The Secretary's Advisory Committee on Genetics, Health, and Society
Summary of Third Meeting
March 1-2, 2004
Bethesda, Maryland

March 1

Welcome, Opening Remarks

Edward McCabe, M.D., Ph.D.
Chair, Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS)

Dr. McCabe welcomed everyone and pointed out that the public was notified of the meeting through the *Federal Register* and the SACGHS website and listserv. He acknowledged the appointment of two new *Ex Officio* members, Mr. Matthew Daynard, Senior Attorney for Federal Trade Commission (FTC), and Dr. Ellen Fox, Director of the National Center for Ethics at the Department of Veterans Affairs. He noted that the inter-meeting task force formed at the last meeting to narrow the priority issues for the committee's work over the next year will guide the issue identification process. Twelve issue briefs found in the briefing books were prepared to facilitate discussion.

Dr. McCabe reviewed the primary goals of this meeting as identifying the top one to three priority issues to be the focus of the Committee's future work; developing a work plan; and beginning deliberations on the first issue. He outlined the agenda as follows:

- **First day:** The Committee will hear about the work of the task force in advancing the priority-setting process and take a straw vote of Members and *Ex Officios* on their priorities. Discussions on the priority issues under consideration will be followed by presentations and a roundtable discussion on coverage of and reimbursement for genetic technologies and services.
- **Second day:** The Committee will take a second straw vote, continue discussions, make final determinations about the prioritization of issues and develop a work plan.

Review of Process and Outcome of Inter-Meeting Priority Setting Project

Emily Winn-Deen, Ph.D.
Chair, Inter-Meeting Task Force

Dr. Winn-Deen provided an overview of the work of the inter-meeting task force appointed in October 2003 to begin the prioritization process. The task force conducted two straw votes of Members and one straw vote of *Ex Officios* on all of the issues considered during the first two SACGHS meetings. She explained the issue identification and ranking process that guided the delineation of the top twelve priority issues. Issue briefs on the top priority issues were developed by staff with guidance from the task force in order to facilitate discussion during this meeting. The top twelve priority issues were access; coverage and reimbursement; direct-to-consumer marketing; exceptionalism; genetic discrimination; genetics education and workforce; large population studies; oversight; pharmacogenomics; patents and access; public awareness; and vision statement. Although genetic discrimination did not rank in the top twelve because action on the issue had already been taken, the task force elected to consider it a standing committee priority because of its potential impact and an ongoing issue to be monitored.

Discussion and Votes on Twelve Priority Issues

The discussion began with some general comments. Dr. Francis Collins noted that the National Academy of Sciences recently began an 18-month study on patents and access, looking at the impact of patents on genetics, genomics, and biotechnology, which will ultimately include recommendations for steps that might be taken to maximize benefits to the public. Mr. Brad Margus recalled that the NIH was planning to commission the Institute of Medicine (IOM) to assess the large population study idea. Dr. Collins reported that the assessment is not yet under way and remains under discussion.

Many Members noted that there was considerable overlap among issues, and it was agreed that although subgroups of topics with common threads could easily be created, they would be kept as separate topics for now. It was suggested that rather than using the access topic as a separate issue, it could be applied as a framework for deciding what kind of consideration issues merit from the committee. It also was pointed out that Members and *Ex Officios* appeared to approach the ranking process in different ways, and discussion ensued regarding why these differences may have occurred and how they may have affected the outcome. Dr. McCabe suggested that the differences might have occurred because Committee members may have ranked according to their assessment of importance, but agencies may have ranked according to what they thought agencies could accomplish or according to the interpretation of DHHS's role and where action could be taken in the short term.

Dr. McCabe suggested that issues of ranking would probably emerge for discussion between the first and second straw votes, but that some prioritization had to be achieved through clarifying the important priority issues the Committee will address versus areas where quick recommendations could be made. Dr. Winn-Deen said that Members should keep in mind the Committee's timeframe and be aware that that some issues are important but are already being addressed by others.

A straw vote was taken and was tallied during break. It was agreed that evaluating topics according to the following four questions (drawn from a list presented to the Committee) during discussion would help members prioritize topics on the second straw vote (to be held the following day):

- Does the government have jurisdiction/authority over the issue?
- Does the issue raise concerns that only the government can address or would government involvement be duplicative?
- Is another body addressing the issue or better equipped to address the issue?
- Have the policy solutions to the issue already been worked out?

The following four issue categories were established (with the third category added during the discussion on access, below):

1: Not high enough priority to pursue in the initial consideration.
2: High enough priority, but can be dealt with through a brief statement or through another rapid approach and/or through monitoring.
3: Integral to each of the topics and thus taken off the table for consideration as a separate priority issue (added during discussion on access).
4: High enough priority to merit substantive deliberation by the committee.

Access (ranked first by Members and tenth by *Ex Officios*)

Ms. Barbara Harrison presented an overview of the access issue, explaining that it overlaps with several other issues also being considered. Dr. David Feigal noted that the Federal government has both a direct and indirect role in affecting access to genetic technologies, and that its role depends on what aspect of access is being discussed. This comment led to a discussion of whether the Committee's scope extends beyond HHS. Ms. Sarah Carr explained that although the Committee's main purpose is to advise the HHS Secretary, the participation of non-HHS agencies in the Committee's allows for recommendations to be made on issues outside the purview of HHS and that the establishment of priorities should not necessarily be limited to issues that the HHS Secretary has jurisdiction.

Ms. Cynthia Berry posed a question about what the Committee's goal would be if it were to address the access issue. Dr. Huntington Willard questioned whether a genetic exceptionalist approach is appropriate for addressing this issue or whether it is better to simply acknowledge that it is part of a larger issue. Mr. Margus and others noted that the pervasiveness of the access issue may make it better suited to be considered in the context of each of the other issues. After further discussion, it was unanimously agreed that access should be a Category 3 issue, i.e., integral to each of the topics.

Coverage and Reimbursement (ranked second by Members and ninth by *Ex Officios*)

Ms. Harrison initiated discussion on the issue of coverage and reimbursement by considering the guiding questions and suggesting that earlier work by the Secretary's Advisory Committee on Genetic Testing (SACGT) could be used as a springboard for addressing this issue fairly quickly.

In response to the question of whether the government has jurisdiction and authority over this issue, Dr. Winn-Deen explained that the government does to the extent that there is a public insurance system (e.g. Medicare, Medicaid) but that private insurance accounts for approximately 60 percent of the health insurance market, which is beyond the Secretary's authority. Dr. Kaytura Felix-Aaron noted that it was for the latter reason that the Agency for Healthcare Research and Quality ranked this issue toward the bottom. Dr. McCabe noted that the government does have influence over the private sector in that decisions made by the Centers for Medicare & Medicaid Services (CMS) since decisions made in the public programs are frequently adopted by the private sector. Mr. Paul Miller suggested that it was for this reason that the issue ought to be ranked highly. Dr. Robinsue Frohboese remarked that the previous work of SACGT and the Secretary's initiatives on the uninsured, underinsured, Medicare reform and prevention make for ideal timing for addressing this issue. After further discussion, a unanimous vote designated this as a Category 4 issue, that is a high priority issue requiring in-depth study.

Several members identified particular areas in the realm of coverage and reimbursement that warrant the Committee's attention. Dr. Debra Leonard remarked that billing codes are inadequate to provide information to third-party payers about what test is being performed. She also predicted that if the reimbursement issue is not addressed, laboratories will increasingly be unable to subsidize genetic testing. Ms. Harrison reported that genetic counseling experiences similar problems. Ms. Agnes Masny identified a need for better funding for, access to, and integration of prevention and health promotion genetic technologies. Dr. McCabe noted that other groups are working on related issues (U.S. Preventive Services Task Force) and suggested that if SACGHS tries to tackle this issue, it should incorporate their expertise and products.

