The Secretary's Advisory Committee on Genetics, Health, and Society
Summary of Second Meeting
October 22-23, 2003
Washington, DC

Welcome, Opening Remarks

Edward R.B. McCabe, M.D., Ph.D.
Chair, SACGHS

Dr. McCabe opened the second meeting by approving the minutes of the June meeting and asking the Committee to consider the possibility of transforming the meeting minutes into the first report to the Secretary chronicling the current and foreseeable state of genetics research and technology and the issues associated with their use.

Dr. McCabe then gave an overview of the agenda:

- A briefing on the role, activities, and plans of the regulatory agencies regarding oversight of genetic technologies, marketing and laboratories;
- A session on Federal and professional societies and organizations' efforts to enhance the adequacy of the genetics workforce and the education and training of health professionals in genetics;
- A roundtable discussion with representatives of the UK Human Genetics Commission and the Australian Law Reform Commission about their countries' approaches to addressing issues involving genetic technologies and recent reports of their commissions; and
- An update on House efforts to prohibit genetic discrimination in health insurance and employment.

He reminded the members that the Committee had sent a letter to the Secretary urging the Administration to press for passage of the Genetic Information Nondiscrimination Act of 2003 in both the House and Senate, and noted the Committee's delight in the Senate's passage of the legislation and the Secretary's positive response to the letter.

Session on Oversight of Genetic Technologies, Marketing, and Laboratories and the Role of Pharmacogenetic Tests in Therapeutics

Oversight of Genetic Testing Laboratories Through the Clinical Laboratory Improvement Amendments (CLIA)

Judith A. Yost, M.A., M.T.
Director, Division of Laboratories and Acute Care, Centers for Medicare & Medicaid Services

Ms. Yost presented an overview of the regulatory applications of the Clinical Laboratory Improvement Amendments (CLIA) that apply to laboratories providing health-related test results, including genetic test results. The Centers for Medicare & Medicaid Services (CMS), the Centers for Disease Control and Prevention (CDC), and the Food and Drug Administration (FDA) are jointly responsible for administering CLIA. CMS, in particular, is responsible for administrative and operational aspects of CLIA. Laboratories are certified based on their most complex test, which CMS classifies as either waived, moderate, provider performed microscopy, or high. Most genetic tests are classified at the highest level of complexity. To be certified, a laboratory must meet five quality standards: personnel qualifications and responsibilities, quality control, patient test management, proficiency testing, and

quality assurance. Although CLIA attempts to assure analytical validity of laboratory tests, it focuses on the quality of the laboratory and not the individual tests it performs. This focus limits CMS's ability to assure the clinical validity of genetic tests. Regarding enforcement of CLIA, Ms. Yost emphasized that CMS takes an educational approach in encouraging laboratories to be become CLIA-certified is flexible with timelines for meeting quality standards.

Plans for Augmenting CLIA to Specifically Address Genetic Testing Laboratory Issues and Concerns
Joseph Boone, Ph.D.
Associate Director for Science, Division of Laboratory Systems, Public Health Practice Program Office, Centers for Disease Control and Prevention

Dr. Boone provided an update on the progress being made in the development of a genetics specialty under CLIA. In 1997, the Clinical Laboratory Improvement Advisory Committee (CLIAC) proposed the addition of genetic testing as a specialty under CLIA. As a first step, a Notice of Intent was published in 2000, encouraging input from all interested parties. CLIAC reviewed the public comments but found differing perspectives on key issues. For example, while most comments supported the creation of a specialty, half considered the definition of a genetic test to be too broad. The relevant agencies are currently in the process of developing a Notice of Proposed Rule Making that will be published for public comment.

Roundtable Discussion
Facilitator: Emily Winn-Deen, Ph.D.

Dr. Winn-Deen opened the session by asking for questions. Dr. Hunt Willard asked the presenters whether there were any issues unique to genetic testing that are not also issues in other specialties, for instance radiology. Dr. Boone noted that given the significant amount of genetic research in the U.S., there is a public expectation for a specialty in genetics. He also been mentioned that there are broader issues to be considered, such as facilitating reimbursement, that are not unique to genetics. Ms. Yost added that CMS is still assessing its authority to regulate pre-and post-analytical concerns such as clinical validity.

Dr. McCabe asked for confirmation regarding requirements for a foreign laboratory sending test results on U.S. patients. Ms. Yost clarified that any laboratory carrying out testing on U.S. patients must be CLIA-certified. Dr. McCabe noted that this creates problems for individuals in rare disease communities, because often there may be a single laboratory in the world that provides the necessary test but it may not have CLIA-certification. In response to a question about the cost to small laboratories for CLIA certification, Ms. Yost replied that the problems are mainly in perception; the cost is actually quite low beginning at $450 every two years for the smallest laboratories.

Dr. Reed Tuckson asked SACGHS to consider how the Federal government evaluates the overall effectiveness of the system in protecting the public from harms associated with genetic technologies. Dr. Muin Khoury noted that CDC takes a non-regulatory approach gathering data on the full range of parameters of genetic testing, from the analytic performance and clinical utility to ethical issues and utilization rates. Ms. Yost noted that CMS already has a significant amount of data from proficiency testing in CLIA-certified laboratories that is made public every year. She noted that there has been improvement since monitoring began in 1995.

