Roundtable Introductions

DR. McCABE: Dr. Zerhouni, thank you very much for opening this first meeting of the Secretary's Advisory Committee on Genetics, Health, and Society.

The public was made aware of this meeting through notices in the Federal Register, as well as through announcements on the Secretary's Advisory Committee on Genetics, Health, and Society, SACGHS, website and listserv. I want to welcome all of the members of the public who are in attendance, all of you here in the meeting room, those of you viewing the proceedings from the overflow room in the hotel, as well as those of you who may be watching from offices and homes via the webcast.

Members of the public in attendance who wish to address the committee may do so during the two public comment periods. If you do want to make comments and haven't yet signed up, please do so at the registration desk. We do ask that you register for those comments.

We're going to go through a round robin, go around the table also to our representatives of the various agencies, and introduce ourselves. I would ask everyone to try to keep that to one to two minutes of introduction so that we can have time to fit everyone in. I will begin, and then we'll go around in alphabetical order here, and then alphabetical order by the agencies from the ex officios.

So with that, let me begin by introducing myself. I'm Ed McCabe. I'm Physician-in-Chief for the Mattel Children's Hospital at UCLA. I actually began in genetic testing at the age of 15 when I was given the good fortune of working in a pediatric research laboratory at the University of Maryland School of Medicine. My job was to screen urines from Rosewood State Hospital, an institution for the mentally retarded at the time, looking for PKU and other inborn errors of metabolism. Then at the University of Colorado, when I was a little bit more mature as a fellow, I actually learned how to run a biochemical genetics diagnostic lab and began my career as an independent investigator.

In the late '70s, I was involved in the expansion of the Colorado and Mountain State Screening Program. I've done research on newborn screening. I chaired the American Academy of Pediatrics Committee on Genetics, which was very involved in newborn screening, and then began to do research showing that DNA was in those dried blood spots and moving from technologies that we still use that were developed in the '50s and '60s to molecular genetic technology.

When I moved to Baylor College of Medicine, I began to be involved in broader genetics policy and was chair of an advisory committee to the Texas Genetics Network for organization of private and public genetic services in the State of Texas, and also began to work with Tom Caskey on the Human Genome Initiative, first with the DOE and then the NIH.

I continue to be involved with the American Academy of Pediatrics, co-founding the AAP section on genetics and birth defects. I've been president of the American Board of Medical Genetics, president of the American College of Medical Genetics, have served as a genetics consultant to the Clinical Laboratory Improvement Advisory Committee, and chaired the Secretary's Advisory Committee on Genetic Testing under Secretaries Shalala and Thompson.

We have established the UCLA Center on Society, the Individual, and Genetics to look at the tension between the interests of society and the interests of the individual, and I think that it's interesting that it reflects very much the broadening of the scope of this committee.

I'm really honored to be appointed to the Secretary's Advisory Committee on Genetics, Health, and Society and to have the opportunity to provide institutional memory. I think it's important to use that rather than the term "continuity," because we are a new committee, but hopefully we can learn from some of the things that we experienced and began to develop with the Secretary's Advisory Committee on Genetic Testing.

I'm pleased to have been appointed along with two other colleagues who also served on the SACGT, Cynthia Berry and Reed Tuckson. The SACGT was one of the most exciting and fulfilling experiences in my career. I look forward to working with all of you to assist the Department of Health and Human Services and Secretary Thompson to address the issues in our charter. I anticipate that our service on the SACGHS will be an exhilarating experience for all of us.

Thank you very much for your kind comments.
MS. BERRY: Good morning. I'm Cynthia Berry. I am General Counsel and Managing Director of Wexler and Walker Public Policy Associates. It's a government affairs firm in town. I'm an attorney with a trial background and also health policy background, so I care very much about the intersection of medical science and discovery and public policy, as well as the ethical issues involved with that.

I currently serve as co-chair of the Virginia Birth-Related Neurological Injury Compensation Program. That's a mouthful. It's one of only two such programs in the country, and it's quite a challenge. We have a special program and fund set up to care for children who suffer from birth-related injuries due to oxygen deprivation. Instead of going through the tort system, they receive all of their care, all of their help for the rest of their lives, as well as their family, through our program, and that's been quite a challenge. There are actually issues that we face all the time -- health-related, public policy-related, ethical issues -- in that group, and I'm looking forward to working here on the innumerable challenges that we face in this area.