Possible directions discussed for addressing this issue included developing guidelines for determining whether evidence is sufficient to warrant coverage for a particular genetic technology and identifying gaps in the evidence base.

Education and Workforce (ranked fifth by Members and second by *Ex Officios*)

Dr. Willard said that the two questions that frame this topic are related to genetic exceptionalism and the extent to which this is a federal/DHHS issue, rather than an academic or professional society issue. Even though the issue does not fall under the direct jurisdiction of the government, there is much the government can do to influence the establishment of educational programs that will enhance the integration of genetics/genomics into health care services. It also was suggested that the anticipated relevance of genetics to all of medicine does make it somewhat exceptional. It was noted that training encompasses those already in the pipeline as well as practicing professionals and that diversity is an important issue in this area.

The role of federal agencies was stressed, including, for example, HRSA's health professions programs. Dr. Feetham suggested that a federal role might be to encourage interdisciplinary education, training, and practice. The Centers for Disease Control and Prevention (CDC), which has a number of projects ongoing, also considers education a top priority, both in the sense of workforce development in general and specific training in genomics for the health workforce. The CDC-sponsored Centers for Genomics in Public Health are active in providing training and technical assistance to the states and health professionals.

Private groups, such as the National Coalition for Health Professional Education in Genetics (NCHPEG), are involved in educating and training health professionals. NCHPEG primarily focuses on generating materials that professional societies can integrate into their own educational efforts. Dr**.** Collins presented gaps as identified by NCHPEG, including licensure, diversity and curricular development, and noted that assessing the gaps would be practical before making recommendations. It was noted that a SACGT working group focused on education as well.

The Committee placed this issue in Category 2, with the following caveats: SACGHS endorses and supports the efforts that HRSA, NCHPEG, CDC, and others have undertaken in working toward the goal of integrating genetic training and realizes that the Committee's recommendations may have some influence over AAMC, ACGME, and other groups in their efforts, as well as other key organizations and federal partners. The Committee may wish to confer with some of these organizations in the future. The Committee could assist if there were specific actions that could be taken to address a specific gap. The Committee can hear presentations from other organizations involved with this issue at future meetings, continue to monitor it to ensure that movement is occurring quickly enough, note changes, and issue statements or take other directions as appropriate. Issues involving education and training can be addressed as the Committee looks at other priority areas, and diversity is understood as a key component of education and an important part of this discussion.

Large Population Studies (ranked fourth by Members and sixth by *Ex Officios*)

Dr. Willard explained that a significant number of large population studies are ongoing in other countries and that discussions are under way regarding the need for and design of large population studies in the United States. Dr. Collins provided information about a meeting that the National Human Genome Research Institute (NHGRI) convened in December 2003 to get input from a group of experts in genetics, epidemiology, and environmental health sciences on the merits and feasibility of carrying out a large-scale cohort study in this country to study both genetics and environment and how they interact to play a role in common disease. He said that such a study would involve individuals from childhood to late adulthood with a family-based approach covering three or four generations. He discussed possible

connections with other ongoing studies and the major issues covered, including study design, power, affordability, and the environmental, genetic, and phenotypic data that could be collected. He noted that the issues discussed were not resolved but that the group agreed that a large-scale cohort study in this country looking at both genetics and environment and how they interact to play a role in common disease would be extremely valuable.

To achieve the kind of power required for useful data, at least a half million people would be needed, and it would be an extraordinarily expensive undertaking that would continue for maybe two or three decades. NIH would find this difficult to approach in its current circumstances, and to make this a viable option, a substantial effort to raise consciousness about the project's importance would be needed, akin to that involved in launching the Human Genome Project. Dr. Collins explained that follow-up discussions have been held, and the plan is to formalize these discussions by assembling a working group of experts to try to flesh out some of the questions that were not answered during the two-day workshop.

SACGHS Members discussed arguments for conducting such a study in the United States. Funding possibilities were considered, including the possibility of public/private partnerships. It was agreed that large population studies are important for the realization of the anticipated medical benefits of the Human Genome Project. Members pointed to other issues that need to be addressed including cost, infrastructure, other resources, and logistics. It was noted that this study would need to be made a national priority, as its successful completion would require more resources than are available to any of the research agencies represented around the table.

The value of conducting a longitudinal cross-sectional study with a population-based sampling strategy not focused on any particular disease was discussed, and it was pointed out that the Department of Veterans Affairs is also working in this area. A consensus has not emerged regarding whether the best strategy would be to conduct one large population study or to conduct a number of different studies that are balanced with respect to gender, include children and include some targeted studies with more immediate endpoints to develop evidence that would allow genetics to move into the practice of medicine more quickly.

Discussion also involved whether this Committee is the appropriate one to make a decision about scientific merit or study design. Members agreed that if the logistics and the scientific merit were demonstrated, the Committee could serve as a forum to discuss progress in this country and around the world, look at opportunities offered by different models, and discuss ethical, legal, and social issues (ELSI) issues and issues such as access, education, training, oversight, equity, discrimination, and public awareness. The Committee could make this information available to the public and provide guidance as the large population-based study was conducted.

Members agreed that the Committee could, after learning more about what such a project might mean for medicine and society and weighing that information, decide whether to endorse such a large longitudinal population study. Dr. Collins said he would welcome any connection with this Committee and that a full presentation could be planned to evaluate progress and determine what areas might merit further attention. The Committee would also like to hear about models being used in other countries and ELSI issues they have faced. Questions might include whether this study could use existing large-scale cohorts (for which DNA samples have already been collected) and what the possibilities might be for public/private partnerships in this area. Members agreed that it was a topic worth considering further, and all voted in favor of placing this issue in Category 4.

Public Awareness (ranked seventh by Members and first by *Ex Officios*)

Ms. Berry characterized this topic as broad and in need of more concrete definition. Public awareness is important in the context of consumers having direct access to genetic technologies. There is also more potential for harm if there is insufficient regulation or oversight or insufficient involvement of educated health care providers. The federal government can play a role in educating the public, and many groups and agencies are working on certain aspects of public awareness, including the Department of Energy, NHGRI, HRSA and the March of Dimes, and American Association for the Advancement of Science. Some academic organizations have taken this on, and some government websites are providing information to the public. Policy solutions are much needed.

Getting the message out in a variety of ways to increase public awareness is important, as is providing credible, accurate, unbiased information and its sources. Department websites and brochures that can be disseminated through doctors' offices were discussed, as were the importance of including non-Internet sources and of protecting people from inaccurate information. It was noted that the Internet has some good information about genetics and genomics available, including the NIH website. Dr. Winn-Deen and Dr. McCabe mentioned that SACGT had been working on a patient brochure that was not completed and that might be useful for this Committee and to the Secretary; therefore, completing it might be worthwhile. There was discussion on whether work in this area would be done by the Committee using department resources for ongoing efforts or mostly by the Department.

Discussion ensued about genetic exceptionalism, with the suggestion that genetics is different because of its predictive nature. Dr. Feigal suggested the committee use its bully pulpit to get important messages across, and Dr. McCabe emphasized the need to be particularly sensitive to language and cultural diversity in increasing awareness. Dr. Collins stressed the importance of identifying the teachable moment and ensuring that the right information is available at that moment, which makes educating health care professionals valuable and important, and he noted that it is difficult to evaluate the impact of an educational investment. It was suggested that clearly identifying the message is essential. The importance of educating those in K-12 was discussed.

