Secretary's Advisory Committee on Genetics, Health, and Society
Summary of Fifteenth Meeting
February 12-13, 2008
Bethesda, Maryland

Committee Members Present

Reed Tuckson, M.D., Chair, Day 1
Steven Teutsch, M.D., M.P.H., Chair, Day 2
Mara Aspinall, M.B.A. (appointment pending)
Sylvia Mann Au, M.S., C.G.C.
Paul Billings, M.D., Ph.D., FACP, FACMG (appointment pending)
James P. Evans, M.D., Ph.D.
Andrea Ferreira-Gonzalez, Ph.D.
Kevin FitzGerald, S.J., Ph.D., Ph.D.
Julio Licinio, M.D.
Barbara Burns McGrath, R.N., Ph.D.
Paul Steven Miller, J.D. (appointment pending)
Joseph Telfair, Dr.P.H., M.S.W., M.P.H.
Marc S. Williams, M.D., FAAP, FACMG
Paul Wise, M.D., M.P.H. (appointment pending)

***Ex Officios*/Alternates Present**

Gurvaneet Randhawa, M.D., M.P.H. (HHS/Agency for Healthcare Research and Quality)
Muin J. Khoury, M.D., Ph.D. (HHS/Centers for Disease Control and Prevention)
Judith A. Yost, M.D. (HHS/Centers for Medicare & Medicaid Services)
Steven Gutman, M.D., M.B.A. (HHS/Food and Drug Administration)
Denise Geolot, Ph.D., R.N., FAAN (HHS/Health Resources and Services Administration)
Phyllis Frost, M.D. (HHS/National Institutes of Health)
Robinsue Frohboese, J.D., Ph.D. (HHS/Office for Civil Rights)
Michael Carome, M.D. (HHS/Office for Human Research Protections)
Inyang Isong, M.D., M.P.H. (HHS/Office on Public Health and Science)
Martin Dannenfelser (Administration for Children and Families)
Michael Amos, Ph.D. (Department of Commerce)
Scott McLean, MC, USA (Department of Defense)
Peter T. Kirchner, M.D. (Department of Energy)
Sherrie Hans, M.D. (Department of Veterans Affairs)
Matthew Daynard, J.D. (Federal Trade Commission)

Executive Secretary

Sarah Carr, NIH Office of Biotechnology Activities

FEBRUARY 12, 2008

Welcome and Opening Remarks

Reed V. Tuckson, M.D.
SACGHS Chair

Dr. Reed Tuckson, Chair of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), welcomed those in attendance and stated that the public was made aware of the meeting through notices in the Federal Register and announcements on the SACGHS website and listserv. He noted that there were technical problems with the video portion of the live webcast. Dr. Tuckson stated that his appointment as Chair of the Committee was ending that day. The Secretary of Health and Human Services (HHS) selected Dr. Steven Teutsch as Dr. Tuckson's successor, and Dr. Teutsch would transition into his new role on day 2 of the meeting.

Dr. Tuckson reviewed the strategic plan of the Committee and provided an overview of the agenda. He stated that in June 2004, SACGHS developed a resolution about the importance of educating and training health professionals in genetics. After a roundtable session was held on this topic at the November 2007 meeting, it became apparent that there were still critical needs in education and training. The Genetics Education and Training Task Force was therefore created, chaired by Dr. Barbara Burns McGrath, who will present a draft charge for the Task Force on day 2.

Dr. Tuckson explained that since the SACGHS Report on Coverage and Reimbursement of Genetic Tests was written, several technical developments took place that affected the Medicare billing options available to genetic counselors. In light of these developments, he recommended that the Committee ask the Secretary to clarify these billing options, as legislative action might be required to remedy the situation. Dr. Tuckson stated that a draft letter addressing the issues would be distributed for Committee review during the meeting, and asked that suggested changes be submitted to staff member Suzanne Goodwin. He noted that the final Pharmacogenomics (PGx) Report was planned for delivery to the Secretary in March 2008 and would be made available to the public 30 days later.

Dr. Tuckson informed the Committee that as part of the Personalized Healthcare Initiative, the Secretary's Office was organizing an informal workgroup with representatives from HHS agencies and the Federal Trade Commission (FTC) to explore direct-to-consumer (DTC) genetic testing services. The Personalized Healthcare Initiative Workgroup would discuss the roles and responsibilities of Federal agencies in DTC marketing and performances of genetic tests, the challenges associated with communication of complex genetic information to the public, and assess the services offered by various companies engaged in DTC marketing, including the quality of information provided and confidentiality provisions.

In March 2007, SACGHS was asked to respond to a series of questions posed by the Office of the Secretary (OS) on the adequacy of the oversight system for genetic testing. A 33-member Task Force, chaired by Dr. Andrea Ferreira-Gonzalezwas formed to develop a report in response. The draft report was released for public comment from November 5 through December 2, 2007. This effort resulted in 64 sets of public comments, which were summarized in the briefing book. Dr. Tuckson stated that the agenda on day 1 and part of day 2 would focus on the draft recommendations on oversight. The first goal for the session was to finalize the recommendations for submission to the Secretary at the end of February 2008. The second goal was to receive approval on the content of the final report, so that it could be edited and transmitted to the Secretary in April 2008. Dr. Tuckson stated that a public comment period would take

place prior to the Committee's discussion, so that Committee's deliberations could benefit from the public's perspective.

Executive Secretary Sarah Carr reviewed the Committee's ethical responsibilities. Dr. Tuckson turned the floor over for public comment. He introduced Paul Radensky from the Coalition for 21st Century Medicine.

Public Comments

Paul Radensky, M.D.
Coalition for 21st Century Medicine
McDermott, Will, & Emory

Dr. Radensky stated that he worked for the law firm McDermott, Will, & Emory, which served as counsel to the Coalition for 21st Century Medicine. The purpose of the Coalition was to develop workable solutions to support public health concerns about appropriate oversight for new technologies concerning in vitro diagnostic multivariate index assays (IVDMIAs) and analyte specific reagents (ASRs), as well as to provide incentives to continue development. Members included laboratories that developed laboratory developed tests (LDTs) and manufacturers of ASRs. Dr. Radensky focused his comments on a proposal developed in response to the Federal Drug Administration's (FDA) draft guidance on IVDMIAs, which he believed was related to the recommendations in the Oversight Report.

The Coalition identified concerns in the draft FDA guidances issued in September 2006 and July 2007. These concerns included transparency, the "fox guarding the henhouse," risk-based regulation, clear definitions, clear and predictable pathways, a transition timeline, and continued incentives toward innovation. With those principles in mind, the Coalition developed a proposal that would involve creation of a registry over a 3- to 5-year period that would be publicly available and would include a role for FDA to review and comment on any claims made. They believed this would allow for an independent review of the validity of data supporting the claims and gather evidence to help determine the appropriate level of oversight.

Dr. Tuckson thanked Dr. Radensky and introduced Jeff Kant from the College of American Pathologists (CAP).

Jeffrey Kant, M.D., Ph.D.
College of American Pathologists, Professor of Pathology and Human Genetics
Director of the Division of Molecular Diagnostics
University of Pittsburgh Medical Center

Dr. Kant spoke on behalf of the College of American Pathologists (CAP), where he chaired a resource committee on proficiency testing (PT) programs in genetics. CAP is a national medical specialty society representing more than 17,000 pathologists who practice anatomic pathology and laboratory medicine in laboratories worldwide. CAP's Commission on Laboratory Accreditation accredits more than 6,000 laboratories in the United States and abroad and has been a leader in developing quality improvement programs for laboratories, including programs in genetic testing. Dr. Kant stated that laboratorians have some of the strongest measures of quality in medical practice. CAP's experience from its PT and laboratory accreditation program is that the overwhelming majority of mainstream genetic tests performed in the United States are safe and effective. CAP's laboratory accreditation program stresses both analytic and clinical validation prior to introducing any test into practice, recognizing that tests will continue to be

improved periodically after introduction, with each improvement revalidated by the laboratory before patient samples are sent. CAP has a keen interest in ensuring that their ability to provide high quality diagnostic services to patients and physicians is not comprised by overly burdensome regulation. They recommended that changes to Federal oversight of laboratory tests be made within the context of the Clinical Laboratory Improvement Amendments (CLIA). CAP supported further enhancement of laboratory testing through educational efforts, improvement in the quality of CLIA inspections, and additional Federal resources for access to controls and standards. CAP agreed with SACGHS that appropriate resources should be directed to the Center for Medicare and Medicaid Services (CMS) for required oversight of CLIA, and that PT should be expanded. CLIA already requires assessment of analytic validity for all assays offered by a laboratory, requires knowledge of the clinical utility of tests for use in routine clinical practice, and stipulates qualifications and responsibilities of the laboratory to patients. Dr. Kant stated that requiring FDA approval for every LDT would result in numerous unintended consequences that would not benefit patients, but would delay implementation of new tests, reduce innovation, increase costs, and limit access to beneficial assays. CAP also supported the emphasis in the draft report on public-private partnerships for assessment of LDTs. Registration of genetic tests through such partnerships could have a positive impact, but he said that the system should be voluntary and created with broad stakeholder input. Since CLIA already requires submission of test lists by laboratories as a condition of inspection, additional information submitted should remain within the context of CLIA and CMS. New mechanisms for the collection of information should be tested before implementation to ensure that the most useful information has been captured and that submission is not overly burdensome for laboratories. This information could be made publicly available, assuring clinicians and patients of the analytic and clinical validity of the tests they are ordering, while not impeding the medical practice of CAP.

Dr. Tuckson thanked Dr. Kant and introduced Mr. David Mongillo from the American Clinical Laboratory Association (ACLA).