Another part of my background is that I was Washington counsel for the American Medical Association. I also worked on Capitol Hill for Senator John Kyle when he was on the House side, handling health policy issues for him as well. Prior to that, I practiced law in Nashville, Tennessee for several years, did trial work as well as commercial work. Again, just looking forward to working with everybody on this committee and hearing all of the public comments, because no matter what your background is, we may all think we're experts in a particular area but we all have so much to learn. We're plowing new territory.

I look forward to receiving all of that information and soaking it up like -- is it Sponge Bob Square Pants or whatever?

(Laughter.)

MS. BERRY: Thank you very much.

DR. McCABE: Thank you.

DR. HOOK: Good morning. I'm Chris Hook. Professionally I'm a hematologist and medical ethicist at the Mayo Clinic in Rochester, Minnesota. A good portion of my practice in hematology is in the areas of coagulation and non-malignant hematology, so I deal with genetic diseases and maladies on an almost daily basis in that practice. Some of the other specific ethics activities that I do at Mayo include chairing

their ethics council, but also I have served in the past for our IRB in the development of its policy regarding genetics research, and one of the outcomes of that process was to be invited to speak to President Clinton's Bioethics Commission, and then also to an NIH group concerning how our institution had dealt with issues of genetics research.

Presently, we're expanding activities to focus not only in genetics research in general but in psychobehavioral genetics research. I'm working with our chair of psychiatry to spearhead an ELSI support committee for that activity.

On a personal level, I have significant concerns about genetic issues, as all of us do. Next week my wife is going to have bilateral prophylactic mastectomies because of a significant family history of breast cancer. My oldest son is on the autism spectrum, and we're learning more and more about the genetic indications of those sorts of disorders. I carry a bicuspid aortic valve and look forward to becoming a cyborg in the next 10 years with a new prosthesis.

So these issues are personal to each and every one of us, and like Cynthia, I'm looking forward with great anticipation to learning from my colleagues on the committee and from our friends and colleagues in the community, to learn of their concerns and how we can serve them. I'm very honored to be here. Thank you.

DR. McCABE: Thank you.

MR. MARGUS: So I have absolutely no institutional memory. I'm the new person. I feel very honored and appreciative of the opportunity to meet everyone here and to serve on this.

Ten years ago this week, two of my sons were diagnosed with a brutal genetic disease called ataxia telangiectasia that combines muscular dystrophy-type symptoms, loss of muscle control with a deficiency and about a 40 percent cancer rate. At the time, I was pretty much a Forrest Gump running a shrimp company and I had no knowledge of microbiology or molecular genetics or how government funds research or how scientists think. But over the last 10 years, a lot has happened in my life.

I'm currently the volunteer president of an organization called the A-T Children's Project that helped to find the gene but also continues to orchestrate research to try to find a treatment for kids. At the same time, I'm currently the CEO of a company called Perlegen Sciences. It started out in Mountain View, California two years ago. It uses high-density oligonucleotide microarrays to hold genome scanning, trying to find the genetic basis of common diseases or drug response.

I've been on several boards but on the board of the Alliance of Genetic Support Groups, which is now called the Genetic Alliance. It's an umbrella organization representing a lot of people with genetic disease. So I've dealt with those issues.

Besides the kids having A-T and having this terrible disease, it turns out that heterozygotes with one mutation like me supposedly have a three- to four-fold risk of cancer. So we don't worry about that day to day. All we care about are our kids, but there are issues related to some insurance discrimination that we've dealt with.

DR. McCABE: Thank you.

DR. TUCKSON: My name is Reed Tuckson. I'm trained as an internist and a general medicine physician. Early in my career I spent a lot of energy and time on being a consultant on sickle cell disease.

I served as the administrator for the Mental Retardation Developmental Disabilities Administration for several years in this city, eventually becoming the Commissioner of Public Health in this city for several years; then Senior Vice President of the March of Dimes Birth Defects Foundation, where, among other things, I was responsible for the research agenda for the March of Dimes.

I then had the opportunity to serve time, six years as the President of the Charles Drew University of Medicine and Science in Los Angeles, and again given a good opportunity I think to understand the issues of research and science. I was then able to serve as the Senior Vice President for Professional Standards of the American Medical Association, where I supervised medical education, ethics, public health, research, science and technology. Now I'm serving as the Physician Senior Vice President of UnitedHealth Group, which is a large multifaceted health care organization that touches the lives of about 40 million Americans and is very rich in the use of data in being able to improve the quality of health care for those that we insure and those who we organize health care services on behalf of.