Although there are efforts under way by different government agencies and private organizations, there was agreement that significant gaps exist in the area of public awareness. If SACGHS were to focus on this issue, it would need to determine what role to play, from monitoring to more proactive leadership, using the Committee as a bully pulpit and/or and making concrete recommendations and creating partnerships with different agencies and groups. Members discussed how this issue in many ways seems to be one that transcends the other issues. A vote was taken, and the topic was unanimously assigned to Category 3.

Public Comment Session

Margaret Gulley, M.D.
College of American Pathologists (CAP)

Dr. Gulley described CAP's involvement in the area of CPT coding and reimbursement regarding genetic technologies and discussed the College's Genetic Testing Work Group's efforts and conclusions. She said that the numeric alpha code modifier system is the most viable solution and described CAP's efforts towards such a system. She reviewed CAP's position on patent policy, saying that gene patents pose a serious threat to medical advancement, medical education, and patient care and that they set an extraordinary and dangerous precedent and affect the availability of diagnostic testing. She discussed progress on CAP's approach to addressing genetic test oversight utilizing existing regulatory mechanisms that include laboratory accreditation and proficiency testing programs. She suggested that instead of

increasing federal regulations or developing duplicative federal programs, it would be best to work through the existing programs to improve oversight of genetic testing.

Judith Lewis, Ph.D., RN, FAAN
International Society of Nurses in Genetics (ISONG)

Dr. Lewis noted that ISONG is committed to working toward ensuring that the nursing workforce is well prepared to serve the needs of patients and the public for genetic information. She described ISONG membership and the role of the nursing workforce in genetic health and programs that help ensure that nurses have the genetic competencies needed for practice at the basic level as well as enhanced competencies for advanced practice nurses. She also discussed programs for preparing nursing faculty who may have been educated in the pre-genomic era and other programs serving doctorate nurses and doctoral students. Current programs, while providing a valuable service, do not have the capacity to meet the demand. If we are to continue to prepare an educated workforce, the profession needs resources to enhance its education and outreach efforts.

Sharon Terry, M.A.
Genetic Alliance

Ms. Terry suggested that the metric for measuring and weighing the 12 issues that the Committee has identified is improved human health. Success for genetics means translating the knowledge into technologies and treatments that improve health, and the government should be involved in facilitating success. Noting that genetic exceptionalism exists, she commented that coverage and reimbursement, genetic discrimination, genetics education and training, oversight, direct-to-consumer advertising, patents, and public awareness are all subsets of access. She said that pharmacogenomics and large population studies could not be done well in the current regulatory climate. She emphasized the importance of integrating genetics into medicine and noted a lack of incentives for early adopters of proven genomics technologies. She urged the Committee to make policy recommendations that could facilitate the climate needed for these studies.

The question before this Committee, she said, is whether it is ready to look at these issues in a fresh way and whether it is committed to discovering the real roadblocks in the system rather than just dealing with the symptoms. She indicated that the Committee has the ability to recommend changes and systems that will no longer allow politics to set the scientific agenda and that the answer might include universal health care. She noted that it is time for science and politics to be integrated, in order to formulate a vision for the future. The Committee requested her written comments and she indicated she would provide them to a staff member.

Andrea Ferreira-Gonzalez, Ph.D.
Association for Molecular Pathology (AMP)

Dr. Ferreira-Gonzalez provided information on issues that are affecting the ability of laboratories to provide genetic testing services, including the inadequacy of CPT coding and reimbursement for genetic tests and the negative effect of gene patents on molecular diagnostic laboratories. She provided an update on advances in the oversight of genetic testing laboratories. She said that AMP asks that SACGHS remains cognizant of the progress in implementing the proposed changes in molecular genetic test coding and of the impact on payment for molecular genetic testing in the future. AMP encourages the Committee to examine the negative impact on medicine of current practices in the patenting and licensing of genetic sequences and to work to eliminate restrictions on the medical use of genetic information. AMP also asks the Committee to review the changes implemented by CAP addressing test

validation and oversight concerns to determine whether these changes address the concerns previously raised by SACGT.

Session on Coverage and Reimbursement for Genetic Technologies and Services

Private Health Insurance Coverage and Payment Policies and Decision-making Process for Genetic Technologies and Services
Michele Schoonmaker, Ph.D.
Specialist in Genetics, Congressional Research Service

Dr. Michele Schoonmaker provided an overview of how private health plans make coverage and payment decisions for genetic technologies and services. Dr. Schoonmaker presented her own views and not the views of the Congressional Research Service.

In the United States, health insurance coverage is provided by public programs such as Medicare and Medicaid or by private health plans purchased individually or through one's employer. In 2002, a total of 44 million Americans had no health insurance.

In general, the scope of coverage provided by a health plan is outlined in broad benefit categories; decisions about specific services are made on a case-by-case basis during claims processing. Health plans may develop formal coverage policies that describe whether a particular service is covered and the conditions under which it will be covered. Medical directors, the federal and state governments, medical policy advisory committees, employers, and unions are among the entities involved in making coverage decisions.

Medical necessity is the primary criterion used by health plans when making coverage decisions, although the definition is subject to interpretation. The Blue Cross/Blue Shield Association's Technology Evaluation Center, which has formally defined its coverage decision criteria, requires final approval from the requisite regulatory body (i.e., the Food and Drug Administration) or compliance with requisite rules (e.g., Clinical Laboratory Improvement Amendments (CLIA)) before it will be considered for coverage. Also, the scientific evidence must permit conclusions concerning the effect of the technology on health outcomes; the technology must improve net health outcome and be as beneficial as established alternatives; and the improvement must be attainable outside investigational settings.

In general, health plans cover genetic technologies when the following criteria are met: personal or family history indicates a high risk for inherited conditions, when the sensitivity of a test is known, when the results will directly affect the treatment or management of the patient, when the diagnosis remains uncertain following conventional workup, and when pre- and post-test counseling is provided as appropriate. Genetic testing is typically not covered for population screening without a personal or family history of disease regardless of ethnicity, for informational purposes only, for testing minors for adult-onset diseases, or (with some exceptions) for a patient's family member who is not also a member of the health plan. Even with these general conditions for coverage, there is still tremendous variation in the level of detail of policies and in the specific analyses covered.

Several factors are considered when setting payment rates, including the setting in which the service is provided, geographic area, and the usual and customary charges billed by providers in that location. Reimbursement rates can be a percentage of charges or fees that have been negotiated between the insurer and provider.

Common coding systems, such as the Current Procedural Terminology (CPT), Healthcare Common

Procedure Coding System (HCPCS), and the International Classification of Disease (ICD), 9th edition, facilitate the billing process by conveying what service or procedure was performed and why. The CPT codes available for genetic testing are limited in that they often require using multiple codes to bill for a single test, which may result in overall payment that is lower than it would be if a single code were available.

Dr. Schoonmaker concluded by commenting that although testing for most inherited genetic conditions is covered by most private insurers, including counseling, reimbursement may not be adequate. Furthermore, there is a perception that insurers are slow to cover new technologies, which insurers may argue is a result of a lack of data to support the medical benefit of new tests. She recommended that providers present cost analyses to insurers to convey to insurers how costs are applied and where reimbursement rates are failing.

Genetic Services: The HMO Model
Ronald Bachman, M.D.
Chief, Genetics Department, Kaiser Permanente of Northern California

Dr. Bachman described the clinical and financial aspects of delivering genetic services in a health maintenance organization (HMO) setting like Kaiser Permanente of Northern California (KPNC).