David Mongillo, M.P.H.
Vice President for Policy and Medical Affairs
American Clinical Laboratory Association

Mr. Mongillo focused his comments on Recommendation 4 in Chapter 4 of the draft Oversight Report, stating that it could have unintended consequences if not carefully communicated to the Secretary. The recommendation said that SACGHS supported FDA regulation of LDTs and the flexible, risk-based approach the agency was taking to prioritize them, stating that the approach should be robust enough to accommodate new genetic testing technologies and methodology. Mr. Mongillo said that if the recommendation was interpreted to mean that FDA's Food, Drug, and Cosmetic Act requirement should be applied to LDTs without interagency coordination, needless duplication of effort would result. He stated that FDA and CMS should coordinate and streamline their quality validation procedures, which currently had separate inspections, separate quality system requirements, separate reporting and labeling requirements, and additional requirements for design control, corrective action, and prevention. Mr. Mongillo said it was premature for SACGHS to definitively support FDA regulation of LDTs without recognizing the important first step of interagency coordination and requirement harmonization. He stated that ACLA and others had proposed regulatory models that would build on interagency coordination, be consistent with principles of least burdensome regulation, fill regulatory gaps, avoid overlapping and potentially conflicting regulatory oversight, and allow for a participatory approach that would draw on the expertise of industry stakeholders, CMS, and FDA. By employing public-private partnerships, these models would avoid significant new costs for Federal agencies. Mr. Mongillo asked that SACGHS revise

Recommendation 4, Chapter 4, by adding the words, "an interagency role for FDA," and "CMS's regulation of LDTs."

Dr. Tuckson thanked Mr. Mongillo and introduced Suzanne Feetham from the American Academy of Nursing.

Suzanne Feetham, Ph.D., RN, FAAN
American Academy of Nursing

Dr. Feetham commented on the draft Oversight Report on behalf of the American Academy of Nursing and the Genetic Healthcare Expert Panel of the Academy. The American Academy of Nursing comprises more than 1,500 top nursing leaders and is constituted to anticipate national and international trends in health care and address resulting issues of health care knowledge and policy. Ms. Feetham said the Academy was concerned about the decision of CMS not to create a genetic testing specialty and associated PT, a reversal of their previous position. The Acacemy strongly supported establishing a genetic testing specialty and associated PT for all laboratories performing genetic tests. Ms. Feetham asked that SACGHS recommend that CMS take action to establish a required minimum degree of quality for any laboratory performing genetic tests and conduct further study on the issue of performance assessment while instituting genetic-specific PT. The Academy was also concerned that the Committee had not recommended that HHS allocate resources to address these knowledge deficiencies. The Academy recommended an adjustment in education strategies for all health care providers, away from traditional education approaches in schools to a focus on system and practice change. Ms. Feetham stated that evidence of knowledge embedded into practice should become a component of every patient record for hospital and institution accreditation. She said that if there was evidence of the application of genetics and genomics in practice, regulators would be influenced to include the expectation of this knowledge for all health care providers in licensing and accreditation. To facilitate a shift of the education focus to practice, Ms. Feetham said SACGHS could invite the representatives of accrediting bodies to a meeting, including the Joint Commission and Health Facilities Accreditation Program. She stated that the Committee's recommendations on communication and clinical support would not be realized without the foundation of an adequate health care practitioner knowledge base and the number of health care providers with genetic expertise was not sufficient to support best genetic test practices. Ms. Feetham said many clinically available tests (such as Oncotype DX, a multiple-gene assay performed on early stage breast cancer tumors where standards of practice for utilization support are in the domain of the oncology specialist) are supported by practitioners other than genetics experts.

Dr. Marc Williams, a member of the Education and Training Task Force, said SACGHS would address the issues Ms. Feetham raised through that Task Force, and noted that the Oversight Report referenced the intensified effort of SACGHS in addressing education and training.

Dr. Tuckson thanked Ms. Feetham and introduced Dr. Peter Lurie.

Peter Lurie, M.D., M.P.H.
Deputy Director, Health Research Group of Public Citizens

Dr. Lurie stated that he was Deputy Director of the Health Research Group of Public Citizens, an advocacy group in Washington D.C.. They take no money from either Government or industry. He said that from the patient perspective, there is no distinction between a genetic test and any other kind of laboratory test. He also said that the public does not understand the regulatory framework and assumes that the amount of regulatory oversight associated with all tests is equal. He stated that a form of genetic

exceptionalism was taking place, because the vast majority of genetic tests were barely regulated, while the vast majority of other tests fall under the purview of FDA. He said patients are owed that amount of equality and comprehensiveness in oversight. Dr. Lurie said the voices of consumers had not come before the Committee to a significant degree. He noted that all 33 members of the Oversight Task Force were from Government, academia, or industry; and only two comments submitted to the record were from consumer or advocacy groups. He said it was notable that many professional for-profit groups disagreed with the thrust of the Task Force's recommendations. Dr. Lurie described his disagreements with the Task Force's recommendations, beginning with the failure to endorse a genetic testing specialty. He also said that despite well documented reasons for expanding FDA regulations and problems with current FDA oversight, the draft report endorsed the status quo, rather than recommending vigorous FDA oversight. He disagreed with the recommendation for a voluntary registry, stating that there had been a voluntary system for 14 years, i.e., GeneTests, which he said had many deficiencies.

Dr. Tuckson thanked Dr. Lurie and introduced Mark Sobel from the Association of Pathology Chairs.

Mark Sobel, M.D., Ph.D.
Managing Officer, Association of Pathology Chairs

Dr. Sobel stated that the Association of Pathology Chairs (APC) represents departments of pathology and laboratory medicine in all accredited medical schools in the United States and Canada. He made several points in response to the draft Oversight Report. Dr. Sobel said SACGHS used a very broad definition of genetic test, going beyond heritable changes to include somatic variations, and going beyond DNA and RNA to include proteins and other analytes. Under this definition, he said the tests referred to should more accurately be called molecular tests, not genetic tests. He said the Committee needed to define the differences between genetic and genomic applications and which intended uses were included in the proposed oversight of genetic testing. He noted that the report concluded that genetic tests should not be considered as significantly different from other clinical tests, and the APC agreed with this perspective. Given this position, however, he said it was unclear why genetic tests were proposed as requiring greater oversight than non-genetic tests that are similarly molecular, laboratory developed, complex, and potentially high risk. Although tests for heritable diseases are unique in several respects, at the technical level, the diagnosis of genetic disease by molecular methods does not differ significantly from the techniques used to diagnose infectious diseases and neoplastic diseases. Therefore, he said it was not logical to establish more stringent technical and personnel standards for molecular genetic testing than already exists, including molecular oncology and molecular microbiology testing.

Dr. Sobel said that Dr. Kant's statement on quality assurance and CLIA versus FDA regulations adequately expressed the opinion of APC, i.e., that further regulation by FDA would be inappropriate, could be duplicative, and could have unforeseen consequences, such as delaying innovation. Concerning the system for test registration, the APC endorsed the Committee's recommendation to develop a public-private partnership for voluntary registration of tests.

Dr. Tuckson thanked Dr. Sobel and introduced Linda Avey of 23andMe, who was addressing the Committee by phone.

Linda Avey
Co-Founder, 23andMe

Ms. Avey stated that 23andMe is a private company in California that enables people to access their genetic information through the use of research tools being used by laboratories across the country and

across the world. They use large-scale genotyping microarrays to give people information and they provide a context for it, so that people understand new findings coming out of the research community. 23andMe was founded on the premise that individuals have the right to access their own genetic information and know what their bodies are made of without having to pay for the services of a health care professional. Ms. Avey stated that consumers cope with risk-based information every day, and history shows that fears about how consumers will respond to information are usually overblown and inaccurate. She stated that Federal and State governments, as well as physicians, should not impede information development and dissemination based on an old-fashioned and paternalistic view of what the average person can and cannot understand. The company was developing a way for consumers to engage in a new research effort, called consumer-enabled research. They believed progress in genetic research would be greatly enhanced by the development of a large database of genetic and phenotypic information contributed voluntarily by interested individuals. Ms. Avey emphasized that the focus of 23andMe was research.

In response to a question from Dr. Jim Evans about the company website, Ms. Avey stated that a white paper on the website explained the process their scientists go through before reporting on any particular finding. The focus was on common diseases that are multigenic, not Mendelian disorders.

Dr. Muin Khoury stated that he co-authored an article in the *New England Journal of Medicine* in January 2008 about the premature use of these kinds of research tools, but he appreciated the fact that the company was trying to educate consumers, rather than sell genetic tests. However, he noted that if these genetic tests were to be offered for prevention of disease or health promotion, they would not pass the tests of analytic validity, clinical validity, or clinical utility. He asked Ms. Avey to clarify the distinction between an educational tool versus a tool that could be offered for health purposes. Ms. Avey said she agreed with the points made in Dr. Khoury's article. She stated that the company was collecting information from their customers and explaining to them that they were participating in research. She cited the Framingham Study as a model. 23andMe wanted to move the concept of the prospective long-term study to the Internet and into a social networking capability in which people could share information directly and dynamically. Ms. Avey said they welcomed oversight and were eager to receive input on their efforts from the medical and research communities. She agreed to speak to the Committee again and provide more detailed information.

Dr. Tuckson thanked Ms. Avey and introduced Dr. Michael Watson from the American College of Medical Genetics (ACMG).

Michael Watson, Ph.D.
Founder, American College of Medical Genetics

Dr. Watson represented ACMG, an organization that bridges laboratory testing and the clinicians who deliver genetic tests to the population. He co-chaired the Secretary's Advisory Committee on Genetic Testing (SACGT) in 1995 and was not certain that much progress had been made since then. He suggested a careful look at the factors that were impeding progress. He noted that no one group is really well placed to deal with all of genetic testing. He also pointed out the large number of tests available. Dr. Watson stated that the complexity of different types of tests makes it difficult to determine their clinical validity. He said the vast majority of genetic tests are for rare diseases, which means there is little incentive for studies in the marketplace that would lead to development. It is therefore left to the laboratories to develop tests, if they want to be accessible to the patient population. However, laboratories are not in a strong position or well resourced enough to lay out guidelines and clinical validity at a general population level. There is tremendous diversity in the tests they offer, both analytically and clinically. Dr.