As mentioned earlier, I did enjoy my service as a member of the previous, although non-continuous committee, and I'm happy to be here with my colleagues.

DR. McCABE: Thank you.

MS. ZELLMER: My name is Kim Zellmer, and unfortunately I'm not here because of my scientific expertise. I'm here because I have a daughter who is six and a half who has neuronal ceroid lipofuscinosis. Thankfully for us lay people, they also call it late infantile Batten's disease. My daughter, Maddy, developed normally until about age 3. At age 3, she began having seizures, and that was the first sign that there was any problem. So for the next year and a half, we struggled with trying to get seizures under control, and when we thought we were winning that battle, unfortunately she began to lose skills. She lost her ability to walk, her ability to talk, she lost her eyesight.

About that time, we finally got the diagnosis, which is obviously a devastating disorder. It's a terminal disorder. Her life expectancy is eight to twelve years old. So I have first-hand knowledge, I guess, of the devastation that genetic disease can cause to a family.

I have a three-and-a-half-year-old daughter, Megan, who is a carrier of the disease but thankfully is not affected. Through Madeleine's disease for the last two years, I have been fortunate enough to meet a lot of families who are struggling with various genetic disorders, who have children affected or who are affected themselves.

When I'm not spending my time being a mother to Madeleine and Megan, I'm an attorney. I work for the Husch and Eppenberger law firm in Kansas City. I do estate planning. I'm also president of Heart of America Batten Disease Support and Research Association. Hopefully I can bring a different perspective to the committee.

DR. McCABE: Thank you.

DR. LEONARD: Hi. I'm Debra Leonard. I direct molecular pathology at the University of Pennsylvania. I'm boarded in molecular genetic pathology, and my practice is really the translation of the Human Genome Project into diagnostic tests, not only genetic tests but also cancer-related testing, infectious disease testing,, identity testing methods. The focus of my career has been basically setting laboratory standards and practices for laboratory testing, including genetic testing. I've been President of the Association for Molecular Pathology and work with the College of American Pathologists on their molecular pathology committee, as well as working with government relations.

I'm also interested in the training of medical students, physicians, physician trainees in genetics and laboratory practices and work with the American Board of Pathology on their molecular genetic pathology committee to set the examinations that will be taken by physicians to be board certified in molecular genetic pathology. My research interests are the impact of gene patents on clinical genetic testing as well as research, and I work on this with Mildred Cho and John Mers, and it's really an honor to be a member of this committee and work with all of you. Thanks.

DR. McCABE: Thank you.

DR. LANDER: I'm Eric Lander. I'm a Professor of Biology at MIT and a member of the Whitehead Institute for Biomedical Research in Cambridge, Massachusetts. I direct the Whitehead MIT Center for Genome Research, which is one of the centers that's involved in the Human Genome Project and continues to be involved in the successors to the Human Genome Project.

My background is originally as a mathematician, and my scientific interests are human genetics, particularly understanding the genetic basis of the susceptibility to complex disease. I'm very proud of the work of the Human Genome Project over the last 15 years, and my interest in serving on this committee is that I want to be very proud of the work of the Human Genome Project 15 years from now. It will take an awful lot of work here to make sure that that is the case.

DR. McCABE: Thank you.

DR. REEDE: Good morning. Joan Reede. My background is in pediatrics and child psychiatry. I've served in a number of places before coming to Harvard Medical School, where I am now. So I've worked in community health centers, in juvenile prisons, in public schools, and ended up at Harvard Medical School, where I'm now Dean for Diversity and Community Partnership and oversee the Minority Faculty Development Program and the Office for Community Outreach programs, and also run a center of excellence in minority health and health disparities.

My faculty positions are in medical school, the Harvard School of Public Health, and my focus is really on bridging academic medical centers to communities, and also on the training and education of physicians to deal particularly with issues of minority health, the health of minority populations and disadvantaged communities, with a focus on areas of health policy. I recently stepped off the Secretary's Advisory Committee on Minority Health, and also served on the Board of Governors of the NIH Clinical Center.

DR. McCABE: Thank you.