KPNC has five genetic centers and a molecular and cytogenetics laboratory staffed by 207 full-time employees, including 11 medical geneticists, 53 genetic counselors, 17 genetic nurses, three metabolic nutritionists, four Ph.D. laboratory directors, and administrative support. The services provided at these centers include prenatal services (e.g., screening for cystic fibrosis), neonatal services (e.g., newborn screening), screening for disorders based on ethnicity (e.g., sickle cell and thalassemia), fetal pathology, adult genetic services, multi-specialty clinical services for treating common genetic disorders, and genetic education. Last year, KPNC's Genetics Department saw over 103,000 cases.

Dr. Bachman also reviewed how new programs are introduced into the Genetics Department. First, the New Technologies Committee, comprised of geneticists and genetic counselors, discusses the proposed program. If approved, the proposal is submitted for review as part of the KPNC budget process and then sent to the KPNC administration. If the proposal is found to be standard of care, cost-efficient, and well-supported, a cost basis for the new service is established, productivity and actual costs are monitored. In 2002, the budget for KPNC's genetics programs is expected to exceed $24 million in 2004, or 65 cents per member per month.

In the future, Dr. Bachman predicted that there will be a greater demand for testing and screening, which will require more genetics providers, a greater reliance on, and thus increased educational needs for, primary care providers as well as the provision of new types of services, such as preimplantation genetic testing, microarray testing for genetic disorders, and pharmacogenomics. To meet these demands, genetic counselors will need to work closely with primary care providers and the internet will be replied upon for patient triage, collection of medical and family history information, pedigree construction, and patient and provider education.

A Laboratorian's Perspective on Reimbursement for Genetic Technologies and Services
Andrea Ferreira-Gonzalez, Ph.D.
Associate Professor and Director, Molecular Diagnostics Laboratory, Virginia Commonwealth University

Dr. Ferreira-Gonzalez provided an overview of payment rates for genetic testing in clinical laboratories.

CPT and HCPCS codes are used to facilitate payment for health services. There are 14 methodology-specific CPT codes available (e.g., molecular isolation or extraction) for billing for genetic technologies and test interpretation and consultation. CPT code modifiers may be added to provide additional information about the procedure or interpretation. CMS establishes HCPCS codes for procedures for which a CPT code does not exist. Often these codes do not adequately account for differences in performing the various procedures involved in genetic testing.

CMS uses a National Laboratory Fee Schedule that establishes a national limit for each laboratory CPT and HCPCS code. States then determine the actual payment rate for their particular area, which may be below or at, but not above, the national limit. This rate-setting approach can result in significant variation in payment amounts for the same procedure. Payment rates by private health plans are largely identical to Medicare's fee schedule, but may be lower if the private plan does not recognize modifier codes. Recently, Federal legislation was passed that freezes Medicare laboratory fee schedule payment rates at the current amount through 2008, despite steadily increasing costs of personnel, overhead, and other expenses.

Royalty payments, as outlined in the licensing agreement between the laboratory and patent holder, also affect payment rates for patented procedures and genes or sequences. Royalty payments are generally comprised of an upfront, one-time payment plus a flat per-test fee or percentage of charges. Several years ago, a survey study by Cho et al. reported that 25 percent of responding laboratories stopped performing tests after receiving orders from the patent holder to cease testing.

Using payment data from her experience at Virginia Commonwealth University (VCU) Medical Center Molecular Diagnostic Laboratory, Dr. Ferreira-Gonzalez showed that the actual costs of performing genetic tests may exceed Medicare's payment rate in Virginia. For instance, genetic testing for Factor V Leiden costs $72 but is reimbursed only $68. Genetic testing for Fragile X using Southern hybridization analysis costs $266 to perform but is reimbursed only $62. Laboratories have been able to recoup some of these losses through over-reimbursement of other tests; however, with the recent freeze on payment rates, their ability to do so will be diminished.

The frequencies in which claims are paid also vary tremendously. For instance, Medicare reimburses 89 percent of the claims submitted by the VCU laboratory, while Medicaid reimburses 72 percent of the time, and private insurers reimburse 61 to 85 percent of the time.

A Clinician's Perspective on Reimbursement for Genetic Technologies and Services
Marc Williams, M.D.
Pediatrician and Medical Geneticist, Gundersen Lutheran Medical Center

Dr. Williams presented information on billed services, multidisciplinary evaluation, access to services, and problems with the current system.

Clinicians use CPT evaluation and management (E&M) codes to bill for office visits, which are, in general, reimbursed at half the rate of procedures (e.g., surgery). The elements used to determine the appropriate E&M code – history, physical exam, and complexity – are poorly defined, subject to interpretation, and do not include certain key aspects of genetics consultations. The American College of Medical Genetics had submitted to the American Medical Association (AMA) CPT Editorial Panel, which is responsible for the CPT coding system, a request for the creation of two new CPT codes for pedigree analysis that would address some of these inadequacies. However, shortly before the CPT Panel was to vote on this request, it announced plans to revamp the entire E&M coding system. The new

system emphasizes time and obviates the need for special pedigree analysis codes. The maximum time level component is 60 minutes, however; genetic encounters are frequently two to three hours in duration, not including pre- and post-encounter time and coordination of time. While there are some CPT code modifiers that can be used to augment the basic code, it is not unusual for third-party payers to reject these modifiers. The establishment of evaluation and assessment codes for genetic counseling could help address some of these problems. The Health Care Professionals Advisory Committee, which is a body of non-physician allied health professionals advisory to the CPT Editorial Panel, has recently submitted a request for the development of such codes.

Physician profiling, an audit technique used by third-party payers to adjust charges, can also be problematic for geneticists. Many geneticists are trained in another specialty (e.g., pediatrics, internal medicine) and listed as such in a health plan's provider network. Because of differences in their patient mix and office visit complexity, geneticists typically submit higher-level codes than primary care providers. If profiled according to their other specialty, however, their billing practices can seem out of line. This can lead to down coding and low reimbursement and even fraud and abuse investigations.

Another problem with the current billing system is that patients with genetic conditions are often seen by multiple providers on the same day, yet providers are not permitted to use the same ICD code if the patient is seen the same day by another provider for the same condition. This restriction can impair coordination of care, inconvenience patients, and decrease the quality of care.

Additionally, few states license genetic counselors, and genetic counselors are ineligible for unique provider identification numbers (UPINs) used for billing Medicare. As a result, genetic counselors are not recognized as a billable entity by many public and private health plans and, thus, are only able to bill incident to their supervising physician or, in the case of hospital-employed genetic counselors, as part of the hospital facility fee. "Incident to" providers are limited in which codes they are allowed to use when billing Medicare. For instance, genetic counselors may only bill Medicare using a CPT code that was developed to cover counseling of patients on the side effects of immunizations which is typically conducted by nurses and lasts five minutes. Some states had developed local codes that allowed genetic counselors to directly bill, however, the Health Insurance Portability and Accountability Act ended states' ability to have separate codes. These billing limitations affect the availability of genetic counseling services and financial viability of a medical practice. Planned changes to the UPIN system are expected to allow genetic counselors to be issued a provider ID number, which will facilitate their ability to bill health plans willing to allow genetic counselors to bill directly.

Medicare Coverage Policies and Decision-making Process for Genetic Technologies and Services
Sean Tunis, M.D., M.Sc.
Chief Medical Officer, Centers for Medicare & Medicaid Services

Dr. Tunis provided an overview of Medicare's coverage process as it applies to genetic technologies and services.