The discussion concluded with questions from Dr. Debra Leonard regarding collaboration with the international community on laboratory standards and CLIA's reach. Ms. Yost noted that there is an international laboratory quality standard that was published recently and CMS is considering adopting that standard as equivalent to CLIA for international laboratories. Dr. Leonard also asked whether CLIA

reached problematic laboratories or whether any CLIA-certified laboratories offered problematic tests. It was noted that CMS requires the participation of all laboratories offering testing for U.S. patients but does not have a formal program to determine that all laboratories that should be complying are doing so.

Impact of Pharmacogenomics on Prescription Drug Labeling
Lawrence J. Lesko, Ph.D.
Director, Office of Clinical Pharmacology and Biopharmaceutics, Center for Drug Evaluation and Research, Food and Drug Administration

Dr. Lesko discussed the role of pharmacogenomics in FDA's regulation of drugs. Pharmacogenomics is the study of hereditary differences in either gene expression profiles at the RNA level or nucleotide sequences at the DNA level for the purposes of better understanding variability in disease phenotypes, disease progression, and dose-response. The ultimate goal is to develop biomarkers to increase the specificity of diagnosing and treating disease. Pharmacogenomic data typically includes microarray or genotyping data. Data on cytochrome P450 enzymes involved in drug metabolism constitute about 75 percent of the data being collected presently. Most pharmacogenomic data are hypothesis generating and exploratory, and except for some of the metabolizing enzyme variants, not well validated.

Pharmacogenomic research initiated a new era that previous regulators did not foresee. FDA drafted Guidance for Industry Pharmacogenomic Data Submissions to help interpret regulations and facilitate progress in pharmacogenomics. Although the regulatory pathway for pharmacogenomics is still unclear, the draft guidance is intended to begin a dialogue between the FDA and industry. Dr. Lesko indicated that sponsors use of the data in drug development will be key to subsequent use of the data in a regulatory scheme. In accordance with Federal regulations, pharmacogenomic data will need to be submitted to FDA if the data is the basis for the sponsors' conclusions on safety or efficacy of the product. The FDA's use of pharmacogenomic data will be based on the validity of the biomarker and will be consistent with the U.S. Code of Federal Regulations. Dr. Lesko also noted that FDA is encouraging voluntary submission of exploratory pharmacogenomic data for the purpose of increasing understanding of the data and using the information to develop regulatory standards.

FDA's Current Approach to the Review, Labeling and Marketing of Genetic Tests
Steven I. Gutman, M.D., M.B.A.
Director, Office of In Vitro Diagnostics, Center for Devices and Radiological Health, Food and Drug Administration

Dr. Gutman discussed regulation of labeling of medical devices. FDA treats all devices the same; it does not take a genetic exceptionalist approach for regulatory purposes. FDA sets labeling requirements and monitors the content of labels to ensure that they are appropriate for approved uses and only for approved purposes. FDA's role in regulating advertising is to ensure that off-label use is not promoted. With regard to genetic tests, FDA requires that packaging for analyte specific reagents (ASRs) include a disclaimer stating that the test has not been approved by FDA and the laboratory is responsible for correctly using the ASR. In addition, Dr. Gutman noted that genetic tests performed in-house may not be sold over-the-counter if an ASR is used in the test.

New trends at FDA include the use of abbreviated 510(k)s and a *de novo* 510(k) process. The agency is focusing their review on relevant endpoints and is fostering a "one-stop shopping" approach. FDA also has adopted a variation of the review template developed by the Secretary's Advisory Committee on Genetic Testing (SACGT) for genetic tests for its review of all devices, allowing the agency to standardize and streamline the regulatory process. The planned adoption of electronic submissions will further streamline the review process.

FDA's Future Plans to Enhance Oversight of High-Risk Genetic Tests
David W. Feigal, Jr., M.D., M.P.H.
Director, Center for Devices and Radiological Health, Food and Drug Administration

Dr. Feigal described FDA's review process for genetic tests. FDA provides basic protections through risk-benefit management (i.e., deciding when first human use is safe, when widespread use is safe, and when adverse experience evaluation and corrective actions, such as recall warnings and market withdrawal, are needed), evidence-based regulatory decisions, and integrity assurance (i.e., enforcement against fraud, bad manufacturing practices, and research misconduct). To perform these functions, the agency uses consumer protection tools, including truth-in-labeling requirements, pre-market safety controls, pre-market efficacy controls, post-market study requirements, post-market event reporting, and standards conformance. To better serve the public and sponsors, FDA has adopted an integrated life-cycle approach to regulating products that begins during a product's investigational use stage and continues through pre-market review, quality systems regulations, medical device reporting, recalls and safety alerts.

Dr. Feigal noted that in-house laboratory testing has a long history and is an established practice regulated by CLIA. FDA regulates ASRs, the active ingredients of some in-house tests. He described a regulatory gap between CLIA, a program that is systems-oriented and focuses on analytical performance and quality control within laboratories, and FDA, which is device-specific and focuses on analytical and clinical performance of the products and requires manufacturing quality standards. The clinical performance requirements of a product regulated by FDA are different from the performance requirements of laboratories regulated by CLIA.