DR. WILLARD: Good morning. I'm Hunt Willard, Director of the Institute for Genome Sciences and Policy at Duke University and Vice Chancellor for Genome Sciences at Duke and the Duke University Health System. Prior to taking on those jobs, earlier this year I had been Chairman of the Department of Genetics at Case Western Reserve University at Cleveland for the past 11 years during a time at which we attempted to put together, along the lines of the old pathology model, both basic science and clinical genetics activities under the same departmental umbrella and fuse training activities across that spectrum, trying to anticipate precisely the kinds of activities that we're doing on this committee.
In the institute at Duke University, this is a somewhat unusual institute in that it's campus-wide. So although the center of gravity for the institute is decidedly at the medical center and the health system, it in fact bridges across the entire campus. So I'm working within the institute with people from the law school, the business school, the engineering school, the divinity school, the School of the Environment, et

cetera, in order to take head-on this charge of figuring out how to translate genome sciences into issues that are relevant to society and the public policy issues that are critical to that effort.

It feels like I'm a lifelong member of the American Society of Human Genetics. I served as its president a few years ago, and I'm pleased to be on this committee as the manifestation of a commitment on many of our parts and now the government's part to make sure that the products of the Human Genome Project and human genetics more generally do indeed translate into the betterment of society and the public good.

DR. McCABE: Thank you.

MS. HARRISON: Good morning. My name is Barbara Harrison. I'm a Certified Genetic Counselor. I work at Howard University. I am the Co-Director of the Genetic Counseling Program there, and so I'm very concerned about the training of competent genetic counselors and have a special interest in training people of color to be involved in genetics both as geneticists and genetic counselors, and just increasing the competency in terms of being able to serve people of color as well.

I'm in the unique position as a genetic counselor of having most of the scientific knowledge, as well as knowing first-hand about the issues that families deal with that have to do with genetic conditions. So I'm hoping to bring that perspective to the committee, and I'm very honored to be here.

DR. McCABE: Thank you.

MS. MASNY: Hi. I'm Agnes Masny. I'm a Nurse Practitioner and a Research Assistant at the Fox Chase Cancer Center in Philadelphia. I work primarily in a family risk assessment program where we service women who are seeking information about genetic testing or, actually, risk reduction measures if, in fact, they are at risk for cancer. As well, I work as an educator and have some funding through the National Cancer Institute in training nurses in this whole area of cancer genetics and the counseling and education that needs to go along with that.

I've had the opportunity to serve on the National Coalition for Health Professional Education in Genetics, and I'm very interested in this area about health professional education. I also work as a project manager with some network hospitals that belong to our institution in seeing how this whole area of cancer genetics and the risk assessment programs can be actually integrated into community-based settings. So I oversee the program as well as the training of the nurses who are running the program there.

I serve also as the coordinator for the Oncology Nursing Society's Cancer Genetics Special Interest Group, again trying to see the ways that nurses can keep up to date with the cancer genetic information that is emerging. I had the opportunity to give a presentation at the prior Secretary's Advisory Committee meetings on behalf of ONS, so now it's a real honor for me to actually be able to sit on the committee. Thank you.

DR. McCABE: Thank you.

DR. WINN-DEEN: I'm Emily Winn-Deen. My job title is Senior Director for Genomics Business with Roche Molecular Systems. I have a Ph.D. in biochemistry and have spent pretty much my whole working career working in the development of diagnostic test reagents. About the same time that the Human Genome Project began, I started working in the area specifically of DNA diagnostics and over the years have felt that there were a number of different areas that needed focus. Certainly, there was a need for technology development. There's a need to understand the underlying genetic associations with human disease.

But I now feel that beyond those sort of technical issues, we also have a number of public policy issues that we need to deal with in order to really realize the potential of the Human Genome Project to make an impact on health care, and as such I'm currently sitting on the Roche Joint Programs in Applied Genetics Public Policy Committee, and it's a tremendous honor to be sitting on this committee as well.

DR. McCABE: Thank you.

I'm now going to go through the various agencies and departments. I would ask that you focus really on the efforts of your department or agency. You can certainly briefly introduce yourselves, but we're really most interested in how the interests of your group will relate to this committee.

So we'll start with Department of Commerce.

DR. BEMENT: Thank you. I'm Arden Bement, Director of the National Institute of Standards and Technology. My background is in materials engineering, materials science and engineering. I've had appointments in academia at MIT, Purdue University, adjunct appointments at Case Western Reserve, Ohio State University. I had appointments in the Defense Department, the National Science Board, and currently serve in the Department of Commerce.

The National Institute of Standards and Technology is active across a number of fields in biotechnology and tissue regeneration. Our focus is primarily on measurement science and standards, standard reference materials and standard databases. Primarily in genomics and proteomics, we maintain at several universities a protein data bank which is getting more and more hits per month. We set standards for forensics DNA. We have a DNA database which we operate through the National Institutes of Justice.