Watson also described the tremendous variation among different populations, which makes genetic testing additionally complex. He said the best way to approach these problems, particularly a registry, is through a public-private partnership. He stated that a registry should have more information than a listing of what people are selling in their laboratories around the country. It should also have information about why tests are clinically valid in particular clinical situations. Such a venture would require the participation of all interest groups and would be very expensive. Key decisions would have to be made on how to find resources for the registry, who the participants would be, and how it would be organized. Dr. Watson said ACMG would be willing to work with the Committee on the registry.

Dr. Watson said CLIA should be able to address analytical validity through inspection, specifically PT. He stated that an FDA rule on the analytical side of laboratory developed tests would not translate directly to the clinical laboratory environment, and therefore, would not be of much help. He noted that an FDA evaluation of a genetic test provides clinical plausibility, but the agency is not in a position to say that a payer should pay for a specific test. Dr. Watson said clinical utility is valuable, but genetic tests often do not have the level of statistical power desirable for a clinical utility analysis. Testing in large populations requires clinical utility, but Dr. Watson stated that in the rare disease world, it is difficult to get beyond the utility of an etiological diagnosis in the test itself. He said that ACMG requested that GeneTests send their entire library to them. It was then built in to a complete Access file of every test and gene available in the database, with clinical validity for the various intended uses of those tests as the first goal. Dr. Watson said clinical validity is not easily constrained by a regulatory perspective, because that perspective has many exemptions for the practice of medicine.

Dr. Tuckson thanked Dr. Watson and introduced Emma Kurnat-Thoma from the International Society of Nurses in Genetics (ISONG).

Emma Kurnat-Thoma, M.S., RN
International Society of Nurses in Genetics

Ms. Kurnat-Thoma said ISONG is a global organization dedicated to fostering the scientific and professional growth of nurses in genetics and genomics. She stated that ISONG supported the Committee's recommendation to enhance interagency coordination of genetic testing oversight, especially the development of steps to foster resources, education, and knowledge. She highlighted four considerations related to analytic validity and clinical validity. ISONG disagreed with the Committee's conclusion that gaps could be identified and addressed without the creation of a genetic testing specialty. Second, ISONG was aware of gaps in the extent to which clinical validity could be generated and evaluated for genetic tests. They supported the recommendation to create public resources and recognized that the American public would be best-served if diverse ethnic, racial, and geographic subgroups were represented. Third, ISONG disagreed with the recommendation to establish voluntary genetic testing registration to reduce system gaps and improve oversight. They felt this approach would not be sufficient, given gaps in the enforcement of existing regulations. Fourth, ISONG applauded the Committee's concern regarding certain types of health-related genetic tests marketed directly to consumers and agreed that there was insufficient oversight of the laboratories that develop them.

Ms. Kurnat-Thoma also stated that ISONG fully supported the idea of HHS collaboration with relevant agencies and private parties. They believe genetic expertise is essential when providing and interpreting genetic tests. In closing, she said that as the largest body of health care providers, nurses have continual and close contact with patients and could intercede to prevent and/or reduce the public harms that might result from DTC genetic tests.

Dr. Tuckson thanked Ms. Kurnat-Thoma and introduced Dr. Michelle Schoonmaker from the Association of Molecular Pathology (AMP).

Michelle Schoonmaker, Ph.D.
Association of Molecular Pathology

Dr. Schoonmaker summarized AMP's comments on the draft Oversight Report. She stated that the SACGHS definition of genetic test would be more accurate for a molecular test. AMP encouraged the Committee to define which intended uses are included in the oversight of genetic testing. Dr. Schoonmaker said AMP was concerned that certain types of genetic testing marketed directly to consumers fell outside the current regulatory oversight of CLIA, and hoped the Committee would further explore the issue of potential harm by health-related DTC marketing. She said CLIA regulations already stipulate the responsibilities of laboratory directors and clinical consultants, and AMP recommended that these roles be reemphasized with regard to genetic testing. They asked that the Committee modify Recommendation 1B by requesting that CMS work with professional organizations such as AMP to develop interpretive guidelines for their inspectors regarding the levels of expertise required for different kinds of genetic testing. Dr. Schoonmaker said AMP offered their expertise to help define the molecular targets that would be regulated analytes to promote expansion of PT programs for better oversight of DTC marketing of clinically dubious genetic tests and would assist in reassuring the public and Congress about the quality of genetic tests. She noted that voluntary consensus organizations such as the Clinical and Laboratory Standards Institute (CLSI) developed detailed practice guidelines that filled many holes in the FDA and CLIA regulatory framework. She stated that a team approach in which Government, industry, and practicing clinicians work together is a viable and desirable alternative to regulation for many genetic and genomic tests. AMP was concerned that registration of genetic tests would duplicate information already submitted to CMS as required under CLIA. The organization strongly supported CMS enhancement of the mandatory CLIA registration of nonwaived laboratories by strengthening CMS's infrastructure. AMP supported the proficiency survey programs available, with analytes added as necessary. They intended to publish best practices and laboratory and clinical practice guidelines, working with organizations such as CAP and ACMG. AMP strongly favored reliance on peer-reviewed literature; consensus statements by professional practice organizations; and collaborative studies by the CDC, other agencies, private investigators, and manufacturers to establish clinical validity. They supported integrated efforts to collect postmarket data to meet clinical, regulatory, and reimbursement goals. Dr. Schoonmaker stated that AMP was concerned that Recommendation 1.4 could result in a duplicative system of oversight for LDTs and the laboratories performing the tests. She closed by reiterating AMP's commitment to assist the Committee in their efforts.

Dr. Tuckson thanked Dr. Schoonmaker and introduced Ms. Sharon Terry of the Genetic Alliance.

Sharon Terry, M.A.
President and CEO, Genetic Alliance

Ms. Terry stated that the first steps in improving the oversight of genetic testing would be through enforcement of existing regulatory authority under the CLIA program; applying the available funding resources to provide for additional personnel, consultants, and training; and providing the mandated level of transparency of CLIA laboratories under the current statute. She said it was important to take action on the identified interim steps within the agencies' discretion and to implement the necessary steps for PT enhancements for genetic testing. Ms. Terry also stated that it was clear that mandatory genetic test registration was necessary to provide stakeholders with information that would improve the oversight of genetic tests. She said that making test performance characteristics and reference information publicly

available would increase confidence and improve the appropriate utilization of genetic tests. The Genetic Alliance believed the registry should be housed at and managed by a Federal agency, such as FDA or NIH. They agreed that more public resources should be committed to fill gaps and they supported the establishment of a laboratory-oriented consortium for sharing information on method validation, quality control, and performance issues. Such an undertaking should prioritize its goals based on clinical need, availability of information, and appropriate resource allocation. To maximize benefits and minimize harms, Ms. Terry stated that a public-private consortium of stakeholders should be created to assess the clinical utility of genetic tests, including the establishment of evidentiary standards and increasing the number of systematic reviews. She agreed with the SACGHS report's concern about FDA exerting regulatory authority over clinical decision aids. Ms. Terry said DTC access to testing must be carefully regulated to ensure public safety. The Genetic Alliance also believed that HHS should convene HHS agencies and interested stakeholders to provide further input into the development of a risk-based framework for the regulation of LDTs. Ms. Terry said HHS must require Federal agencies to work collaboratively and avoid turf battles.

Dr. Tuckson thanked Ms. Terry and closed the public comment session.

Session on Oversight of Genetic Testing

Overview
Summary of Public Comments on Draft Report and Goals of Session
Andrea Ferreira-Gonzalez, Ph.D., Chair of Oversight Task Force

Dr. Ferreira-Gonzalez provided an overview of the goals of the oversight session. She reviewed the charge given to the Committee by OS, the process used to draft the report, and the fact that the draft had gone out for public comment. She stated that the focus of the session would be to reach consensus on the report's recommendations and approve their transmission to the Secretary at the end of February. The Committee also would be asked to approve the spirit of the report's text, which would be edited following the meeting and sent to the Secretary on April 30, 2008.

The charge from OS asked for a comprehensive map of the steps needed for evidence development and oversight of genetics and genomic tests, with the improvement of health quality as a primary goal. The charge also tasked the Committee with evaluating existing pathways that examine analytical validity, clinical validity, and clinical utility; attributable harms if pathways were inadequate; and the roles and responsibilities of relevant Government agencies and private sector organizations. The Committee was asked to consider whether genetic tests are different from other laboratory tests for oversight purposes. The charge also addressed the adequacy and transparency of PT processes, communication pathways to guide the use of genetic testing, and new public and private sector approaches for demonstrating clinical validity and developing clinical utility. Finally, SACGHS was asked to consider whether additional or revised Government oversight would add value to the oversight system.

Drafting of report chapters began in May 2007, after several Task Force meetings were held to discuss the approach to the report. Face-to-face meetings were held in July and September 2007 to advance the report and develop draft recommendations. The draft report was released for public comment from November 5 through December 21, 2007. The Task Force received 64 sets of comments from a range of stakeholders; including professional organizations, industry, Government agencies, health care professionals, advocacy organizations, academicians, and individuals. Copies of the comments were sent to all Task Force members, with an initial analysis performed by the Task Force's Steering Group, which was composed of the six SACGHS members on the Task Force. In January 2008, weekly conference calls were held by the

Steering Group to discuss the public comments and any revisions needed to enhance the recommendations and report text. The full Task Force met via conference call to provide their perspectives on January 23. On January 30, a conference call was held with the full Committee to obtain its input.