Roundtable Discussion
Facilitator: Debra Leonard, M.D., Ph.D.

Dr. Leonard began the discussion with a question to Drs. Feigal and Gutman about FDA's plans regarding SACGT's recommendation that FDA provide oversight of laboratory-developed (home-brew) genetic tests. Dr. Gutman responded that FDA is using a variation of the template that SACGT produced but has no plans beyond revisiting the ASR rule to increase its regulation of laboratory-developed tests. Dr. Feigal emphasized that CMS regulates these activities through CLIA. Dr. Francis Collins asked whether FDA's legal review of its authority to regulate laboratory in-house tests yielded any conclusions, Dr. Feigal stated that the issue has not been resolved. Dr. Gutman described FDA's plan to review a proposal submitted by the manufacturing community for a new category of devices called *in vitro* analytical tests (IVAT).

A discussion ensued about the use of pharmacogenomic tests in regulatory decision-making. Dr. Lesko confirmed that FDA would prefer that laboratory tests mentioned in drug labels also are FDA approved. However, if the test is available as a home brew, FDA's Center for Drug Evaluation and Research would request information about the test but it would not go through the same review as a test kit. Dr. Lesko also noted that evidence supporting the use of pharmacogenetic tests will have to come from prospectively designed studies.

Dr. Tuckson asked whether FDA considers cost-effectiveness during regulatory decision-making. Dr. Feigal replied that since its focus is on safety and effectiveness, FDA does not consider cost-effectiveness. Given that CMS would be involved in cost-benefit analyses, the Committee wondered how the agencies coordinate the sharing of data. Dr. Gutman responded that the agencies have undertaken some innovative efforts to improve coordination of activities and to communicate on both an informal and semi-formal basis, and he emphasized that this is challenging task but one that is not being ignored.

In response to a question whether companies are expected to provide data from diverse study populations when they seek approval, Dr. Lesko stated that while sponsors already do so for Phase I and II trials, enrollment of diverse populations in Phase III trials has lagged. A lack of ethnic diversity would not necessarily delay approval but may cause the product to have a narrower label.

FTC's Regulations of the Advertising and Promotion of Consumer Products and Its Application to Genetic Technologies
Matthew Daynard, J.D.
Senior Attorney, Advertising Practices Division, Bureau of Consumer Protection, Federal Trade Commission

Mr. Daynard reviewed the role of the Federal Trade Commission (FTC) in oversight of the marketing of genetic tests. FTC is responsible for monitoring advertisements for unfair or deceptive acts or practices. A representation, omission or practice is deceptive if it is likely to mislead consumers acting reasonably under the circumstances and if it is likely to affect consumers' conduct or decisions with respect to the product at issue. A practice is unfair if it causes or is likely to cause an injury to consumers that (1) is substantial; (2) is not outweighed by countervailing benefits to consumers or to competition; and (3) is not reasonably avoided by consumers themselves. He noted that FTC does not interfere with patient-doctor relationships or police the scientific literature.

While FTC regulates health claims, the potential injury to consumers must be serious for FTC to intervene. He noted that no actual harm need have taken place for FTC to act. Mr. Daynard gave examples of FTC's actions in privacy initiatives involving Internet commerce with health claims. In order to have the greatest impact, FTC focuses its attention on products or services that present significant safety concerns, have claims concerning serious diseases, and are national in scope. While FTC thus far has not taken action on a genetic testing advertisement, it is certainly within their mandate to do so should an actionable claim be found.

Roundtable Discussion
Facilitator: Kimberly Zellmer, J.D.

Ms. Zellmer began the discussion by asking Mr. Daynard how an advertisement gets the attention of FTC. Mr. Daynard replied that colleagues in other agencies, such as FDA and NIH, send advertisements to FTC. Dr. Tuckson asked for confirmation about FDA's authority to regulate the marketing of materials for genetic tests. Dr. Gutman responded that FDA does not have authority to regulate laboratory advertisements or labels or direct-to-consumer (DTC) advertising of laboratory tests. Mr. Daynard confirmed that FTC does have the authority in this area but relies on competitors and watchdogs at the state and consumer levels to relay information to the FTC. FTC can address complaints through several mechanisms, including litigation, educational campaigns, and letters to marketers that continue to promote unsubstantiated claims. These efforts are often effective in getting advertisers to voluntarily revise their advertisements. On occasion, FTC will convene health claims "surf days" during which FTC staff search websites for deceptive claims and follow-up with warning letters. This strategy is often successful; 45 percent of deceptive or unfair claims were removed after a recent effort. Dr. Collins noted that even though the problem in genetic testing advertisements may be small at present, taking action now will establish a standard before the problem grows and becomes widespread.

Dr. Leonard asked who decides when an ad for a genetic test is deceptive or may be only partially true. Mr. Daynard said that FTC neither decides nor regulates quality of care, nor does it get involved in ethical issues. Rather, it relies on experts to decide what is reasonable. If an advertisement were for a marketed kit, Dr. Gutman said FDA's concern would be about claims about its safety and effectiveness. He was not sure how FDA would react if the test had no health implications.