We're quite active in single molecular measurements, automatic gene sequencing techniques. We have a dental materials center. We're quite active in developing microarrays and studying microphilytics involved with microarrays, automatic gene sequencing chips, gene chips and the use of micropores, both natural and synthetic, for studying DNA and RNA sequencing.

That's not exhaustive. We work closely with the University of Maryland Biological Institute, and also several institutes in the National Institutes of Health, more recently quite active with the newest institute, the National Institute for Bioimaging and Bioengineering.
DR. McCABE: Thank you.

Next we have the Department of Defense.

DR. TURNER: Good morning. My name is Martha Turner and I'm honored to be here representing the Department of Defense. I was asked to represent the Department of Defense in my capacity as the ethics consultant to the Air Force Surgeon General, but in fact I represent all three services here.

The Department of Defense has broad interest in genetics in three main areas: force protection, operational readiness, and health care for all of our beneficiaries. These include the active duty members, their families, and those who have retired. Across all of these, we consider the multiple ethical implications, including access, confidentiality and discrimination, some of the things you've already mentioned.

For force protection, we're looking at survivability in the field, and one example would be early exposure identification and individual protection for the soldiers. For operational readiness, we look at

conventional weapons and weapons of bioterrorism and the human response to those weapons. Related to operational readiness, we also have some interest in the forensic application of genetics such as the identification of remains. In health care, we look at drug response, disease-specific prevention, diagnosis and treatment. Across all of these, again, in practice and research and education, we strive for balance of science and ethics.

DR. McCABE: Thank you.

We were just notified that the speakers need to get closer to the microphones because there's some difficulty hearing, and we certainly want the people who are listening to us on the Internet and in the other room to be able to hear. So please do get closer to the microphone.

Next we have Department of Health and Human Services, Administration for Children and Families.

MR. DANNENFELSER: Hi. My name is Martin Dannenfelser. I'm Deputy Assistant Secretary for Policy and External Affairs with the Administration for Children and Families. In first coming to Washington in 1981, I served on Capitol Hill for about 14 years and worked for a member of Congress with a very strong interest in the area of medical ethics and continued involvement in that area in the non-profit world for about six years before coming to HHS about two years ago. We deal with the human services side primarily of the issues at HHS, such things as Head Start, child care, child welfare programs, welfare reform, and so on.

As part of the Secretary's initiative, the one department initiative, I am often the representative from ACF on a variety of task forces dealing with such things as HIV/AIDS, program coordination, biomedical ethics, health disparities, domestic violence, and issues like this here. So I try to bring a perspective of the overall well-being of children and families and how we can make a contribution in this area as well.

DR. McCABE: Thank you.

Next we have the Agency for Healthcare Research and Quality.

DR. FELIX-AARON: Good morning. My name is Kay Felix-Aaron and I serve as the Senior Advisor for Minority Health for the Agency. I am a physician and health services researcher, and my career has been guided by improving access and quality of care for low-income communities.

The Agency for Healthcare Quality and Research, AHQR, is interested in the intersection between health care and genomics, and particularly its application in everyday practice. The agency studies research and practice activities that do a number of things: one, improves quality of care; two, ensures safety and efficiency in health care. So AHQR studies the experience of Americans with its multiple databases, including the MEPS, Medical Expandable database, its hospital administration database, and its consumer assessment of health plans database.

I think AHRQ can contribute in several ways and it's interested in several ways. One is in the evidence base that supports practice and technology, the use of practice and technology. This information can guide purchasers, providers and consumers. It's also interested in studying and supporting how providers integrate diagnostic and therapeutic technologies in everyday practice. A third area of interest for the agency is in access to care and quality improvement. Thanks.

DR. McCABE: Thank you.

Next is Centers for Disease Control and Prevention.

DR. KHOURY: Good morning. My name is Muin Khoury. I'm the Director of the CDC Office of Genomics and Disease Prevention. I'm a pediatrician by training, medical geneticist, epidemiologist, trained specifically in genetic epidemiology at the time when the field was almost being born or non-existent. I joined the CDC in 1986 and I've been there ever since. I've been fascinated by the work of public health and disease prevention that the CDC does in collaboration with many groups.

CDC, as you may know, is the nation's prevention agency. We do a number of activities in terms of surveillance, investigating health outcomes and disease outbreaks, and having a pulse on the health of the nation with the idea of implementing and evaluating prevention activities that work in real communities.

In 1997, the CDC created the Office of Genomics and Disease Prevention in response to the Human Genome Project to figure out how to integrate advances in genomics into public health research policy and practice. I have been very lucky to be part of the initial creation of this office and be part of the whole movement at CDC.