Dr. Ferreira-Gonzalez stated that the overall tenor of the comments was very positive, as most commenters thought the report was responsive to the Secretary's charge. The following recurring themes were noted: several commenters were concerned about the report's broad definition of genetic test; there was strong support for increased PT and the development of standards and reference materials needed for PT; a mandatory rather than voluntary approach to a genetic tests registry was favored, but there was no clear indication where such a registry should be housed; commenters expressed concerns about DTC advertisers and consumer-initiated testing; FDA's authority to regulate laboratory tests was not questioned, although some comments criticized FDA's IVDMIA draft guidance; the importance of establishing clinical validity was affirmed; commenters called for more evidence and analysis of clinical utility and for increased education efforts; and the public asked that the costs, benefits, and harms to patients be considered before increasing oversight. The Task Force worked collaboratively to revise the report and recommendations in response to the public comments. Dr. Ferreira-Gonzalez said the Committee needed to discuss and finalize the recommendations and approve the final report in principle before the end of the meeting in order to meet the deadlines for delivery to the Secretary and the public. She introduced Dr. Clifford Goodman from The Lewin Group, a Federal contractor, who addressed the group via conference call. The Lewin Group had developed a comprehensive map of the oversight of genetic testing, with an accompanying document listing more than 30 gaps in oversight that were identified by the Task Force.

Presentation of the Comprehensive Map of Genetic Testing Oversight
Clifford Goodman, Ph.D.
Senior Vice President
The Lewin Group

Dr. Goodman stated that the comprehensive map consisted of five main sectors: research and development, CLIA-exempt States (e.g., New York and Washington), the CMS/CLIA pathway, the FDA pathway, and availability and reimbursement. He described each sector individually.

Research and development generally includes understanding gene-disease interactions, followed by basic research, prototype design, preclinical development, and clinical testing. Possible routes from clinical development include LDTs and commercial tests, including companion diagnostic tests, which are tests developed in parallel with drugs or biologics. Gaps in this sector included the need for standard reference materials for assay, analyte, and platform validation and lack of FDA premarket review of LDTs.

The second sector, CLIA-Exempt States, applied primarily to New York and Washington, and referred specifically to the oversight of laboratories. This sector had five main sections: PT, quality assurance, quality control, personnel standards, and reagent and equipment inspection. Dr. Goodman noted that an important aspect of oversight in these States is biennial inspections. States that do not fare well in the inspection process could lose their CLIA-exempt status or might not be able to offer certain tests. Identified gaps in this sector included the lack of resources and means to develop PT for all genetic tests and lack of data on the effectiveness of PT versus alternative assessment.

The CLIA Regulation sector included sections on personnel standards, quality assurance, quality control, analytical validity, and PT. Quality control includes inspection and survey requirements that can be

performed by CMS or its agents. CLIA accreditation can be achieved through meeting these requirements. A laboratory with CLIA accreditation can provide clinical testing. Biennial inspections are required, and if the laboratory does not meet its requirements, it could lose CLIA accreditation or might not be able to offer some tests. Biennial inspections provide important feedback information to the CLIA regulatory process and to the research and development process. Identified gaps in this sector included inadequacies in CLIA requirements for PT and that the analytical validity is reviewed after tests are on the market, not before.

The FDA sector of the map outlined FDA's oversight of commercial products that emerge from research and development. They typically come from device-makers of test kits and test systems, Co-developed therapeutics created in parallel with tests also require FDA review. Identified gaps included inadequate resources to review the analytical validity and clinical validity for many tests and clinical validity data may not be available at the time the tests are offered clinically.

The fifth and final sector of the map described the availability and use of genetic tests. Dr. Goodman said that a small number of tests are subject to clinical utility review. The U.S. Preventive Services Task Force (USPSTF) and the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) program provide this function for some tests. Reimbursement is carried out by Medicare, Medicaid, private insurance, the Veterans Administration (VA), the Department of Defense (DoD), and other entities. Another component of the sector included postmarket surveillance, which is done largely, but not entirely, by FDA. Some postmarket surveillance information is fed back to those responsible for reimbursement, because payers have an interest in what happens to tests once they are on the market. The postmarket data might affect coverage and payment decisions. Postmarket surveillance could also feed back to the research and development sector and to the FDA review process. For example, postmarket information might be used to revise product labeling of tests. Once a test becomes available for clinical use, various organizations in the public and private sectors conduct outcomes research. The findings can be fed back to clinicians and others who might use the data to inform their decisions about test use. Payers are interested in outcomes research because findings might affect reimbursement decisions. Identified gaps in this sector included insufficient evidence of the clinical utility for most tests; inadequate, outdated systems for coding, coverage, and payment for genetic tests and services; and limited information and transparency on the number and type of genetic tests used in clinical and public health practice.

Dr. Goodman explained that the map represented a high-level snapshot of the current oversight system. He stated that the diagram was extremely complicated because it accommodated an evolving diversity of testing and testing services and an uneven patchwork of legislation and regulation. Dr. Ferreira-Gonzalez led the group in a discussion of the map.

Dr. Paul Billings stated that the report should describe how the map relates to the overarching issues of public awareness, access, and the fostering of innovation. Dr. Marc Williams added that the map should identify the parts of the process that are transparent to the consumer and those that are not transparent. Dr. Khoury asked how LDTs can be offered directly to consumers, bypassing the system presented in the map. Dr. Ferreira-Gonzalez replied that the companies offering the tests claim they do not fall under CLIA statutory regulation because they are not offering diagnostic or medical services. Dr. Steve Gutman said the map omitted a small but important niche line of submissions, the investigational device exemption (IDE). He said he would provide The Lewin Group with several other technical corrections. Dr. Teutsch suggested that the map clarify where the issues with analytic validity, clinical utility, and clinical validity were occurring, so that it would be easier to see where gaps existed. He recognized that the problems were not in isolated places, but occurred across the spectrum. Dr. Khoury pointed out that EGAPP was designed not only to conduct a clinical utility review, but as a way to review analytic validity

and clinical validity. Dr. Evans commented that Dr. Goodman's presentation of the map was so helpful that the report should provide a website address to facilitate access to it. Dr. Gurvaneet Randhawa suggested adding the fact that outcomes research information, as well as clinical utility data, informs the USPSTF and EGAPP recommendations. Dr. Ferreira-Gonzalez suggested that the guidelines developed by professional organizations be reflected in the CLIA-Exempt State and CLIA Regulation sectors. Dr. Tuckson recommended that a commentary be added to the map on accountability by major Government agencies.

Dr. Ferreira-Gonzalez thanked Dr. Goodman for his presentation and introduced a discussion of the final oversight recommendations.

Discussion of Final Oversight Recommendations
Facilitators: Dr. Tuckson and Dr. Ferreira-Gonzalez

Dr. Ferreira-Gonzalez stated that in the January 30th teleconference with the full Committee, several issues were identified in the oversight recommendations that needed further discussion. She said the session would focus first on the recommendations for effective communication and decision support in Chapter 6; followed by the recommendations for the development and evaluation of evidence for the clinical utility of genetic tests in Chapter 5; and then by the recommendations for analytical validity, proficiency testing, and clinical validity in Chapter 4. The overarching recommendation at the end of Chapter 2 would be addressed last. The three primary questions she would ask concerning each recommendation were: Do you need more information to clarify the intent of the recommendation? Does the recommendation adequately address the identified problem? Is the wording of the recommendation satisfactory? During the discussion that followed, the Committee agreed to go back over the recommendations on Day 2 to eliminate redundancy in content.

Chapter 6

Recommendation 1 in Chapter 6 addressed deficiencies in genetic knowledge by relevant stakeholder groups. Concerning recommendation 1A, Ms. Mara Aspinall stated that public and private payers should be included as examples of groups in need of genetic education efforts. She suggested that a timeline for implementation be added. Dr. Williams stated that the Education Task Force should develop the timeline, and the Committee agreed to discuss the specifics of timelines the following day. Dr. Khoury's suggestion that the phrase, "deficiencies in genetic knowledge and education," be changed to, "deficiencies in knowledge of appropriate genetic and genomic applications and practice" was accepted.

During discussion of recommendation 1B, Dr. Tuckson stated that the Secretary of HHS should ensure that there are adequate research resources available to advance analytical validity, clinical validity, and clinical utility for multiple purposes. He therefore suggested editing 1B to read, "Based upon increased research regarding analytic validity, clinical validity, and clinical utility, sufficient resources should be provided for the translation of this knowledge into evidence-based clinical practice guidelines that enhance the quality of clinical care and public health outcomes. The Committee recommends the Secretary ensure the availability of information regarding the clinical use of tests to determine the adequacy of information and its translation meets the needs of improved clinical care and outcomes." The Committee voted to accept this revised wording.

Recommendation 2 called for FDA to engage with relevant HHS advisory committees and other stakeholders to gather perspectives on an appropriate regulatory framework for clinical decision support systems because the areas in which FDA was choosing to exert control was not clear. Dr. Gutman pointed

out that the regulatory authority used by FDA in this area had been in effect for a long time; however, the Committee agreed that since current decision support tools did not exist at the time the relevant statute was enacted, there was a need for clarification of FDA's approach. Dr. Williams added the phrase, "As part of this process…" at the beginning of the second sentence of the recommendation, which stated that FDA should develop a draft guidance on clinical decision support systems.

Recommendation 3 recognized the need for genetic expertise to support best genetic testing practices and requested that HHS act on the recommendations in the 2006 SACGHS Coverage and Reimbursement of Genetic Tests and Services Report. The Committee accepted the recommendation as written.

Recommendation 4 requested that HHS allocate resources to the Agency for Healthcare Research and Quality (AHRQ), the Centers for Disease Control and Prevention (CDC), the Health Resources and Services Administration (HRSA), and the National Institutes of Health (NIH) for research and development of clinical decision support tools and resources. In response to public comments, the Task Force had revised the recommendation to include the need to engage providers and payers in education efforts and to provide incentives on protections to ensure participation in the design, dissemination, and implementation of clinical decision support. The Committee accepted the recommendation as written.