Health technologies and services must meet several criteria in order for them to be considered for Medicare coverage. First, technologies that are under the regulatory purview of the Food and Drug Administration (FDA) must be FDA-approved for at least one indication, although Medicare has flexibility to cover off-label uses. Home brew genetic tests not under the purview of FDA do not require approval to be reimbursed by Medicare. Second, the service must fall into one of the statutorily defined benefit categories (e.g., diagnostic services). A patient's strong family history would not by itself qualify genetic testing as a diagnostic service. Most pharmacogenetic tests, on the other hand, would be considered diagnostic because they are typically performed in patients who have existing signs and

symptoms of disease. Third, the service must be found to be reasonable and necessary for treatment of illness or injury. To be deemed reasonable and necessary, there must be adequate evidence to conclude that the item or service improves net health outcomes experienced by Medicare patients (e.g., improved functional status, quality of life or psychological outcomes or reduced morbidity or mortality), and the service must be as good as or better than currently covered alternatives. Evidence is weighted based on the source and methodology, with more emphasis placed on evidence from sources with less potential for bias. For genetic tests, the critical pieces of evidence are test performance (i.e., test sensitivity and specificity) and clinical utility (i.e., impact on patient management and outcomes). Cost, cost-effectiveness, and cost-benefit analyses are not considered formally in making reasonable and necessary determinations.

CMS has had a longstanding policy, based on interpretation of the Medicare statute, that excludes coverage for screening and preventive services in the absence of signs, symptoms, or personal history of disease or illness. While nothing in the statute prohibits CMS from designating testing in high-risk individuals with no signs, symptoms or personal history of disease as diagnostic, Congress has historically legislated exceptions to this policy (e.g., diabetes screening in high-risk individuals was legislated by the Medicare Prescription Drug, Modernization and Improvement Act of 2003). Thus, any change to this policy would require a rulemaking process.

Medicare coverage decisions are made at both the national and local levels. CMS has a formal process for making national coverage decisions. In the absence of a national policy, Medicare contractors who process Medicare claims make local coverage decisions. Local decisions are based more heavily on expert opinion, whereas national decisions rely more on empirical evidence. Currently, there is only one national coverage decision for a type of genetic test – cytogenetic testing for acute myelogenous leukemia, acute leukemias, congenital anomalies and myelodysplasia – and several local decisions (e.g., HER-2 neu and BRCA testing, although most do not cover BRCA testing because they consider it to be screening). Covered tests accounted for 270,000 claims that were paid at a total cost of $13 million. With advances in genetic testing, pharmacogenomics and personalized medicine, there will be more coverage decisions pertaining to this area of medicine.

Medicare Payment Rates and Decisions
Donald Thompson, M.S.
Director of Ambulatory Services, Centers for Medicare and Medicaid Services

Mr. Thompson provided an overview of Medicare's payment decision process as it applies to genetic technologies.

Payment rates for new genetic tests fall under the rubric of the Medicare clinical laboratory fee schedule. Payment rates for such laboratory tests are handled through two methods – gap-filling and cross-walking. Gap-filling is a decentralized process that involves asking local Medicare carriers to determine a local payment amount for a new test with a new CPT code based on charges for the test and any discounts, resources required to perform the test, clinical studies, and information from local clinicians, manufacturers and other interested parties. Cross-walking refers to the process by which a new laboratory test (with a new CPT code) is assigned the same fee as a similar, existing test. These processes include public meetings with public testimony and recommendations from interested parties on whether a gap-filling or cross-walk process should be used and, if the latter, what existing CPT code should be used. Tentative determinations are posted on the web for additional public comment, and final determinations are usually made in early November.

The Medicare Prescription Drug, Improvement, and Modernization Act of 2003 included provisions that

affect the clinical laboratory fee schedule. One provision prevents CMS from updating existing laboratory fees until 2009. A second provision requires CMS to publish formal regulations about the process CMS uses to determine payment rates for new laboratory tests. A formal rule-making process is planned. A third provision allows for a demonstration project for competitive bidding for laboratory tests. Since CMS does not have statutory authority to revise the clinical laboratory fee schedule, this last provision could be an approach to remedying current deficiencies. A second potential approach would be for CMS to invoke its inherent reasonableness authority for existing tests whose payment rates are deemed inherently unreasonable.

Cost-Effectiveness Determinants and Data Needs
David Veenstra, Pharm.D., Ph.D.
Assistant Professor, Department of Clinical Pharmacy, University of California-San Francisco

Dr. Veenstra discussed the role of cost-effectiveness analyses in coverage and reimbursement decisions for genetic technologies.

Dr. Veenstra began by saying that cost-effectiveness analyses provide a framework for evaluating the complex and conflicting factors, in addition to cost, that are involved in making coverage and reimbursement decisions in health care. He stressed that although results of cost-effectiveness studies are sometimes used in making reimbursement decisions in the United States, most of the decisions are evidence-based. Currently cost-effectiveness information is used mainly in considering expensive and novel technologies expected to have a large impact on health care practice and costs. Genetic technologies are not yet on the radar screen of payers; however, genetic tests will generate more interest in cost-effectiveness as they begin to be used more frequently and influence utilization of expensive drugs, and when regulatory authorities are more involved (e.g., FDA labeling changes).

In conducting cost-effectiveness of genetic tests, factors considered include:

1. *The severity and frequency of outcomes of interest*: For pharmacogenomics, this includes the seriousness of drug side effects, duration of the drug prescription, and the cost of the drug and monitoring. For tests looking at disease risk, this includes the health and quality of life impacts of the disease and treatment costs.
2. *Cost* (e.g., test and drug costs, additional clinic visits, genetic counseling)
3. *Alternatives to the test* (e.g., dose adjustments made during periodic office visits, other screening strategies)
4. *Strength of genotype-phenotype association:* Genes that have a higher penetrance are more cost-effective.
5. *Prevalence of the gene variant*: Even small changes in the prevalence of the genotype can have a large impact on the incremental cost-effectiveness ratio. This may have implications in terms of who is covered.

He concluded by emphasizing that cost-effectiveness evaluations for genetic technologies are particularly challenging given the significant data needs. The field would benefit from more precise estimates of the induced costs associated with genetic testing (e.g., the cost of adverse drug reactions) as well as more information on patient preferences and quality of life needs.

Roundtable Discussion

Dr. Joan Reede began by asking for comments on what might be done to address the widening gap between the insured and uninsured in access to genetic technologies. Dr. Williams acknowledged the

existence of a two-tiered health care system in which a large segment of the population does not have access to any medical care, but noted that these access issues are not unique to genetic services. In fact, the nature of genetic disorders may allow children and adults with these conditions to qualify for certain programs and funds such as Medicaid that are unavailable to others. Dr. Tunis remarked that there may be genetic services of high value and cost-effective but that are not currently part of benefit packages. Dr. Williams noted that there are some low-cost interventions such as family history taking and genetic counseling that reduce the number of high-end technologies being utilized, especially in integrated health care delivery systems.

Dr. Willard suggested that as the Committee examines the future of applying genetic and genomic technology for an increasing number of patients, it take advantage of the body of expertise and knowledge that demonstrates that, despite upfront costs, genetic services can produce savings to the health care system in the long run. He also noted that this approach may be better received than putting forth arguments that these services are underpaid. Dr. Veenstra remarked that cost-effectiveness methods have been fully developed and there is much research being done in this area but that it is just beginning to have a role in coverage decision-making. Dr. Schoonmaker said that before we can use cost-effectiveness information, a better way of collecting data is needed to assess effectiveness, such as through better coordination between CMS and the FDA in the evaluation of new technologies. Dr. McCabe noted that the SACGT had examined the tension between FDA's focus on safety and effectiveness and CMS' focus on reasonable and necessary, but because any action would require legislative changes to the agencies' enabling statutes, it opted not to pursue this incongruence further. Ms. Berry added that the five to ten year budget window that the Congressional Budget Office's examines when reporting on the economic impact of proposed laws may also be problematic when trying to demonstrate the cost-benefit of genetic services. Dr. Veenstra further noted that the typical units of measurement used in cost-utility studies – quality of life and life expectancy – are not conducive to this process. Dr. Williams remarked that some testing situations (e.g., for susceptibility to malignant hypothermia reactions following anesthesia) can produce cost savings in short timeframes.