We have a few priorities I'd like to mention briefly. The first is the evaluation for what genes mean in terms of health and disease in real communities in real time, sort of what do you do with a gene when you find one and how it impacts the health of the public in a real community. We have many challenges there, including the integration of genomics and the acute public health response in terms of epidemics and clusters and generally the health of the public.

We also have a challenge to evaluate, from a population perspective and with prevention in mind, how genetic information can be used to improve health and prevent disease both in terms of screening as well as everyday health care for the purpose of prevention.

Finally, we do spend quite a bit of energy working with our partners in state and local public health in terms of beefing up the public health capacity to respond, developing the workforce, and also funding and sponsoring different public health activities in this regard, and we also work with schools of public health and all the federal agencies that are represented here. I'm glad to be part of this.

DR. McCABE: Thank you.

Food and Drug Administration.

DR. FEIGAL: Good morning. I'm David Feigal. I'm an internist by training. I've been at the Food and Drug Administration for about 11 years in the Center for Drugs, the Center for Biologics, and I'm currently the head of the Center for Devices. I'm here actually representing the entire FDA. The types of products and issues that we have with genetic testing is quite broad-ranging. It includes genetically guided therapies, gene therapies themselves and, as was a particular focus of the previous committee, diagnostic devices.
The consumer protections that we are responsible for is to ensure that investigational use of products are safe, that market approval is based on an evidence standard set in statute that products be safe and effective or equivalent to products that are already marketed, we have the responsibility for inspecting and ensuring manufacturing quality, and monitoring problems in the marketplace in the use of the test or drug, such as adverse experiences. We're part of a system that provides the corrective actions when there are problems, along with the manufacturers, including recalls of products, safety alerts and changes in labeling.

The basic scheme of FDA is risk-based consumer protection, and part of the concerns of the previous committee in the debate has been to think about which are the riskier kinds of information, what are the riskier types of products. It's also a complex area because these are consumer protections that are shared with CMS, which is responsible for administration of the Clinical Laboratories Improvement Act, the CLIA program, which supervises laboratories. Our authorities and responsibilities overlap with states which license laboratories and license health professions.

Let me just conclude by thanking you in advance for all of your efforts. These are difficult issues and we all have a stake in this.

DR. McCABE: Thank you.

The Health Resources and Services Administration?

DR. FEETHAM: I'm Suzanne Feetham and I'm briefly sitting in for Dr. Sam Shekar. I am a nurse with clinical practice with children with birth defects and their families. I was Deputy Director at NINR, the National Institute of Nursing Research, at NIH. My program of research has included studies of families' decision-making in families considering genetic testing when they have susceptibility for cancer, and families of children with genetic conditions. I also have published extensively in genetics education in nursing and families and genetics.

Dr. Shekar is the Associate Administrator for the Bureau for Primary Health Care, and he leads the HRSA Genetic Work Group to provide recommendations to the agency on the activities for the Agency in Genetics. We are the access agency and the programs of HRSA focus on providing health care to underserved and vulnerable populations to move towards eliminating health disparities.

HRSA has recognized the significance of genomics for health and the understanding of mechanisms of disease and prevention. We feel that access and quality of care is affected by the ability of health professionals to apply genetic knowledge to their practice. We partner with several of our other federal agencies -- NIH, AHQR, CDC and others -- and also with professional organizations in the programs that we fund in regards to genetics.

Our genetics cover three key areas. The first is our well-known Maternal Child Health Bureau for the Genetic Services Branch, which provides programs for genetic testing and counseling, information development, and dissemination programs related to hemophilia, screening of newborns for sickle cell anemia, and genetic disorders and follow-up. That bureau also focused on programs in public education.

We also have a major program in our Bureau of Health Professions for the education of health and public health professionals. Since 1996 to '01, the funding in this area has increased five-fold in our recognition of the significance of genetic education to health professionals.

Another area of significance for HRSA is that our National Center for Workforce Analysis conducted a study of the genetic counseling workforce and is currently funding a study that is being done in regards to the genetic workforce in primary care, including nurses and physicians, genetic specialists, in partnership with the Maternal Child Health Bureau and our colleagues at NIH, the Human Genome Institute.

Our agency has significant networks that can facilitate the dissemination of genetic knowledge to help professionals. Our programs of HRSA and our commitment to this area can inform the work of this committee, and we are pleased to be part of this as an ex officio. Thank you.