Recommendation 5 requested that HHS step up its efforts to assess the implications of DTC advertising and testing and implementation of strategies to protect consumers. In response to public comments, the Task Force had revised the recommendation to include the concepts of social stigmatization and privacy concerns as potential harms, and added HRSA to the list of relevant Federal agencies that should be involved in issues related to DTC advertising and testing. The Committee discussed previous and ongoing collaborative efforts by the FTC and FDA in warning consumers about potential harms related to DTC genetic tests. They considered referring to this collaboration in the recommendation and/or report. Dr. Tuckson stated that the recommendation should be more directive, perhaps asking the Secretary to determine within a short timeframe which Federal agency or agencies have oversight authority for DTC tests. Since this issue was also addressed in a recommendation in Chapter 4, the Committee agreed to review the two recommendations during the Chapter 4 discussion and decide whether both were necessary. The Committee agreed, however, that both DTC advertising and testing should be covered by the regulatory system and that the final SACGHS recommendation should be directive in nature.

Chapter 5

Recommendation 1 in Chapter 5 called for HHS to create and fund a public-private entity to assess the clinical utility of genetic tests. The Task Force had revised the recommendation to include examples of evidentiary standards and levels of certainty for different situations and added that data from electronic medical records (EMRs) should be used in research. Part B of the recommendation called for the development and funding of a research agenda that would address gaps in knowledge of analytical validity, clinical validity, and clinical utility and on the population health impact of genetic tests. The Committee accepted the recommendation as written.

Recommendation 2 requested that HHS act on the recommendations in the 2006 report on Coverage and Reimbursement of Genetic Tests and Services, and asked public and private health care payers to develop mechanisms, such as coverage with evidence development or phased reimbursement, to facilitate the collection of clinical utility evidence for high priority tests and applications. The Task Force had revised the recommendation to include the need to evaluate whether mechanisms to collect clinical utility evidence enhance or hinder innovation, understanding of effectiveness, and proper utilization. Dr. Tuckson noted that the recommendation did not address who would pay for these efforts and where they

would be housed. The Committee discussed combining Recommendation 2 with Recommendation 1, so that these issues would be addressed by a public-private stakeholder group. Several members advocated for keeping the recommendations separate, and Dr. Randhawa agreed, stating that Recommendation 2 had a different focus than Recommendation 1, i.e., the implementation of research for high priority tests and applications. He agreed with Dr. Tuckson that the question of who would pay for these activities remained unaddressed. Dr. Randhawa volunteered to draft revised wording for Recommendation 2 that would take into account the various entities, in addition to health care payers, that could fund special research topics in clinical utility.

Recommendation 3 requested that HHS conduct public health surveillance to assess health outcomes or appropriate surrogate outcomes, practice measures, and the public health impact of genetic testing. Several Committee members noted some redundancy with Recommendation 1B in Chapter 6. It was agreed that the two recommendations should be cross-referenced, and the following phrase was added to Recommendation 1B in Chapter 6: “See also Recommendation 5-3."

Recommendation 4 asked HHS to advance appropriate use of interoperable patient-level data for research and to enhance the quality of decisionmaking. The Task Force had revised the recommendation to include implementation efforts. Dr. Tuckson noted that the recommendation urged coordination of SACGHS with AHIC, which was in the process of transitioning to a public-private partnership. The recommendation was revised to apply to AHIC and its successors.

After completing the discussion of the recommendations in Chapters 6 and 5, Dr. Ferreira-Gonzalez explained that the ad hoc experts and *ex officio* members of the Task Force had been invited to provide input directly to the full Committee. She introduced Dr. Kathy Hudson, who had prepared comments on the draft oversight report.

Kathy Hudson, Ph.D.
SACGHS Oversight Task Force

Dr. Hudson stated that she was the Director of the Genetics and Public Policy Center at Johns Hopkins University, and she was honored to serve on the Oversight Task Force. She commented on three issues: the recommendation for enhancements to CLIA, the recommendation for a test registry, and DTC genetic testing. Concerning enhancements to CLIA, she noted that the Task Force recommended an expansion of PT, but not the creation of a genetic testing specialty. She said the Task Force was correct in ascertaining that CMS did not want to create a new specialty, and therefore it was wise to focus on the goal of additional PT. She stated that the key issue would be which PT programs CMS would approve and noted that there would be a market to create PT programs for tests offered on a widespread basis. Dr. Hudson said that implementing this recommendation would require changes to CLIA regulations. Her organization had drafted a model regulation that would fulfill the requirements of the report and avoid concerns about genetic exceptionalism, which she offered to share with the Committee.

Dr. Hudson said the draft report included a recommendation for the creation of a voluntary test registry, possibly as an extension of GeneTests. She noted that the majority of public comments on the issue recommended that the registry be mandatory. Several commenters urged that the registry be housed and managed by a Federal regulatory body. Questions that remained unanswered included: What functions would be carried out by the registry? Would they facilitate data submission? Would the registry conduct any quality control? What leverage would there be to demand that data be submitted, and would there be penalties for noncompliance? Dr. Hudson stated that it was not clear whether the various Federal agencies or HHS would have sufficient authority to require the kinds of information envisioned for the registry.

She stated that Secretary had the ability to re-delegate authority to the agencies to resolve these issues. Dr. Hudson said the registry should be housed in an agency that has documented experience with creating and running publicly accessible registries, such as FDA, CDC, or NIH.

Concerning DTC testing, Dr. Hudson commented on the map presented by Dr. Goodman. She said the map had a separate line for “DTC non-CLIA certified;” however, the CLIA statute applies to laboratories that provide assessment of health, irrespective of how they are marketed. She did not agree that there was a distinct pathway and said the map could be misleading.

Dr. Tuckson thanked Dr. Hudson and continued the session on the final oversight recommendations.

Discussion of Final Oversight Recommendations (Continued)
Facilitators: Dr. Tuckson and Dr. Ferreira-Gonzalez

Chapter 4

Recommendation 1 in Chapter 4 proposed steps to support and augment the CMS action plan to address gaps in genetic testing oversight in lieu of a genetic testing specialty. The Task Force had revised Part A of the recommendation to call for CMS to require PT for all high complexity tests for which PT products were available. Dr. Ferreira-Gonzalez explained that PT ensures the analytical validity of a test. In response to a question from Dr. Tuckson, Dr. Ferreira-Gonzalez clarified that the Task Force did not recommend the addition of a genetic testing specialty under CLIA because genetic testing was already covered by personnel requirements for high complexity laboratory testing and by quality control requirements. In addition, much genetic testing is already covered because it cuts across current CLIA specialties. Dr. Ferreira-Gonzalez added that since the technology for genetic testing was continually evolving, specific requirements imposed by a genetic testing specialty could become problematic in the future (e.g., limit future development). The Task Force believed that making changes to the requirements for PT would address many major concerns related to the lack of a genetic specialty.

Dr. Tuckson asked for clarification on the key gaps in oversight that had caused concern and whether wording could be added that would summarize them. Dr. Ferreira-Gonzalez replied that gaps included a lack of PT, which was being addressed by the recommendation, and insufficient inspection of genetic testing laboratories. She said CMS was developing new guidelines for inspections. Ms. Judy Yost added that CMS was analyzing oversight needs and using existing mechanisms and information to address them, rather than taking 6 years to implement proposed and final rules to add a genetic testing specialty.

After discussing the meaning of high complexity tests versus moderate complexity tests and hearing about the FDA standards that must be met for waived tests, the Committee changed the wording of the recommendation to, "CMS should require PT for all nonwaived tests for which PT products are available" (rather than for all high complexity tests). The next part of the recommendation stated that alternative assessment methods must be used for tests without PT products (as required by CLIA). Recommendation 1 also urged HHS to fund studies to identify additional ways to conduct PT. Part B expressed support for the steps CMS was taking to improve processes and procedures in genetic testing laboratories, primarily through the training of inspectors. Part C supported the 2006 recommendation by the Government Accountability Office (GAO) Report on Clinical Laboratory Quality stating that CMS should use revenues generated by the CLIA program to hire sufficient staff to fulfill CLIA's statutory responsibilities. Several editorial changes were agreed upon and Recommendation 1 was accepted.

Recommendation 2 requested that HHS ensure funding for the development of reference materials, methods, and samples for assay validation, quality control, and performance assessment; as well as other steps to address gaps in analytical and clinical validity data. Dr. Michael Amos suggested changing the wording to take into account standards for both analytes for specific tests and platform standards for microarrays or mass spectronomy, and the Committee agreed. Dr. Ferreira-Gonzalez explained that the Task Force had revised Part C to state that an initiative for enhancing public reference databases should encourage robust participation and consider mechanisms for anonymous reporting and protection from liability for information-sharing. Part D encouraged professional organizations to develop professional guidance for applying genetic tests in clinical practice. Ms. Aspinall suggested that specific language be added to ensure that the necessary support was available for this effort, as well as incentives to ensure follow-through. She agreed to draft the new text.

Dr. Ferreira-Gonzalez moved to discussion of Recommendation 4, stating that Recommendation 3 would be discussed later. Recommendation 4 asked HHS to convene relevant stakeholders to provide further input on FDA's risk-based regulatory framework for LDTs and consider models for assessing LDTs that are not subject to FDA review. The Task Force had revised Part A by expanding the list of stakeholders and including LDTs offered directly to consumers. They also added that the FDA risk basis should consider intended uses of LDTs and the likelihood of harms to consumers if test results were inaccurate or misinterpreted. Part B, which proposed new private or public-private models for LDTs not subject to FDA premarket review, had been revised to offer alternative assessment models for infrequently performed LDTs. Dr. Ferreira-Gonzalez explained that this recommendation was a subject of controversy both within the Task Force and in the public comments. She also noted that oversight map made it clear that for LDTS that go through FDA, laboratories must comply with both FDA and CLIA inspections, leading to some duplication of effort. Dr. Tuckson asked if the issues addressed in Recommendation 4 left any gaps in oversight, and Dr. Williams and Dr. Ferreira-Gonzalez said they did not.

Ms. Aspinall stated that there should be a recommendation stating whether SACGHS agreed with the IVDMIA FDA guidance on premarket review, specifically whether premarket review should be required of LDTs. Dr. Williams said the Task Force agreed that there needed to be a risk-based strategy for premarket review, but that input from other stakeholders was required to make sure the risk stratification was properly done. Private or public-private models could be developed to address those LDTs not subject to FDA premarket review. After discussion of the current system for premarket review and realistic steps that could be taken by the HHS Secretary, it was decided that Dr. Ferreira-Gonzalez and staff would revise Recommendation 4 and present it to the Committee the following morning.