Dr. Feigal requested information on the percentage of the cost of testing that is paid out-of-pocket by patients. Dr. Ferreira-Gonzalez responded that it is hard to track this information within a large academic center where several departments bill under a global billing process. Dr. Leonard explained that her laboratory requests full payment up front from patients outside her university's health system to ensure that it receives full reimbursement, leaving patients to bear the financial cost of underpayment by their third-party payer. Dr. Williams said that about half the time his patients are able to obtain third-party coverage for BRCA or HNPCC testing. Others either must pay out-of-pocket or seek assistance from an institutional fund that has been set up for those in a dire financial situation. Not reflected in these figures are individuals who forgo testing altogether because of the lack of coverage.

Dr. Winn-Deen asked CMS about its current inability to modify payment rates even if a strong economic argument could be made for higher reimbursement. Dr. Thompson responded that although the clinical laboratory fee schedule is frozen until 2009, CMS's inherent reasonable authority is one avenue that could be pursued to address such a scenario. However, this authority could also lead to a reduction in fees. To the extent that a new CPT code is assigned, there is some flexibility on how the price might be set. Dr. Tunis noted that Medicare's lack of use of cost-effectiveness or economic evaluations as part of either coverage or reimbursement has in part resulted from 20 years of intense lobbying by the medical device industry.

In the discussion of cost-benefit analyses for pharmacogenetics, Dr. Joan Reede cautioned the Committee that science may not be able to explain all health risks and we should not overlook the influence of the environment in health and health disparities. Also, Dr. Winn-Deen pointed out that statistically powerful

studies to determine whether there are genetic predispositions for serious adverse drug reactions cannot be ethically conducted.

Dr. Martin Dannenfelser asked whether Medicaid provides any kind of reimbursement for genetic testing. Mr. Thompson and Dr. Tunis described Medicaid as a decentralized program subject to state laws and policymaking with states sometimes relying on the Medicare national policies and fee schedule or to some of the local Medicare policies within the state. CMS does not mandate Medicaid coverage decisions or control payment rates. Dr. Dannenfelser followed up with a question about whether Medicare would cover prenatal testing. Dr. Tunis explained that while uncommon, some Medicare-eligible groups (e.g., individuals with disabilities) might seek prenatal testing.

In response to a question from Dr. Leonard, Drs. Schoonmaker and Williams clarified that FDA approval is generally not required for genetic tests as long as they are performed in a CLIA-approved laboratory, but that private insurers make these decisions individually.

The discussion then turned to the process of obtaining a CPT code and determining payment rates for new codes. Mr. Thompson said that decisions about whether to cross-walk or gap-fill are made on a case-by-case basis with consultation with clinical and contractor staff and public input. Regarding the CPT coding process, the AMA has a formal, deliberative process that can take one to two years. If a new code is needed more rapidly, there are miscellaneous codes available but these have additional administrative burden associated with their use. Dr. Leonard reminded the Committee that because of the nature of CPT laboratory codes, new genetic tests often are billed using existing codes. Thus, new tests are subject to the existing payment rates associated with those codes. Following up on a question from Mr. Margus about whether laboratories are profiting from technological improvements since payment rates were established for these codes, Dr. Leonard explained that while such technologies may exist, academic laboratories generally do not have the capital equipment budgets to purchase such instruments.

In response to a question from Dr. Ferreira-Gonzalez about CMS' inherent reasonableness authority, Mr. Thompson explained that CMS is currently in the process of developing instructions to ensure that the process of changing payment rates is fair and equitable.

March 2

Public Comment Session

Dawn Allain, M.S., CGC
National Society of Genetic Counselors

NSGC recognizes that realizing the full benefit of genetic technologies requires the integration of clinical genetic services into current health care delivery models. SACGHS should identify and promote additional research that will supply the evidence-based outcome data necessary to tackle billing and reimbursement issues. SACGHS and CMS should work with genetic professional organizations and the AMA to establish CPT codes for clinical genetic services and to recommend to the administration and Congress that genetic counselors be incorporated into federal statute and be recognized as allied health care providers. NSGC encourages SACGHS to identify novel methods to increase the number of qualified providers through genetic counseling training programs, medical genetics residency programs, and genetic nursing programs, and to continue to support the educational efforts targeting primary care providers and allied health professionals. NSGC also strongly encourages SACGHS to evaluate achievable goals in order to move forward the objective of improved access to genetics as part of a global

health care program.

Dr. Tuckson questioned Ms. Allain about certification for practitioners and who can bill as certified genetic counselors. The American Board of Genetic Counselors specifically certifies master-level genetic counselors and individuals who have gone through accredited counseling training programs. Dr. Willard asked whether Ms. Allain could recommend concrete steps for increasing the numbers in the pipeline or provide alternate strategies. She suggested that the Federal government could identify areas where genetic training programs can apply for funding to enhance

Joann Boughman, Ph.D.
American Society of Human Genetics (ASHG)

Dr. Boughman updated the committee on legislative action in the area of genetic non-discrimination. The Senate passed the Genetic Information Non-Discrimination Act of 2003, 95 to 0. The challenge, she said, is now on the House side. H.R. 1910 and H.R. 3636 have been introduced but S. 1053 is still being held at the desk and no hearings have been scheduled. ASHG would prefer to see S. 1053 formally introduced in the House and assigned to committee so it could reach the floor for a vote. Dr. Boughman said ASHG has been working with the Coalition for Genetic Fairness, which is chaired by the National Partnership for Women and Families, on strategies and activities to advance the legislation, including the presentation of facts on and examples of genetic discrimination. She discussed the National Partnership's CAPWIZ website, designed to generate letters from the public for Congress, and she encouraged involvement in these efforts.

While reminding Members they may not lobby Congress while functioning as members of the Committee, Dr. McCabe, asked whether there was anything the Committee could do to facilitate these efforts. Members discussed possible strategies to help move the current legislation forward and to identify the arguments against the legislation, noting that the June meeting would be too late to have an impact on this session. Dr. Boughman recommended sending another letter to the Secretary to remind the Administration of the significance of the issue and the short legislative window. Dr. Tuckson said that is it important to identify the opposition and their concerns in order to help the Committee add the necessary data to present a persuasive argument, and he suggested that Dr. Boughman provide an analysis to help the Committee evaluate whether it could capture the information needed and provide it through the Secretary. Dr. McCabe said that the two arguments he has heard against the legislation are the small business argument about the cost of the insurance and the argument that it is already covered under the Americans with Disabilities Act (ADA). The Committee discussed using the visibility of SACGHS to ask individuals who have been discriminated against to provide testimony. Mr. Matthew Bradley of EEOC was asked whether the EEOC could provide information on the ADA and its use for protection against employment discrimination based on genetic information.

Dr. McCabe asked Dr. Boughman to update the committee in June and to help with strategies for moving forward by providing as much specific information as possible. Dr. Boughman said that she will keep SACGHS informed regarding the data and examples ASHG is gathering. Dr. Willard said that to have a few compelling examples of genetic discrimination would be useful. Further discussion of the proposal to send a second letter to the Secretary was deferred until later in the day.

