we talked about is the challenges of addressing the issues identified by the general public. So far, we are focusing on consumers and patients, meaning people that have some reason to be interested in genetics. We know the general public perhaps has a different orientation to this. The challenge of who is the general public and how to access attitudes from them, we don't have an answer to. We are going to talk about that further. There is a desire to see that we integrate that with this report.

I'm hearing some more about integrating some things about informed consent and research with genetics. We will talk about that.

Here is a scary slide. This is the timeline. We are working now on collecting the data and writing the background. That will go on until summer.

Our next step will be to develop some draft initial recommendations that we will present to the whole

Committee at the June meeting. These will be recommendations based on analysis of the data I just presented. In that meeting we will come to some agreement about the draft recommendations. That will go into a draft of the report, which will be written over the summer and sent to you at the end of summer for your end-of-summer reading. Get your novels done early because you will get this report at the end of the summer.

We will present that draft report in the October meeting, and then it will go out for public comment over the holiday in November. The final report is anticipated to be ready for publication and submission to the Secretary next year, probably in mid 2010.

We are pretty much on track, but I think the heavy lifting is yet to come in terms of the writing.

I would like to stop talking and see if people think from that brief review that we are on the right track. Are there things you would like to add or minimize? I will very much refer to the rest of the people on the workgroups because there is definitely a shared governance committee.

**Committee Discussion**

DR. TEUTSCH: Thank you, Barbara. Any comments from the group? Any thoughts for Barbara? Gurvaneet, we can count on you.

DR. RANDHAWA: I think this is just great work.

DR. FROSST: I second that.

DR. TEUTSCH: Any thoughts for this committee before we turn them loose again? I know they have been working hard.

DR. McGRATH: We can take written comments, too, if you are more awake. You can send Emails.

**Closing Remarks**

**Steven Teutsch, M.D., M.P.H.**

DR. TEUTSCH: Hearing none, it is good to see all the progress, Barbara. Thank you for that.

It is hard to think we are going to break up early. I know people won't know what to do with themselves. I think we have had a productive day, hearing from our agency colleagues on the DTC work, the challenges of privacy and informed consent, and the work of the Education Committee.

We will adjourn, to return tomorrow. We will

hear a little bit more from our colleagues. We will spend most of the day talking about the implications of genetics and health reform, particularly from the payers' perspective.

[Whereupon, at 5:13 p.m., the meeting recessed to reconvene the following day.]

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CERTIFICATION

This is to certify that the attached proceedings

BEFORE THE: **18th Meeting of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS)**

HELD: **March 12-13, 2009**

were convened as herein appears, and that this is the official transcript thereof for the file of the Department or Commission.

SONIA GONZALEZ, Court Reporter