Ms. Zellmer raised the issue of direct consumer access to genetic testing as potentially being a greater problem. Dr. Gutman replied that FDA would probably not get involved in direct access concerns unless the laboratory used an ASR in the test. If the laboratory purchases ASRs for use in test or product, they would only be able to promote to consumers but could not actually market the product without a prescription. Mr. Brad Margus and Dr. Tuckson discussed the Committee's faith in consumers' abilities to discriminate between unfounded treatment claims and beneficial information. Dr. McCabe suggested that FTC involve professional organizations in producing its educational materials in order to teach consumers to be more cautious and to ask questions.

Dr. McCabe asked whether FDA would have any authority if there were no health benefits to the tests. Dr. Gutman answered that if there were no health risks, it is possible that FDA would be unable to intervene. The discussion concluded with a conversation about differences between genetic testing and other home kits such as pregnancy tests. Some Committee members felt that genetic tests are different because in many cases the results are not clear-cut.

Public Comment Session

Shirley Jones, Ph.D., RNC
International Society of Nurses in Genetics (ISONG)

Dr. Jones offered comments on genetics and work force education and training. ISONG's mission is to educate and guide nurses on the integration of and application of genetic knowledge. Dr. Jones provided details on the organization's current activities, including its annual education conference and research agenda that addresses delivery and access issues, and the lag time between knowledge and practice. She voiced ISONG's support of the national effort to identify and describe the issues surrounding work force education and development, and encouraged SACGHS to support this initiative.

Dr. Winn-Deen commended ISONG's efforts and asked Dr. Jones to comment on specific actions that SACGHS could take to encourage efforts to enhance nursing education. Dr. Jones replied that there are programs specific to nursing education as well as for other health professionals that have proven successful and funding for such programs would be especially helpful.

Veronica Feeg, Ph.D.
American Academy of Nursing's Expert Panel on Genetic Health Care

Dr. Feeg described several associations responsible for the production of documents on nursing education, including American Association of Colleges of Nurses, the Association for Women's Health, Obstetric and Neonatal Nurses, and the Oncology Nursing Service. She emphasized the role of the 2.7 million nurses involved in the education of the public on health care choices and the integration of genetics in their education.

In response to a question from Dr. Tuckson about distinctions in the appropriate roles of nurses and genetic counselors, Dr. Feeg stated that nurses are trained to conduct discussions with patients about health but not in genetic counseling per se. Mr. Margus inquired about the current state of genetics education of nurses. Dr. Feeg mentioned that there are ongoing studies that require continued support and that there likely are gaps in current knowledge. Dr. Willard asked whether there were different educational considerations for the education of nurses on organ transplants versus genetics. Dr. Feeg responded that genetic education would be somewhat different.

Continued Discussion of Oversight and Pharmacogenetics

Following the public comment session, the Committee returned to its discussion on oversight of genetic technologies and the role of pharmacogenomics, focusing on funding for pharmacogenomic research and mechanisms to encourage funding, how to get data on drugs already marketed, who would pay for such studies, and how retrospective studies might be done. It was pointed out that the Japanese government mandated a pharmacogenomic analysis of all drugs that are on the market and being used in the Japanese population to determine whether their population reacts differently to drugs that were clinically tested on Western populations. Dr. McCabe suggested that the Committee explore what other countries such as Japan are doing in this area and how they are supporting these efforts. Mr. Margus noted that to date the results from most pharmacogenomics studies have been disappointing, but the Committee ought to encourage FDA to keep abreast of this issue. The Committee also noted that multiple constituencies should ideally share available pharmacogenomic data in order to facilitate informed decision-making regarding quality, costs, and safety. The importance of FDA's concept of safe harbor for pharmacogenomic data was noted. The Committee explored how to build the evidence base needed to make full use of pharmacogenomic data for the optimization of drug safety and protection of the public. One suggestion was to use the cooperative group studies model for pharmacogenetics studies of particular subgroups of drugs such as statins. Dr. Alan Guttmacher noted pharmacogenomics is not likely to be widely used until the cost is greatly reduced. He suspected that this will not happen in the next few years but that it is not decades away.

Discussion then shifted to the current understanding of the value of pharmacogenomics in health care. Given the number of uninsured people, and continually rising pharmaceutical costs, it is important to know when pharmacogenomic tools will be available and whether they will be cost-effective. There is an impression that a considerable amount of pharmacogenomic research occurs within the pharmaceutical industry but the data are not publicly available. While it is true that the pharmaceutical industry is spending considerable money on pharmacogenomic studies, few discoveries with any predictive value or utility have been made and since resources are largely being targeted to the drug discovery phase, investments are not likely to result in diagnostic tests. There was interest in trying to determine how to best improve pharmacogenomics' contribution to increased efficiency of health care delivery and reduction in health care costs.

Dr. Tuckson suggested several key areas for further exploration: the pharmaceutical industry's view of pharmacogenomics' potential, the safe harbor issue, the current research infrastructure for pharmacogenomics, and the relationship between public and private sector research dollars. It was suggested that a subcommittee may be needed to help clarify these issues, especially how the government agencies can coordinate the rational use of scarce public resources to answer these critical questions. The Committee also discussed learning more about current pharmacogenomic knowledge, the potential promise of pharmacogenomic research, and the timeframe for realizing the promise. CDC is beginning to address some of these issues by identifying the 50 highest impact genes on health care or health outcomes.