<u>Continued Discussion and Votes on Priority Issues</u>

Pharmacogenomics (ranked tied for eighth/ninth by Members and fourth by *Ex Officios*)

Dr. Winn-Deen began by noting while the significant scientific progress made in pharmacogenomics

through such efforts as the Human Genome Project, the SNP Consortium, and the HAPMAP project, barriers remain. A central question surrounding this issue is what efforts, including research tools to find associations, might help overcome the barriers. She also discussed how large population studies might help move the research process forward. Dr. Steven Gutman mentioned the progress of FDA's work groups in developing guidances on pharmacogenomics data and on multiplex testing. Dr. Gutman emphasized that drug companies must by law submit any well-established pharmacogenomic data that have relevance to the safety and efficacy of drugs, but that because drug companies are confused how to handle the data, particularly, guidance those who are developing a diagnostic in conjunction with a therapeutic product. It also is important to raise the consciousness of drug and diagnostic companies regarding the economic and scientific advantages of studying the diagnostic during the critical drug studies. He said that both documents will likely be reissued within the next few months and that the Committee could review and comment on them.

Dr. Winn-Deen suggested that pharmacogenomics will first be applied to the understanding of the influence of genetics on the response rate and dosing of existing drugs. The second wave will include new drugs developed based on genomics knowledge. Dr. McCabe said that there are research and funding gaps in this area on which the Committee can advise the Secretary. This was voted as a Category 4 issue, with Mr. Margus abstaining.

Genetic Discrimination (ranked third by Members and eighth by *Ex Officios*)

Reflecting its earlier discussion, the Committee agreed that genetic discrimination should be a Category 2 issue.

Genetic Exceptionalism (ranked tied for tenth/eleventh by Members and twelfth by *Ex Officios*)

Dr. Willard framed the issue of genetic exceptionalism by asking whether the information itself is inherently different and whether the ways it is used are inherently different. One approach the Committee could consider would be to create a background document that details these issues in a way that would be useful to the Secretary or to some of the *Ex Officio* agencies; or the Committee could simply say that the issues involved do not differ significantly from those in other areas. Another suggestion was to present the issues related to genetic exceptionalism in the letter on discrimination.

The *Ex Officios* gave this issue a low ranking, so were asked for feedback. Dr. Michael Carome noted that institutional review boards (IRBs) reviewing research involving genetic testing or banking samples for future unspecified genetic research view the research as unique, perhaps with risks that exceed minimal in relationship to other studies, and as possibly requiring additional privacy and confidentiality protections. IRBs could benefit from guidance on reasonable levels of protection. Dr. Carome said that it is not a priority of the Secretary's Advisory Committee on Human Research Protections, and that this Committee has relevant expertise to advise the Department. Dr. Gutman pointed out that while aspects of genetic information may make it unique, the FDA does not focus on them in its regulatory approach. Mr. Daynard remarked that genetic exceptionalism is not an issue at the FTC, all claims are treated the same. The Committee will identify possible opportunities for assisting the agencies, such as OHRP. It was voted a Category 3 issue.

Oversight (ranked tied for eighth/ninth by Members and third by *Ex Officios*)

Dr. Leonard reminded the Committee that SACGT focused a great deal of attention on oversight of genetic testing and genetic testing laboratories, with some impact, and asked whether more attention is required by SACGHS. Dr. McCabe said that much of what SACGT set in motion is moving forward and

being monitored. Dr. Gutman said he regarded the template suggested by SACGT and FDA regulatory changes as works in progress and that it would likely continue regardless of whether or not the Committee chooses to explore the issue further. He pointed out that test stratification is a difficult task and that, for FDA as well as professional groups, the goal is stay abreast of developments. Dr. Tuckson said that it is not clear where the locus of responsibility in government is for advancing the oversight agenda and providing a coordinated and consistent plan. He suggested that the Committee has a responsibility to the Secretary and to others to be attentive to this issue. Ms. Yost provided an update on the addition of genetic-specific components to the CLIA program. She noted that CDC is beginning to develop and fund a model approach to provide coordination and support to develop a process for sustained evidence-based review of genetic tests. The Committee placed this issue in Category 2 issue. Dr. McCabe remarked for the record that he would vote only in the event of a tie.

Direct-to-Consumer Marketing/Direct Access (ranked twelfth by Members and seventh by *Ex Officios*)

Ms. Berry suggested that the Committee focus on direct access to genetic technologies and services since direct-to-consumer (DTC) advertising is already present and accepted in the drug arena. For genetic testing, there is no gatekeeper or physician intermediary as there is with prescription drugs. She suggested they explore whether there are gaps in the activities being undertaken by federal agencies and private entities. Dr. Christopher Hook said these are areas of concern, given the number of claims that are being made in terms of predictive value for certain products. He said that it does not seem to be on the radar screen of the agencies that are empowered to intervene and may warrant quick action through a statement that these should be areas of intervention for FTC and other agencies. Members discussed whether consumer harm has been documented and whether consumers should seek their own information in these areas, and it was suggested that DTC marketing may add to confusion and diminish public confidence in the utility of genetic information. Also, there was concern that people could be driven to use unnecessary health care resources and that they need support in sorting through the various claims and counterclaims.

The FTC and FDA perspectives and roles were presented, with Mr. Daynard suggesting that the SACGT brochure should be resurrected and should tell consumers and doctors to report misleading advertising to FTC. Mr. Miller remarked that the issue of enhancing consumer awareness is part of the larger issue of DTC, and FTC brings to the table tremendous consumer protection expertise, while this Committee and DHHS in general, in combination with NIH, bring the science perspective. One suggestion was to conduct a one-time priority project involving FTC and DHHS holding a press advisory and strategically placing articles and briefs.

The DTC marketing issue was voted a Category 4 topic. Mr. Daynard suggested that to get FTC to place this on its radar screen as soon as possible, timely communication between the Committee and FTC would be helpful.

Patents and Access (ranked tied for tenth/eleventh by Members and eleventh by *Ex Officios*)

Members discussed how the enforcement and licensing of patents are inhibiting the use of genetic information, particularly in diagnostics. The low ranking by Members and *Ex Officios* may have occurred because it was thought that the Committee's involvement would duplicate some of the efforts of the NAS study being conducted over the next 18 months which will assess the impact of patents on research and clinical practice and make recommendations. Discussion ensued regarding whether it should be placed in Category 2 with monitoring or 4, and whether genetic exceptionalism is involved. It was voted a Category 2 issue.

Vision Statement (ranked sixth by Members and fifth by *Ex Officios*)

Members debated the need for a vision statement, with some comments that it may be useful to have an overarching statement that is informative about the Committee's mission. It was suggested that it could be a short executive summary of goals to help focus attention. Dr. McCabe remarked that the Committee's vision statement is included in the charge from the Secretary and that recasting it could be seen as an effort to change the charge. Dr. Willard agreed, but noted that there may be value in writing a one-pager that delineates the issues the Committee has decided to prioritize. Dr. McCabe said that a document could be written that constitutes a progress report of what the Committee has accomplished in its meetings. This was voted as a Category 2 issue.

Coming to Consensus on the Top Three Issues and Developing a Long-Range Work Plan

The Committee reviewed the second prioritization vote and began developing a work plan starting with the top priority in Category 2.