FEBRUARY 13, 2008

Welcome and Opening Remarks

Dr. Teutsch welcomed everyone to the second day of the SACGHS meeting. He introduced Mr. Rick Campanelli, the Secretary's Counselor for Human Services Policy, who was there to present a certificate of appreciation to Dr. Tuckson.

Presentation of Certificate of Appreciation to Dr. Tuckson

Richard Campanelli, J.D.
Counselor for Human Services Policy
Senior Policy Advisor to the Secretary
U.S. Department of Health and Human Services

Mr. Campanelli thanked Dr. Teutsch for his willingness to serve as the new SACGHS Chair. He stated that he was there on behalf of the Secretary to recognize Dr. Tuckson's important work as Chair over the previous several years. He acknowledged the effectiveness of the Committee in the face of changing genetic discoveries and expressed gratitude for Dr. Tuckson's leadership of SACGHS. Mr. Campanelli noted that Dr. Tuckson had served as the Commissioner of Public Health in the District of Columbia, as a university president involved in health and science, and as an expert in professional standards in the Nation's largest physicians' organization. Dr. Tuckson's work was important not only to the Secretary, but to the Department and the public. Under his leadership, SACGHS developed important recommendations and background pieces on coverage and reimbursement of genetic tests, genetic discrimination, marketing of genetic tests to consumers, genetics education for health professionals, and pharmacogenomics; and a report on the oversight of genetic testing was soon to be released. Mr. Campanelli read a letter from the Secretary, which expressed appreciation for Dr. Tuckson's willingness to lead, sacrificing his private interests to advise on the planning and operation of HHS programs. In recognition of Dr. Tuckson's contributions to SACGHS, the Secretary forwarded a certificate of appreciation. Dr. Tuckson stated that he enjoyed working on behalf of Secretary Leavitt and, previously, Secretary Thompson, and he expressed pride in the achievements of the Committee. He then turned the meeting over to the new Chair, Dr. Teutsch.

Opening Remarks from the New SACGHS Chair

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

Dr. Teutsch said it was a great privilege to follow Dr. Tuckson as Chair of the Committee and acknowledged his vision, leadership, generosity of spirit, clarity of focus, and sense of humor. He said the Committee was composed of an incredibly talented group of people with deep knowledge and experience in many aspects of genetic health and health care, health care policy, and personal experience with genetic conditions. He expressed tremendous optimism that the Committee could build on the legacy of SACGHS to make even greater contributions.

Dr. Teutsch introduced the Public Comment period. He welcomed Dr. Hudson, who had spoken the previous day as a member of the Oversight Task Force, and was present to speak from the perspective of the Genetics and Public Policy Center.

Public Comments

Kathy Hudson, Ph.D.
Director, Genetics and Public Policy Center

Dr. Hudson said the public believes the genetic tests they take to make important health-related decisions are analytically and clinically valid, but that is not always the case. She said the oversight recommendations must ensure that there is adequate evidence and that the evidence is transparent to the public. She stated that increased oversight would not stifle innovation and pointed out that the comments

of AdvaMed, a trade association for device manufacturers, and Roche, a pharmaceutical company, argued that more oversight was needed. Dr. Hudson said there had been no discussion about the deleterious impact of the status quo on innovation. IVD manufacturers face significant disincentives to produce validated test kits. A manufacturer can present evidence to FDA and go to market, but the next day, another genetic testing company can offer the same test or make identical claims without oversight from FDA. She noted the significant numbers of LDT providers on the Task Force and the lack of IVD manufacturers. She stated that the Committee would not fulfill its mandate unless it made recommendations that substantially leveled the playing field for businesses that are focused on innovation and working to obtain FDA approval.

Dr. Hudson made several points about DTC genetic testing. First, she said the map provided by The Lewin Group inaccurately showed a non-CLIA regulatory pathway for genetic tests. She said that selling an LDT without CLIA certification is against the law. Second, the vast majority of DTC tests are subject to CLIA as they make explicit or implicit claims about health assessments. The majority of companies providing DTC testing claim that they are providing those tests from CLIA-certified laboratories, and she called on CMS to verify those claims. Third, Dr. Hudson said there were a number of inaccuracies in statements about the regulatory status of DTC tests. The definition of a clinical laboratory is one that examines samples derived from the human body to provide information about the diagnosis and treatment of disease or for the assessment of health of human beings. She said this definition and all the CLIA regulations cover laboratories whether the tests are being sold directly to consumers or through a provider. Fourth, oversight cannot be bypassed by companies such as 23andMe by claiming that the genotype provides research information. The exemption in CLIA for research applies only if those research results are not provided to the research subject. If results are being provided to customers, the tests must be performed in CLIA-certified laboratories. Finally, Dr. Hudson questioned whether progress was being made in FTC efforts on DTC testing. She stated that she was not aware of any efforts made by FTC since the issuance of a consumer alert 18 months previously, even though numerous consumer complaints of misleading claims were made to the agency, as well as a class action lawsuit. She suggested that the Secretary investigate FTC's evaluation of these misleading claims.

Dr. Teutsch thanked Dr. Hudson and introduced Robert DiTullio from AdvaMed.

Robert DiTullio
AdvaMed

Mr. DiTullio said he was with Sequenom, a molecular diagnostics and research company in San Diego, and was co-chair of AdvaMed's Diagnostics Task Force. AdvaMed is the world's largest association representing manufacturers of medical devices, diagnostic products, and medical information systems. Mr. DiTullio presented a proposal from AdvaMed that he said would be the least burdensome approach for the regulation of all diagnostic tests. The proposal suggested realigning the intensity of regulatory oversight with the patient risk/benefit ratio in mind and allowing FDA to focus its limited resources on only the highest risks. It promoted the FDA oversight of safety and effectiveness of all diagnostic tests. The underpinnings of the proposal included several key principles. First, all clinical laboratories should be subject to CLIA requirements and quality standards, and FDA should oversee the safety and effectiveness of all diagnostic tests no matter where they are made, because they have the same risk/benefit profile for patients. AdvaMed promoted FDA oversight of tests, with oversight focusing primarily on the risk of harm associated with how the test result is used to treat patients. AdvaMed also proposed that low-risk tests and well-standardized tests should be exempt from FDA premarket review or only subject to labeling review of the performance claims. This approach would allow FDA's resources to be used toward higher risk tests, which could be cleared or approved using a risk-based approach that aligned data

submission requirements and the intensity of the review with the risks. Patient access to specialized test categories should not be disadvantaged. The proposal further stated that FDA and CMS should harmonize regulatory requirements for diagnostic tests and leverage each other's standards and resources for oversight of LDTs. The new oversight system should be implemented through notice and comment rulemaking and guidance, as appropriate. The final principle was that CMS should recognize that all new diagnostics must receive timely and adequate reimbursement.

Dr. Teutsch thanked Mr. DiTullio and welcomed Ms. Pam Dixon from the World Privacy Forum.

Pam Dixon
Executive Director, World Privacy Forum

Ms. Dixon stated that the World Privacy Forum is a nonprofit public interest research group that focuses on in-depth analyses of privacy issues and longitudinal research. One focus area is health care privacy issues. They were interested in the aspects of privacy that they felt were underrepresented in the Oversight Report. They were concerned that marketing interests and misused science would crowd out legitimate genetic testing and privacy, particularly outside the clinical sphere. Questionable privacy activities related to consumer-consented health care data were already occurring in the health care sector. She gave an example of DirectMag.com, a direct marketing online magazine for marketing companies, which provides 60,000 marketing lists. A quick search indicated that they have 406 lists keyed to the term "diabetes." Most lists are generated from actual consumer health care data. In this case, 2,186,700 consumers are known and identifiable, and there are 400 data points included about the consumers. These include gender, household income, and type I or type II diabetes. “Selects” can be purchased along with the base list that include the age of the person and their children, education level, ethnicity, prescriptions and over-the-counter medications taken, and purchasing activities of the consumer. She emphasized that consumer-reported data were being disclosed, not clinical data. Ms. Dixon was concerned that as DTC advertising and genetic testing mature and the price drops, the situation could become worse and impact consumers. The World Privacy Forum submitted comments on the Oversight Report that made three recommendations: (1) they asked that privacy be expressly included in the report as an issue for examination, including privacy outside the clinical setting; (2) they recommended that a group be tasked to address the specific privacy issues that are arising in this context; and (3) they asked the Committee to urge the FTC to state that genetic data and requests for genetic tests on websites be off the table in terms of advertising, using the data for marketing purposes, or for any purposes other than health care.

Dr. Teutsch thanked the commenters and turned the Committee’s attention back to the discussion of the final oversight recommendations.

<u>Session on Oversight of Genetic Testing</u>

Discussion of Final Oversight Recommendations (Continued)
Facilitators: Dr. Tuckson and Dr. Ferreira-Gonzalez

Chapter 4, Continued

Dr. Ferreira-Gonzalez presented the new wording developed for Recommendation 4 in Chapter 4. It read:

"The Committee is concerned by the gap in oversight related to clinical validity. The Committee believes that it is imperative for this gap to be closed as expeditiously as possible. To this end, the Committee makes the following recommendations:

All high-risk LDTs should be reviewed by the FDA in a manner that takes advantage of its current experience in evaluating laboratory developed tests. In order to accomplish this recommendation, the Committee recommends convening a multistakeholder public and private sector group to determine the criteria for risk stratification and a process for systematically applying these criteria. The multistakeholder group should also explicitly address and seek to eliminate duplicative oversight procedures. For all other tests, this multistakeholder group is also charged with the development of a review process that meets the needs of protection of the public. This group should also consider existing regulatory models and data sources (e.g., New York State), and responsibility for overseeing this review process should be defined by this group. To expedite and facilitate the review process, the Committee recommends the establishment of a registry, as noted in Recommendation 3."