Before concluding the roundtable discussion, the Committee reviewed other pending issues: whether to pursue the issue of oversight of genetic testing further (in general, it was noted that CMS and FDA are moving forward on issues within their agencies' scope of authority but that some marginal problems remain); genetic exceptionalism; the adequacy of Federal protections and monitoring mechanisms regarding genetic technologies; reimbursement for genetic testing; and DTC marketing. Dr. Leonard noted that current billing codes for genetic testing do not provide adequate reimbursement. Regarding DTC marketing, the Committee discussed sending examples of egregious advertising of genetic tests to FTC and suggesting ways to teach the public how to discriminate between legitimate and illegitimate advertising. Although the States regulate direct access to laboratory tests, the Committee discussed the

need for guidance documents that could differentiate tests appropriate for direct access. Dr. McCabe suggested convening an intra-meeting task force to identify and prioritize all of the issues and identify what needs to be done.

Genetic Discrimination in Health Insurance and Employment
Kristin Fitzgerald
Professional Staff Member
U.S. House Committee on Education and the Workforce

Ms. Fitzgerald reported that the recent Senate passage of genetic nondiscrimination legislation (S. 1053) has brought this issue to the attention of the House. She emphasized that this issue is not new and has been addressed by many state laws, as well as by the Health Insurance Portability and Accountability Act (HIPAA), and potentially by provisions in the Patient's Bill of Rights (which did not pass). After a radio address by the President in June 2001, the House held several hearings on the issue of genetic nondiscrimination to address the state of the science, existing laws, and the proposed bills (specifically for unintended consequences). Although the House has not focused on the issue in the way the Senate has, Ms. Fitzgerald indicated that there is general agreement among House members that persons should not be discriminated against on the basis of genetic information. Ms. Fitzgerald commented that legislation in this area is unique in that it is anticipatory and is premised on science that is changing very rapidly. From the House perspective, S. 1053 reflects many improvements including attempts to conform to the HIPAA privacy regulation and to other labor laws in terms of enforcement and damages, and attempts to anticipate and address dual regulatory issues. However, the House is still considering the definitions of genetic information and genetic test, and how these definitions will affect the bill, as well as how the bill interacts with the HIPAA privacy regulation and protections. Although genetic nondiscrimination likely will not be addressed by the House in 2003, Ms. Fitzgerald indicated that staff will continue to work on it and any legislation will follow the typical legislative process in the House.

Roundtable Discussion
Facilitator: Cynthia Berry, J.D.

Ms. Berry led the discussion by asking whether the House voiced any major objections to S. 1053, other than the concern that the bill is anticipatory (i.e., there are apparently few actual cases of genetic discrimination but the fear of such discrimination has negative effects such as discouraging individuals from seeking medically useful services). Ms. Fitzgerald responded that they would like to be sure that the bill has as few unintended consequences as possible, a problem that has been seen with past discrimination laws. To be sure that the legislation addresses a clearly defined problem in the least disruptive manner possible, the members of the House will undertake a thorough evaluation process. The House had been concerned about the amount of damages that could be sought through Equal Employment Opportunity Commission (EEOC) enforcements, but since the provision was dropped from the Senate bill, there is no single issue that is of particular concern for the House. The members are expected to look at all impacts of the legislation and to consider all of its various parts. Dr. McCabe inquired why the bill passed unanimously in the Senate despite the concerns of employers. Ms. Fitzgerald responded that generally there is strong support for anti-discrimination measures, however the House process would probably be quite different from that in the Senate. While the health title of this bill amends HIPAA and, as such, appears very similar to the HIPAA nondiscrimination provisions, the employment title was drawn directly from the Civil Rights Act. Dr. Willard wondered whether the bill has the support of the House leadership and how this might affect its prospects and whether the issue is electorate-driven. Ms. Fitzgerald responded that an issue this complicated generally is not electorate-driven. The House leadership is strongly behind the regular order process. Since the House has not acted on this issue in the past, there is a real need for the members to learn about the issue and deliberate it thoroughly. Ms. Berry asked whether legislation is likely to pass the House before the end of the 108th Congressional session.

Ms. Fitzgerald replied that the process will be serious and since members will be thoroughly studying the issue, they will take more time if they sense a need for more deliberation. Dr. McCabe closed the discussion by reiterating that the Committee would like to continue to monitor this top priority issue.

International Approaches to Addressing Issues Involving Genetic Technologies

UK Human Genetics Commission
Philip Webb
Member, Human Genetics Commission

Mr. Webb provided an overview of the United Kingdom's (UK) Human Genetics Commission (HGC), including its structure and organization, accomplishments, and future plans. HGC was established in 1999 to provide the UK government with analysis of developments in human genetics, including the impact on human health and health care and the ethical, social, legal, economic, and technical implications; to promote dialogue and collaboration; and to advise on strategic research priorities and regulatory frameworks.