DR. McCABE: Thank you.

National Institutes of Health?

DR. COLLINS: Good morning. I'm Francis Collins, Director of the National Human Genome Research Institute at the National Institutes of Health. I first want to say what a wonderful group we have here in the mosh pit --

(Laughter.)

DR. COLLINS: -- this very dedicated and talented group of members of this committee that I'm really looking forward to hearing from and working with, because I think we have a very exciting opportunity and a challenging one in front of us, with a whole host of topics that I'm sure we'll be wrestling with in the coming days, weeks and months.

The National Institutes of Health is the government's most major investment in biomedical research, with a budget approaching $30 billion a year, research which is carried out now on a host of fronts, from very basic science-oriented enterprises to very translational research, up to and including clinical trials. So this is a very major area of interest for the NIH, to make sure that all that research ends up being applied in a fashion that has maximal public benefit.

The NIH is constituted of some 27 institutes and centers, of which the Genome Institute is but one. I can tell you that all of the other institutes have considerable areas of interest in the purview of this particular committee and have already provided input to me as the NIH representative that I hope will be valuable in informing the committee of NIH's concerns and hopes.

For myself, I'm a physician. I'm trained as an internist and a medical geneticist. My career has been initially in an academic environment at the University of Michigan but for the last 10 years as an institute director at NIH leading the Human Genome Project, with a particular focus on trying to understand the causes and ultimately the cures of diseases that have genetic components, which is in fact virtually all diseases.

I will tell you one other hat that I wear, which is as the chairperson of the National Coalition for Health Professional Education in Genetics, an organization which currently involves some 120-member organizations which collectively is trying to achieve education of health care professionals in the principles and practice of genetics in anticipation that this is going to be a discipline which spills out, and in fact already is, into the mainstream of medicine and for which all health care providers will need to be prepared.
Finally, I'd just like to say that in terms of input to this committee's deliberations, I hope that I can be of some help by the fact that we do have a wonderful wealth of scholarship in many of the issues that you may want to consider, and that comes from research that's been supported, particularly from the ELSI program, the Ethical, Legal and Social Implications program of the Genome Institute, which has been in place since 1990 and which has investigated in rigorous, scholarly ways many of the topics that I think this committee may wish to deliberate on, and happily that means that we do have some evidence upon which perhaps to base some of our discussions. That ELSI program continues to be a critical part of our hopes for the future, with many areas of research bubbling up on an almost weekly basis as new developments occur in this rapidly advancing field.

So again, I'm delighted to be able to be a part of this as a liaison from NIH and looking forward very much to working with all of you and with the chair, the very capable Ed McCabe. Thank you.

DR. McCABE: Thank you.

Office for Civil Rights?

DR. FROHBOESE: Good morning. My name is Robinsue Frohboese. I'm the Principal Deputy Director of our Office for Civil Rights at HHS. I'm very pleased to be able to join you and to bring the perspectives of the Office for Civil Rights. I'm an attorney and a psychologist and actually have a longstanding interest and history in medical ethics issues dating back, like Marty, to the days that I spent on Capitol Hill in the early 1980s when I worked for the Senate Health Committee and was involved with a number of issues around medical ethics.

In my capacity of representing the Office for Civil Rights, I will be bringing to the committee two key important perspectives. The first perspective has been mentioned by a number of the members, and that is ensuring equal access by vulnerable populations and minority groups, eliminating health disparities, and also ensuring non-discrimination on the basis of race, color, national origin, disability, and age, all of which our office is responsible for in HHS-funded programs.

In addition to the traditional civil rights perspective, our office is also responsible for the new privacy rule under the Health Insurance Portability and Accountability Act, which many of you may know went into effect two months ago in April of this year. There are a number of considerations in protecting the privacy of information, including genetic information, and I will be happy to share our perspectives about the rule and to offer guidance about our interpretation and enforcement activities. Thank you.

DR. McCABE: Thank you.

Department of Justice?

DR. MAJIDI: Good morning. I'm Vahid Majidi. I have my degree in chemistry, and I started my career as a professor at the University of Kentucky. I was there for about eight years, after which I went to Los Alamos National Laboratory. Currently, I'm on detail from Los Alamos, and I'm the Chief Science Advisor at Department of Justice. We have a very broad interest in genetics information as it relates to forensic science, as well as all legal aspects of genetic information, genomic technologies, and genetic materials. Thank you.

DR. McCABE: Thank you.

And next is Department of Labor.