Dr. Kevin FitzGerald stated that the recommendation did not define “high risk,” and suggested that the multistakeholder group define this term. Dr. Khoury noted that the Secretary’s Advisory Committee on Genetic Testing (SACGT) attempted to define “high risk” and “low risk” for almost a year without success. Dr. Gutman objected to the recommendation and said it would be better to recommend that FDA refine its risk system. In addition, he was not sure the registry would be legal in terms of FDA statutory authority. Dr. Khoury requested that Recommendation 4 be written in a more direct way, urging specific action on the part of FDA or HHS; Dr. Tuckson agreed.

The Committee discussed whether there was a need to convene the recommended public and private multistakeholder group to develop criteria for laboratory tests. Dr. FitzGerald suggested that SACGHS recommend that FDA continue to address all laboratory tests according to their current standards, while also calling for HHS to convene the multistakeholder group to inform the process. The majority of Committee members agreed. Because of resource constraints on FDA, Dr. Evans suggested advocating for more resources for the agency. The Committee accepted the language advocating that the stakeholder group should eliminate duplicative oversight procedures and examine new and existing regulatory models and data sources. The final statement in the revised Recommendation 4 referred to a mandatory registry, which the Committee addressed in more detail in Recommendation 3, described below.

Dr. Ferreira-Gonzalez led the Committee in a discussion of Recommendation 3, which supported a mandatory system of genetic test registration that would use CLIA registration data as a foundation. Based on public comment, the Task Force had significantly revised the draft recommendation, which had called for a voluntary system of registration through a public-private partnership. However, the Task Force was split on where such a registry should be housed, that is, at CMS or FDA. SACGHS staff therefore explored the issue with *ex officios* from CMS and FDA, which led to unanswered questions about the legal authority to gather and publicly display certain data elements. The Steering Group then modified the recommendation to take into account this significant development. The revised recommendation read:

"There are considerable information gaps about the number and identity of laboratories performing genetic tests and the specific genetic tests being performed. To gain a better understanding of the genetic tests being offered as laboratory developed tests and to enhance transparency in this field, SACGHS reviewed proposals for a voluntary or mandatory test registry and considered the benefits and burdens of each type of system. The Committee decided that a mandatory, publicly available, Web-based registry that is well staffed to maintain an accurate and current database will offer the best approach to address the information gaps. Since genetic tests are not unique from other laboratory tests for oversight purposes, the registry should include all LDTs. The Committee also discussed whether such a database should reside at CDC, CMS, or FDA. Based on the exploratory work, SACGHS concludes that the concept of a mandatory registry offers promise, but recognizes that there are unresolved issues, including practical and

legal questions, that require further analysis before a final decision can be made about how and where to implement the registry. In light of these unresolved issues, SACGHS recommends the following course of action:

A. CDC, in collaboration with CMS and FDA, should convene a stakeholder meeting by September 2008 to determine the data elements to be included in the test registry. CDC should cast a wide net for broad stakeholder representation, including representatives from the private sector who can represent a role for the public-private partnership in developing a registry. CDC, through this stakeholder effort, should assess the level of effort, as well as the burden on the laboratory and the impact on the other key stakeholders, such as patients, physicians, and payers necessary to obtain each data element, including linking to reliable sources of existing information.

B. HHS should perform the requisite legal analysis to determine what data elements, as determined by the CDC stakeholder group, can be required by CDC, CMS, and/or FDA. For example, if clinical validity is a required data element, the legal analysis should determine whether CDC, CMS, or FDA currently have the statutory authority to require reporting of this information for all LDTs. If these agencies do not currently have the necessary statutory authority, the legal analysis should identify specific statutory provisions that may be needed in order to effect the system of enhanced reporting requirements and a statutory authority should be sought.

C. HHS should appoint and fund a lead agency to develop and maintain the mandatory registry for LDTs. The lead agency should work collaboratively with its sister agencies to create a comprehensive registry and minimize duplicative collection of registry information. The lead agency should be staffed with qualified personnel who are experienced in developing and updating large databases in a timely and accurate manner.

D. While awaiting completion of the above processes, HHS should use short-term voluntary approaches, such as incentivizing laboratories to register with GeneTests, and encouraging laboratories to make their test menus and clinical validity data for these tests publicly available on laboratory websites."

Dr. Khoury suggested tasking HHS to create the registry, but disagreed that a legal analysis of the required data elements should be conducted. He said they should be the elements that address analytic validity, clinical validity, and clinical utility. He stated that the bullet describing HHS creation of the registry (C.) should appear first, followed by the bullet describing the stakeholder meeting (A.). He said that bullet should be changed to reference “the lead agency” instead of CDC, as the activities described would not necessarily be conducted by CDC. The Committee agreed to these changes by a show of hands.

Recommendation 5 requested better enforcement of existing regulations for laboratories that do not have CLIA certification but are performing health-related tests. The Task Force had revised Part A to include the fact that laboratories without CLIA certificates cannot be reimbursed by Medicare and Medicaid, yet these restrictions have no consequences for laboratories that perform DTC testing. The Task Force was seeking more direct enforcement ability, that is, CMS should require that the laboratory cease and desist testing, without having to go through the HHS Inspector General. Dr. Evans wanted to make the language stronger than merely stating, “HHS should explore mechanisms…,” and Dr. Teutsch suggested the following wording: "HHS should strengthen its enforcement efforts against laboratories." After a discussion with Ms. Yost about the appropriate way to seek greater authority for CMS in this matter, the

following wording was added: "…CMS should establish and exercise its regulatory authority to take direct enforcement actions against laboratories that perform tests for clinical purposes without proper CLIA certification."

Recommendation 6 called for expanding CMS's statutory authority through CLIA to encompass certain DTC tests that appear to fall outside of CLIA's scope. The Task Force had revised this recommendation to include FDA's authority and regulatory process. The Committee accepted the recommendation as written.

Ms. Yost informed the Committee that CMS was collaborating with CDC and FDA in investigating the type of testing performed by laboratories that were not under the purview of CLIA. They had identified 64 laboratories to date and were conducting continuous followup until they were satisfied that the laboratories were not only enrolled, but in compliance.

Dr. Ferreira-Gonzalez introduced discussion of an overarching recommendation that outlined steps to enhance interagency coordination for oversight activities.

Dr. Paul Wise proposed that the overarching recommendation frame the issues identified by the Task Force, emphasize their importance, and describe the recommendations that address them in a way that would be accessible to the general public. Dr. Tuckson agreed and asked that the concept of accountability be added, that is, the Secretary must use his power to coordinate efforts at HHS to protect the public. Dr. Telfair stated that the overarching recommendation should begin with a strong outcome statement explaining the overall goals of the report's recommendations. Staff members worked with the Committee to draft new language to capture these concepts.

Wrap-up and Decisions on Final Recommendations and Draft Report

The Committee read through all the recommendations to provide an opportunity for final changes. They decided to integrate the unique points of Recommendation 5, Chapter 6 into Recommendation 4, Chapter 4. They also pointed out editorial changes in various recommendations that would be made by staff members subsequent to the meeting. The Steering Group agreed to make additional changes to the overarching recommendation to incorporate the Committee's comments. Dr. Tuckson led the Committee in a vote to accept the recommendations. All recommendations were approved and the content of the report was approved in spirit. The Committee was told they could submit final edits for the report by February 20th. It was agreed that the recommendations would be finalized and sent to the Secretary by February 29th. After the editing process, the final version of the report would be transmitted in April.

Dr. Teutsch thanked the Oversight Task Force and turned the floor over to Dr. Barbara Burns McGrath, who would lead the Committee in a review of the charge for the Genetics Education and Training Task Force.

Session on Genetics Education Task Force

Draft Charge for the Genetics Education and Training Task Force
Chair, Barbara Burns McGrath, R.N., Ph.D.

Dr. Burns McGrath provided background information on the education and training interests of SACGHS. A meeting was held in 2003 that resulted in a 2004 resolution presenting recommendations on genetics education and training. A SACGHS session on the topic was held in November 2007 that covered perspectives from various disciplines, professional education and training programs, diversity in

the work place, family history, emerging issues (e.g., gene-environment), and emerging stakeholders. The Committee decided that the topic continued to be of interest and they formed a new Task Force. The 2004 resolution was used as a starting point. Since November, the Task Force had communicated via email and teleconferences to develop a draft charge. Dr. Burns McGrath read the charge to the Committee:

"Advances in genetics and genomics are leading to a better understanding of disease processes and improved application of genetic testing to guide health decisions. With increased integration of genetics into other medical disciplines, however, health professionals with or without training or expertise in genetics are challenged to keep pace with this dynamic and rapidly evolving field. Education will have to address the growing importance of genetics in common disease, which likely will require more knowledge and understanding about risk assessment and communication. In addition, the accelerated growth of direct-to-consumer genetic services highlights the need for informed decisionmaking. To realize the benefits of genetic technologies and protect against potential harms, the education of health care professionals, the public health work force, and the general public is critical. For these reasons, the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) has formed a Task Force to build on the findings of the Committee's 2004 resolution on Genetic Education and Training of Health Professionals. The Task Force is charged with developing a plan to identify the education and training needs of health professionals, lay health educators, and the general public in order to optimize the benefits of genetic and genomic services for all Americans. This plan will also outline the steps required to meet these needs and evaluate the efficacy of educational and training efforts. The plan includes, but is not limited to, the following activities:

1. Assembling evidence to determine which recommendations from the 2004 SACGHS education resolution were implemented and which ones require additional efforts.
2. Identifying the education and training needs specific to genetics and genomics for health professionals involved in providing care for individuals and for those involved in the development of guidelines, policies, and strategies for incorporating genetics and genomics into clinical care.
3. Identifying the education and training needs of lay health educators who are non-credentialed individuals from the local area trained to promote health and provide general health care services for a specific condition or program.
4. Identifying the education needs specific to genetics and genomics for medical directors, administrators, and policymakers in the public and private sectors to inform policy development, legislation, coverage and reimbursement decisions, and other issues that directly or indirectly impact the provision of genetic services.
5. Identifying the education needs of patients and consumers to assist them in informed decisionmaking about the use of genetic services and enhance their understanding and utilization of results and how these results impact decisions about prevention or treatment.
6. Identifying effective educational tools that can be incorporated into electronic health records, personal health records, and clinical decision support systems that would enhance the appropriate integration of genetic and genomic technologies throughout the health care system without adversely impacting privacy, access, and work flow. In addition, identify gaps where such tools do not currently exist and develop recommendations on how to address these gaps.
7. Assessing the use of evaluative research methods to determine the efficacy of genetics and genomics education and training.