In response to its mandate to engage the public in its deliberations, the Commission utilizes a consultative panel of over 100 citizens affected directly or indirectly by a genetic disorder, and surveys the public about concerns and beliefs about genetics. These public consultations have had a key role in the development of recommendations. For instance, in response to a survey of 2,000 individuals, the Commission recommended, and the government agreed to, a five-year moratorium on the use of genetic information in decisions for life insurance policies[1] less than £500,000 in value, while it further studies and develops long-term advice on the issue.

Mr. Webb discussed two of HGC's recent reports: *Inside Information* and *Genes Direct*. The first report on the storage, protection and use of personal genetic data makes 38 recommendations, including the criminalization of the acquisition of another person's genetic information without consent, which the UK government has since agreed to implement. Other recommendations in *Inside Information* relate to genetic discrimination and the need to maintain separation between the UK's criminal DNA database and the UK Biobank database, a research project that aims to collect 500,000 samples from individuals between the ages of 45 and 69 to discern the relationship between various genes and health disorders. Its second report, *Genes Direct*, focuses on genetic services offered directly to the public without the involvement of a medical practitioner and discusses the increasing availability of genetic tests on the Internet, some of which lack scientific evidence of clinical validity. The report recommends stricter controls, but not a ban, on the provision of genetic tests and that a reliable source of information about genetic tests be available to the public.

Genetic discrimination, paternity and genealogy testing, and genetics and reproductive decision-making (e.g., genetic profiling of newborns) are among the areas that HGC will address in the future.

Australian Law Reform Commission
David Weisbrot, J.D.
President

Mr. Weisbrot provided an overview of the Australian Law Reform Commission (ALRC) and its recent report, *Essentially Yours: The Protection of Human Genetic Information in Australia*. ALRC has existed

[1] In the UK, genetic discrimination in life insurance is of greater concern than health insurance because of its link to house purchases and mortgages. Genetic discrimination in health insurance is not as great a concern as it is in the US because the UK has a national health system.

for 28 years, conducting legal research and public consultation for the Australian government. *Essentially Yours* responds to the request of the Attorney General for advice on how best to protect privacy, prevent unfair discrimination, and ensure adherence to ethical standards. ALRC also engages in extensive public consultation as part of its deliberative process, including public forums and targeted meetings around the country, surveys, requests for written comment, and the development of consultation documents.

Essentially Yours includes 144 recommendations addressing a wide range of issues, including oversight of medical research; genetic databases; genetic discrimination in life insurance and employment; disclosure of genetic information to family members; and DNA collection for law enforcement, educational, immigration, kinship, paternity, and identification purposes. ALRC's recommendations recognize a need for a SACGHS-like body to provide independent, authoritative advice to the government through a public process on these and other genetics issues, such as the identification of sensitive genetic tests that may require restricted access and counseling; education of the public and health providers about genetics; and fortification of the health care delivery and financing system to allow for the integration of genetics into medical practice.

Roundtable Discussion
Facilitator: Christopher Hook, M.D.

Dr. Hook began the roundtable discussion by asking the presenters whether any issues require international collaboration. Mr. Webb responded that Internet-based genetic testing services are of particular concern and would benefit from cooperation. Dr. Arden Bement followed with a question about whether the two commissions had explored accreditation of laboratories and conformity to validity standards. Mr. Weisbrot remarked that, in Australia, sensitive genetic tests are performed only at certain public hospitals to optimize the quality of the tests. In response to Dr. Leonard's query whether laboratories in Australia were responsible for also providing genetic counseling, Mr. Weisbrot stated that laboratories are not responsible for directly providing counseling services but they should have a referral system in place. Dr. Tuckson inquired about HGC's interaction with the UK government, to which Mr. Webb responded that HGC reports to both the health and science ministers, and the placement of HGC staff in the Department of Health facilitates the effective transmittal of the Commission's recommendations.

Session on Genetics Workforce, Education, and Training

Framing the Issues
Joann Boughman, Ph.D.
Executive Vice President, American Society of Human Genetics
Chair, Former SACGT Education Work Group

Dr. Boughman opened the session by framing the issue of genetics education and training and previous efforts to address the issue. Dr. Boughman described the magnitude of the challenge of adequately educating health professionals in genetics to assure the appropriate integration of genetic technologies into the health care system and asserted that rapid advances in genetics, consumer and patient demand, and cost considerations drive the need to do so. She stated that major gaps currently exist in health professionals' knowledge, education, and training in genetics and that these gaps will limit the integration of genetics into all aspects of health care. At the same time, much progress has been made with respect to the many recommendations issued in this area by national organizations, including the National Academy of Sciences (NAS), the Institute of Medicine (IOM), the NIH-DOE Task Force on Genetic Testing, and the SACGT Education Work Group.

The 1975 NAS report recommended curricular and continuing education changes to reflect genetics, an increased focus on preventive medicine, and funding to support these activities. The 1994 IOM report stated that health professional education needed to be overhauled to look at medicine from a genetics point of view and to consider the ethical, legal and social implications of genetic knowledge and advancements. The report also made recommendations for improving the genetics curricula, strengthening training programs, and developing physician competence through continuing medical education. The 1997 report of the NIH-DOE Task Force on Genetic Testing focused on increasing genetics in medical school and residency curricula and on the role of continuing medical education (CME) in developing physician competence. In 2002, the IOM, in the report *Who will keep the Public Healthy? Educating Public Health Professionals for the 21st Century,* recommended that all schools of public health incorporate genomics into their curricula. The SACGT Education Work Group emphasized collaborations in outreach, an educated public and creative, interactive, and accessible programs. Dr. Boughman noted that many people are working hard on closing the identified gaps in the education of health professionals in genetics.