Results of Second Vote, Within Categories

Category 2	**Members**	*Ex Officios*
Genetic discrimination	1	Tied 1 and 2
Education and workforce	2	Tied 1 and 2
Oversight	3	3
Patents and access	4	5
Vision statement	5	4
Category 4		
Coverage and reimbursement	1	2
Large populations	2	3
Pharmacogenomics	3	1
Direct-to-consumer	4	4

Work Plan for Category 2 Issues

Genetic Discrimination

The Committee decided to draft a letter to Secretary Thompson to encourage Administration support for S.1053 and to say that the most expeditious way to address the issue is to move S.1053 through the House. It was clarified that the Committee does not have the authority to lobby the Congress. The Committee was reminded that a Statement of Administration Policy, issued by the White House at the time of the passage of S.1053 by the Senate, indicates that the White House supported the bill and the President would sign it in the form in which it had been passed. The work product will be a letter to Secretary Thompson, and SACGHS will continue to monitor the issue.

Education and Workforce

Dr. Willard said that information could be provided between now and the next meeting by the *Ex Officios* and other interested parties to inform the Committee of actions that could be taken to help in their education and training efforts. Monitoring of this issue should continue. SACGT also had a working

group on education, and the Committee noted that it would be useful to review these earlier efforts. Dr. Collins said he spoke with Mr. Joe McInerny, the executive director of NCHPEG, to get his sense of the gaps found within genetic education of health professionals. He indicated that trying to implement changes in health professional qualifications, licensure, and certification is often a way to move forward. He noted that NCHPEG has not been as successful as it had hoped to be in convincing those responsible for medical and nursing school curricula development to do a better job of incorporating new concepts of genetics and genomics. A statement from SACGHS to AAMC indicating the importance of this could be useful. Mr. McInerny also confirmed the importance of focusing on diversity of the workforce and said that although NCHPEG has identified this as a major priority and has a working group devoted to this issue, they would welcome the chance to interact with SACGHS about possible strategies. There was agreement on the importance of bringing in key groups, such as AAMC, AACN, and ACGME.

Dr. Reed, Dr. Willard, Ms. Zellmer, Ms. Masny and Ms. Harrison were appointed to a task force to draft an education resolution for the June meeting taking into account the elements of diversity, licensure and certification, and curriculum development. The task force will also look at the data collected from relevant groups and sources as a way of identifying the gaps in educational efforts. The Committee will spend time at the June meeting developing the elements of the resolution. Issues related to diversity across the workforce will be incorporated in any resolution. Written documents will be requested before the meeting, and representatives of these groups will be invited to participate in a roundtable discussion with the Committee.

Oversight

The Committee will continue to monitor this issue. Discussion involved the need to keep informed and to hear updates on the various guidance documents that FDA is developing to provide a regulatory framework for the incorporation of genetics into medicine and also on the CLIA regulations when they are published for public comment.

Patents and Access

In light of the work of the National Academies of Science Committee, SACGHS will continue to monitor this issue and the progress of the NAS Committee. They requested that a member of the NAS Committee be invited to present their work to SACGHS when the study is completed.

Vision Statement

Members discussed publishing the issue briefs, prefaced by an introduction summarizing the process up through this meeting. Such a document also would serve as documentation for the Secretary of Committee accomplishments and of where the Committee believes the emphasis should be. It was agreed that the inter-meeting task force would complete the work by drafting the Vision document for discussion at the June meeting. Possible publication vehicles were discussed, and it was agreed that general medicine journals should be considered instead of those specific to genetics. The report will first be submitted to the Secretary, and publication will be sought after the appropriate clearances are obtained.

Work Plan for Category 4 Issues

Large Population Studies

Dr. Collins said that an NIH working group first met in December to discuss a Large Populations Study.

The NIH working group would welcome a liaison from SACGHS and at the appropriate time would welcome a strong endorsement of the need for such a study. A liaison to this group will be identified, and more in-depth discussion could be held during the October 2004 meeting, perhaps for a half day. Dr. Collins will inform members about ways the SACGHS could reinforce the importance of the issue and also identify those who are less enthusiastic about it. The Committee also wants to be informed about other Federal agencies' activities in this area and may want to hear directly from them as well.

Pharmacogenomics

Ex Officios rated this issue as a high priority and were asked to weigh in with the reasons for their enthusiasm. Dr. Gutman talked about the initiatives at FDA and the reconstitution of the Pharmacogenomics Roundtable, which he said would benefit from SACGHS representation. It was noted that a private consortium had sent a white paper to the NIH regarding the forming of a series of national centers for drug discovery and the development of the large clinical trials that would be necessary and it might be useful to hear more about this in the future. The Committee was also interested in understanding how to reach the point where the use of polymorphism data becomes a valuable adjunct to the choice of the right drug for the right patient and the roadblocks involved. The Committee could determine whether there are recommendations it could make about research gaps and regulatory barriers that might be helpful. There are broader issues around pharmacogenomics—beyond providing a safe harbor for pharmaceutical and biotechnology companies so that they will start reporting data—and it would be valuable to hear from companies and from clinicians. Dr. McCabe said that although the decision would be made in June regarding whether this can be done in October, the Committee could begin outlining its efforts in June.

Direct-to-Consumer Marketing

The Committee would like the Secretary to urge FTC to step up its vigilance in tracking DTC marketing and help determine when the issue gets to the point that renewed attention is needed. The Committee and staff can begin to gather related information and share it with FTC. A task force comprised of Dr. Hook, Mr. Margus, Ms. Masny and *Ex Officios* was appointed to draft a short resolution indicating the Committee's concern about the proliferation of direct marketing of genetic tests of questionable scientific validity to consumers. The Committee hopes the resolution will help FTC to invest resources in this area. Further study of this issue will occur after the pharmacogenomics issue is addressed.

Coverage and Reimbursement

Coverage and reimbursement was ranked as the highest priority issue requiring in-depth study and will be the focus of the June 2004 meeting. The Committee will gather data about the major health care systems and other groups, organizations, and agencies and complete a draft identifying gaps and policy options that will be evaluated in June. The Committee will deliberate on the report and develop recommendations at the June meeting with the goal of completing a report on the issue for the Secretary in October. The Committee prepared the following outline of the report:

Report Goal - Improve access to health-related genetic technologies/services by ensuring coverage and appropriate reimbursement in all health care settings.

Introduction - Describe what is unique about genetic technologies and services that has implications for coverage and reimbursement (e.g., genetic counseling, informed consent, molecular basis of CPT codes, problems of uninsured/underinsured, etc.)

Content –

- What is the state of play in coverage of GT/services in private and public settings?
- What are the barriers to coverage in private health plans? In Federal programs?
- Options for addressing each barrier:
  - legislative
  - regulatory
  - private efforts
- What are existing reimbursement mechanisms for genetic technologies/services in private and public sectors? How are they deficient?
- How to effect change?
  - -- legislative
  - -- regulatory
  - -- private efforts

It was noted that, in general, genetic technologies should be referred to as genetic technologies/genetic services. It also should be specified that the Committee is focusing on medically related genetic technologies, not technologies used for other purposes such as forensics.

Two possible communications to CMS were suggested, one involving encouraging the development of CPT codes for genetic counseling and evaluation (exploring beyond AMP's and CAP's position; to be revisited in October 2004), and one involving the need for genetic counselors to be recognized as allied health professionals. It was noted that HRSA does recognize genetic counselors as allied health professionals.

A task force composed of Dr. McCabe, Dr. Leonard, Ms. Zellmer and Ms. Harrison was formed to assist staff in planning the June 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.

/s/ Edward R.B. McCabe	/s/ Sarah Carr
Edward R.B. McCabe, M.D., Ph.D.	Sarah Carr
SACGHS Chair	SACGHS Executive Secretary