8. Promoting active involvement by health professional governing bodies that influence education and training (e.g., residency review, National Board of Medical Examiners) to be more proactive

in their requirements for genetics in curricula, clinical training, and licensing and certification and continuing education requirements.

Dr. Burns McGrath stated that ad hoc members would be added to the Task Force based on the finalized goals. She asked for comments on the scope of the Task Force and indicated that she wanted the goals to be measurable.

Discussion and Finalization of Task Force Charge

Dr. Williams suggested engaging with the Secretary's Office to determine whether any actions had been taken on the 2004 resolution and to move forward from there. Dr. FitzGerald noted that a wide variety of efforts addressing genetics education for particular groups had been undertaken and HHS could coordinate or serve as a focal agency in this area. Ms. Aspinall agreed, pointing out that the representatives from various disciplines at the previous meeting's roundtable said they were not in ongoing communication with one another. She suggested that the Task Force build on the work that was already being done. Dr. Khoury wanted to ensure that HHS would be given specific, unique guidance that was not merely telling the agency to do more of the same things. Dr. Williams added that the scope should be defined by the areas the Secretary has the ability to control (i.e., HHS agencies). He also said there was a significant opportunity to deal with education on clinical decision support and point-of-care education. The Secretary could relate this information to AHIC or AHIC 2, leveraging the new learning methodology that was developing.

Dr. Phyllis Frost noted that NIH had formed a trans-NIH communication group on complex genetics and diseases and was moving forward with an agenda, headed by Dr. Alan Guttmacher. She suggested coordination with those efforts. Dr. Randhawa addressed Goal 6, on effective educational tools that could be incorporated into electronic health records. He stated that a list was needed of various educational mechanisms and modalities (e.g., Web-based, paper-based, and person-to-person education), as well as a list of different populations and the modalities that would work in various settings. Dr. Paul Miller recommended that the Task Force ask for input from the stakeholder community on the 2004 resolution and consider what the end product of the new effort should be. Dr. Billings suggested looking at the educational programs designed for judges. Ms. Sylvia Au emphasized the need for coordination of HHS education efforts for consumers, with sustainability in mind. She said previous efforts by various agencies were scattered and not sustainable.

Dr. Burns McGrath asked for ideas on evaluation. Dr. Telfair said the original recommendation on evaluation was very prescriptive, which was not feasible. However, evaluative methods could be used to determine how different tools and approaches were used by different groups. He said the evaluation piece would have to be developed more specifically after other decisions were made about the effort. Dr. Randhawa said it would be useful to have a sense of the desired outcomes, for example, increased knowledge, improved decisionmaking, or health outcomes over the long term. Dr. Williams added that there was good literature showing what works in terms of retention. Lt. Col. Scott McLean said that, based on his practice, the most important outcome is living longer and living better. Dr. Billings asked that the Task Force address whether there is something specific about genetics education and health that should be highlighted, as contrasted with health education on other topics. Dr. Telfair suggested a focus on specific priority areas and use of a step-wise approach leading to a set of actionable, functional recommendations. He said the focus should start with providers and clients, after which other populations could be addressed. The Committee agreed to submit other suggestions on the charge and scope of the Task Force via email.

Dr. Teutsch asked the Task Force to revise the charge based on the input they received and return to the July meeting with a timeline for their activities. The charge would then be finalized and discussion would take place on such issues as the need for *ad hoc* members.

Session on Planning For July Priority Setting

Future SACGHS Priorities: Issues and Planning Process
Dr. Teutsch

Dr. Teutsch introduced a brainstorming session on new priorities that might be appropriate for the Committee to take up. He said no final decisions would be made that day and that the list of topics would be refined at the July meeting. He noted that the original SACGHS priorities were established through a systematic process undertaken in 2003 and 2004. Dr. Teutsch referred to a description of this process in Tab 5 of the briefing books. He said that Committee members and *ex officios* developed a list of 19 topics during a brainstorming session. A Task Force was formed to narrow down the list using a specific set of criteria and to investigate the remaining issues in preparation for discussion at the March 2004 meeting. Four of the criteria questions used were: "Does the Government have jurisdiction or authority over the issue? Does the issue under consideration raise concerns that only the Government can address, or would Government involvement be duplicative of other efforts? Is another body addressing the issue or better equipped to address the issue? Has a policy solution to the issue already been worked out?" Dr. Teutsch stated that once these and other criteria questions were answered, issue briefs were prepared on the remaining 11 issues.

At the following SACGHS meeting, members and *ex officios* deliberated on the issues and organized them into three categories: those that required in-depth study, those that required short-term action or monitoring, and overarching issues that would be considered within the context of all other issues. The issues categorized as requiring monitoring were genetic discrimination, genetics education and training, patents and access, and oversight. Coverage and reimbursement, large population studies, pharmacogenomics, and DTC marketing were categorized as requiring in-depth study. The overarching issues were access, public awareness, and genetic exceptionalism.

Dr. Teutsch asked for volunteers to serve on a Priority-Setting Task Force. The purpose of the group would be to continue brainstorming the issues suggested during the session, develop a plan for the priority-setting process, and identify the types of background materials needed for the July session. Dr. Paul Wise agreed to serve as Chair, and Dr. Billings, Dr. Evans, Ms. Aspinall, Dr. Khoury, Dr. Teutsch, and Dr. Randhawa volunteered to serve on the Task Force.

Dr. Teutsch reviewed a list of possible priority topics submitted by the Office of the Secretary. The first related to the international genomics infrastructure for clinical research. The next was primary care practice-based approaches for the integration of continuing medical education (CME), curricular, and medical boards. The third topic was clinical research standards for biospecimen collection. The fourth was the economic and diagnostic value of multiplexed genomic tests and how costs are integrated into commercial development plans to determine what factors developers use to assess value. The fifth topic was co-development of molecular targeted agents and diagnostic biomarkers. Dr. Teutsch also pointed out that an article in *Nature Reviews Genetics* requested that SACGHS define terms related to the storage of whole genome data in electronic health records. Dr. Teutsch told the Committee that SACGHS would be hearing a great deal about personalized genome services during a half-day session at the July meeting, which could raise additional issues. He asked Ms. Au to work with staff to help organize the session.

Dr. Teutsch opened the floor for suggestions on priority topics. Dr. Williams suggested exploring the reality of the $1,000 genome in the near future and its possible impact on genetic testing. Dr. Billings raised the issue of the distinction between health-related genetics and recreational or ancestral genetics. He noted that there had been little or no discussion about gene therapy recently. He then suggested a topic on the translation of pharmacogenetics and genomics as they relate to personalized medicine. Dr. Julio Licino raised the issue of privacy and access to medical records by oneself or others (e.g., the military, law enforcement). Dr. Evans stated that the rapid emergence of genetics and genomics into medical practice would stress the fragmented health care system in the United States. He said fundamental health care reforms would be necessary to reap the benefits of genetic medicine and ensure that innovations in genetics do not create more disparities in health care delivery. Ms. Aspinall agreed, and added that reform efforts should address the economic incentives in the system, that is, how they drive care or lack of care at the public health and individual levels. Dr. Khoury suggested that HHS, possibly in collaboration with the private sector, invest money in trials and observational studies that would allow evaluation of the utility and validity of promising applications, so that evidence-based guidelines could be developed. Dr. Randhawa suggested a white paper on research priorities for pharmacogenomics, and suggested exploration of genetic modification labeling of food and genetics and cloning. Lt. Col. McLean said SACGHS could examine a practical public health approach to screen for genetic conditions in populations beyond the newborn period. Dr. Wise raised concerns about the implications of the new genetics for minority health, and Ms. Au added that identity issues for minorities should be explored as well. Dr. Burns McGrath agreed, and stated that the phrase "genetic exceptionalism" should be better defined. She added the issue of HHS policy on promoting stem cell research. Dr. Denise Geolot wanted to examine privacy issues as they relate to large population studies and the use of genetics, especially with regard to informed consent and reconsent. She recommended a possible collaboration with the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC) on this issue. Dr. FitzGerald said the Committee should reexamine the definitions of “genetics, "genomics," and "genetic testing."

Dr. Teutsch closed the session by stating that he would like to solicit directly the opinions of the *ex officios* in a conference call. Dr. Evans suggested seeking public comment on priority issues.

Next Steps and Concluding Remarks
Dr. Teutsch

Dr. Teutsch summarized the accomplishments of the meeting and reviewed next steps. He said the Oversight Report would be copyedited following the meeting, with transmission of the recommendations to the Secretary to take place by the end of February and finalization of the entire report by April 30. The Education Task Force would develop a plan and timeline for their charge, and the Priority-Setting Task Force would set up a process for further discussion of priority issues at the July meeting. Ms. Au and Dr. Licinio would work with staff members to organize a session on personal genome services. Dr. Teutsch closed by stating that the July SACGHS meeting would take place in the Humphrey Building. He adjourned the meeting.

We certify that, to the best of our knowledge, the foregoing meeting minutes of the Secretary's Advisory Committee on Genetics, Health, and Society are accurate and correct.

Reed Tuckson, M.D.
SACGHS Chair (for February 12, 2008)

Sarah Carr
SACGHS Executive Secretary

Steven Teutsch, M.D., Ph.D.
SACGHS Chair (for February 13, 2008)