Federal Efforts to Address Genetics Workforce, Education, and Training Issues
Sam Shekar, M.D., M.P.H.
Associate Administrator, Bureau of Primary Health Care, Health Research and Services Administration (HRSA)

Dr. Shekar provided an overview of Federal efforts to address genetics workforce, education, and training issues. Dr. Shekar stated that the current time it takes to translate relevant science into clinical practice is unacceptably long. Effectively translating genetics into everyday practice requires educating more than 11 million health professionals. Although the primary responsibility for educating and training professionals lies in the non-Federal, private sector, the Federal Government currently is undertaking many activities in support of these efforts.

Dr. Shekar reviewed the results of a survey of the 16 ex officio agencies about their activities in genetics education and training and workforce enhancement. The survey identified over 180 relevant activities supported from fiscal years 2000-2003. Given the increased expectations resulting from the completion of the Human Genome Project and its effect on the way we view disease, these efforts address the fundamental need to facilitate the translation of genetics into practice. Most of the effort at the Federal level is targeted at health professionals. Major types of activities include dissemination of information, educational outreach, consensus building, and generalized training. Across the four fiscal years, spending on these activities totaled more than $102 million, with expenditures increasing each year. These efforts tend to be supported by discretionary funding. The agencies identified two action items in their surveys: describing the appropriate use of genetic technologies and helping with across-the-board integration of genetic technologies into health care and public health systems.

Preliminary Findings of the Genetics Workforce Study
Judith A. Cooksey, M.D., M.P.H.
Adjunct Associate Professor, Department of Epidemiology and Preventive Medicine, University of Maryland School of Medicine

Dr. Cooksey reported on the preliminary findings of the genetics workforce study co-funded by the National Institutes of Health (NIH) and the Health Resources and Services Administration (HRSA). Dr. Cooksey explained that the study assesses who is qualified to provide genetic services to patients, and what roles the various health professionals play in the delivery of these services. Many factors influence qualification to provide genetic services, including competency, regulation, local market factors, setting and organizational structure for care delivery, inter-professional collaboration, and reimbursement. The study also aims to assess the way genetic services are delivered nationally, to describe models for

delivery, and to analyze factors driving change. The study focuses on clinical genetics specialists, physicians that provide genetic services (including primary care physicians), nurses and, to a lesser extent, other health care professionals that provide genetic services.

The study consists of several components. One part of the study, a survey of medical geneticists, found that this professional group is very small with limited patient care time, is pediatrics-oriented, and generally conducts time-intensive patient visits that from a reimbursement perspective are not highly productive. A genetic counselor workforce study concluded that expansion of genetic counseling programs could not be supported due to reimbursement issues, underemployment, and current employment in academic health centers. A genetic services models component found that genetic tests, individual leadership, institutional vision, geneticists' roles, laboratory competition, the nature of services (e.g., outpatient, cognitive), and safety net funding all influence the organization and delivery of genetic services.

Overall and to date, the study has found that:

- Clinical genetics is a very small specialty area in medicine. Most services still relate to counseling, testing, and test interpretation. It is not clear whether the small number of genetic specialists is, in fact, a problem.
- The diffusion of innovation and new technology typically progresses with specialists from the academic health center (which trains specialists) to the broader medical community. Often diffusion to primary care providers is slower.
- The market exerts a strong influence over clinical genetic services through reimbursement, competition, and financial incentives and is restrictive for cognitive services.
- Availability of genetic tests affects the demand for services.
- A "big-picture" approach to the issue of genetic services delivery is lacking.

Roundtable Discussion
Facilitator: Huntington Willard, Ph.D.

The roundtable explored a number of issues with respect to the genetics education and training of health professionals. In response to a question about whether evidence exists to show a measurable increase/improvement in genetics education over the past 10 years, Dr. Boughman stated that measuring improvement, especially in the practice setting, and assessing the adequacy of the genetic-based education in health professional schools are very difficult tasks, but there are some methods that can at least provide a baseline. Although the basics of genetics are being taught (in the form of didactic lecture), it is not clear that genetics is being incorporated effectively into the clinical training of practitioners. If one approaches the problem of an appropriate genetics workforce from the perspective of genetic specialists, it might appear that the battle has been lost since there are only about 1,000 clinical geneticists out of roughly 560,000 specialists. This number might be explained by the fact that there is not a great deal of prestige (as defined through market forces) associated with being a clinical geneticist, or alternatively, that genetics is a component of many specialties. Dr. Leonard pointed out that thinking about genetics only as a specialty area is not consistent with the future model of genetic medicine since all physicians will have to practice genetics. However, genetic specialists clearly have an important and unique role in this future.

Although all physicians will have to incorporate genetics into their practices in the future, genetic specialists will continue to be relied on for difficult cases. The perception of geneticists as only diagnosticians may need to change in order to appeal to medical students who wish to improve the lives of their patients through direct action. Also, reimbursement must be developed to provide incentives to both primary care practitioners and specialists to begin to incorporate genetics into their practices.