MR. ZURAWSKI: Hi. I'm Paul Zurawski, the Deputy Assistant Secretary at the Employee Benefits Security Administration, the agency at the Department of Labor which has jurisdiction over employer-provided benefit plans, including health care. We currently have the regulatory and enforcement responsibilities of HIPAA, at least in terms of the preexisting condition, portability and non-discrimination provisions as they relate to group health plans. Thanks.

DR. McCABE: Thank you.

Equal Employment Opportunity Commission.

MR. MILLER: Thank you. My name is Paul Miller and I am a Commissioner at the U.S. Equal Employment Opportunity Commission. The EEOC enforces all federal workplace discrimination laws

prohibiting discrimination on the basis of race, gender, age, religion, disability, and pregnancy. The EEOC intersects with these issues of genetics in terms of prohibiting and thinking about employment discrimination, particularly genetic discrimination. Most of the bills I think that Dr. McCabe referred to earlier, genetic discrimination bills, foresee an enforcement mechanism through the EEOC, and the EEOC has also litigated the first case of genetic discrimination against a railroad.

My background, I've served on the working group that drafted the Executive Order on Genetic Discrimination and Privacy. I've previously served as a member of the President's Committee on People with Disabilities and was a White House staffer on disability issues, a lawyer by training. It's a pleasure to be here. Thank you.

DR. McCABE: Thank you.

Also, within DHHS, we have the Centers for Medicare and Medicaid Services.

DR. TUNIS: My name is Sean Tunis. I'm the Chief Medical Officer for Medicare and Medicaid Programs, and also the Director of the Office of Clinical Standards and Quality. The perspectives that I bring and will be listening with are both related to the coverage policy, the reasonable and necessary determinations of new technologies for the Medicare program, reimbursement policy that goes along with that, and also the agency shares oversight of the CLIA program with the FDA. So we have a component of CMS that has that expertise. Actually, the primary person with knowledge of that person, Judy Yost, is not here today but she'll be joining us at future meetings.

I'm also invited to these meetings to make sure David Feigal stays within the boundaries of sense.

(Laughter.)

DR. McCABE: Thank you.

Well, I think that one of the things that we all observed is that the breadth of agencies who are involved with this committee is extremely broad, and I think it represents the interests of both the government and our population more broadly in what has come from genetics and the Human Genome Project. So I think it's really wonderful to have so many representatives of the various government agencies here and involved with this committee.

Most important to this committee's function is really the staff of that committee, and we are incredibly lucky that Sarah Carr, who was the Executive Secretary for the Secretary's Advisory Committee on Genetic Testing, is also the Executive Secretary for this committee. Anything that goes wrong I will take full credit for, but I can tell you I won't have too many things that I have to take credit for because Sarah and her staff do such a wonderful job. So I think we should have Sarah introduce herself, since she's really the key person for the success of this committee.

MS. CARR: Well, thank you, Ed. Again, my name is Sarah Carr, and I'm called the Executive Secretary of this committee, but I'm part of a team at NIH that helps staff you. It includes Lana Skirboll, Amy Patterson, Suzanne Goodwin, and this summer Olivia Hess, our summer intern. The other role I have when I'm sitting here with you is to be the designated federal officer, and I'm supposed to keep you from doing anything wrong, and I have great faith in all of you that I won't have a very hard time making sure that you don't do anything wrong.

So it's a pleasure to be with you, it's a pleasure to see this committee start, and we look forward to staffing you and helping you navigate these challenging waters.

DR. McCABE: With typical modesty, Sarah will not tell you of all the other responsibilities that she has. We are not her only responsibility.

Before we go on, I want to take a moment to review the rest of the agenda with you and explain the principal goals of the meeting. Our main objective is to identify and to prioritize issues that the committee will address and develop as a workplan for addressing these issues. We do have an extremely broad range of opportunities. Our challenge will be to identify one or two opportunities over this meeting and the next meeting in priority as our initial areas on which we will focus.

The informational presentations that we've scheduled for today and tomorrow morning are preparatory to our priority-setting deliberations. They're intended to provide the committee with a common foundation from which to consider and prioritize these issues. We will have background presentations on key issues from some of the nation's leading experts in their fields. We will hear perspectives from the public, we will review the work of our predecessor, the Secretary's Advisory Committee on Genetic Testing, and finally we will be briefed about what the federal agencies represented on the committee think are their highest priority issues.

So with that, let us move on, and I want to thank all of you for your informational introductions and also with the brevity of those introductions so we could move on with the work at hand.