Public Comment Session

Paul Billings, M.D., Ph.D.
Laboratory Corporation of America

Dr. Billings noted that empowered, educated consumers drive demand for genetic tests and that there is no evidence that the current system harms or dissatisfies consumers. His remarks focused on the issues of coverage and reimbursement of genetic tests and genetic discrimination.

Chin-To Fong, M.D.
University of Rochester School of Medicine and Dentistry

Dr. Fong discussed the increasing need to keep abreast of advancing genetic knowledge. He emphasized the importance of minimizing the knowledge gap between interested groups such as genetic professionals and the general public. He discussed Project BEGIN (Biotechnology ELSI/Genetics Instruction Network), which uses teachers to pass knowledge down through mentoring networks and to disseminate curriculum to the schools.

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

Ms. Allain commented on the lack of genetic counselors in many areas of the country and encouraged support for the Allied Health Reinvestment Act as well as other federally funded mechanisms for genetic services in underserved populations. Ms. Allain stressed that education in genetics should be tailored to each medical specialty encouraged the Committee to attend to reimbursement issues and state/board licensure of genetic counselors.

Larry O'Connor, M.D.
American Medical Association

Dr. O'Connor described the American Medical Association's activities and collaborations with the American Bar Association and the American Association for the Advancement of Science to educate their members about genomics and its role in clinical medicine. Dr. O'Connor said that AMA was interested in providing primary care physicians with tools for integrating genetics into patient services.

Sharon Olsen, M.S., RN, AOCN
Oncology Nursing Society

Ms. Olsen discussed the Oncology Nursing Society's (ONS) 1995 strategic plan that focused on integrating genetics education and practice initiatives. She highlighted the many continuing education courses available in genetics and encouraged the Committee to support the Nursing Reinvestment Act and other federal programs that will address the projected nursing shortage.

Michael Rackover, PA-C, M.S.
American Academy of Physician Assistants

Mr. Rackover discussed the need to have mentors facilitate the application of genetic knowledge into medical practice and emphasized the need for the use of family history in family practice.

Fred Ledley, M.D.
Mygenome

Dr. Ledley described many of the barriers in addition to genetic discrimination that prevent the health care system from proactively meeting consumer interest in predictive genetic testing. These include inadequate training of professionals, demographics of healthcare utilization, the lack of incentives for payers and provider to create infrastructure, inadequate resources, and the lack of trust in healthcare systems' use of genetic data. Dr. Ledley emphasized the consumer's role in decision-making. He asked the Committee to be innovative but to also take care that its recommendations not inadvertently create new barriers.

Continuation of Session on Genetics Workforce, Education and Training Issues

Overview of Efforts by Organizations and Professional Societies to Address Genetics Workforce, Education, and Training Issues
Joann Boughman, Ph.D.
Executive Vice President, American Society of Human Genetics

Dr. Boughman described the activities of the many organizations and professional societies addressing genetics education at all levels, including general education, undergraduate education, physician education (medical school, residency, continuing medical education), geneticist education, and health professions education. Groups involved with education at these various levels include the Association of American Medical Colleges (AAMC), American Board of Medical Specialties (ABMS), American Society of Human Genetics (ASHG), American College of Medical Genetics (ACMG), American Board of Genetic Counselors (ABGC), and American College of Graduate Medical Education (ACGME).

Activities include developing medical school curricula to incorporate genetics, developing genetics-related questions on the national boards, incorporating genetics into practice in residencies and CME modules, and integrating new genetics concepts into the maintenance of certification (MOC) process. Residencies in pediatrics, OB/GYN, internal medicine, neurology, and family practice have incorporated approved genetics curricular elements to assure that physicians trained in these areas have a basic working knowledge of genetics. Several formal combined residency programs in pediatrics/medical genetics, internal medicine/medical genetics, and pathology/medical genetics have been approved and three others in psychiatry, obstetrics/gynecology, and family practice are pending.

Current gaps include teaching genetics concepts in college, a declining specialist pool, insufficient training faculty, understanding genetics and common disorders, and large (and growing) information gaps for specialists. Key areas that need more work include developing teaching models (curricular guidelines/elements, professional guidelines, programming for the specialties, and faculty development); educating the public, patient and consumer; enhancing undergraduate education and premed requirements; and involving and educating the media. There are several barriers to addressing these issues, including a perceived lack of relevance of genetics to practice; a current focus on rare diseases and disorders; an overcrowded curricula; and the probabilistic nature of genetic information.

Educational Programs for Health Professionals: Addressing the Gaps
Joseph D. McInerney, M.S.
Director, National Coalition for Health Professional Education in Genetics

Mr. McInerney described the many ongoing activities in the education of health professionals in genetics. The National Coalition for Health Professional Education in Genetics (NCHPEG), an association comprised of over 127 member organizations, has developed a set of core competencies for health professionals in genetics that have influenced many of the educational efforts currently underway.

NCHPEG has an annual meeting, maintains a web site called GROW (Genetics Resources on the Web), and develops a web-based program annually for a particular discipline, in addition to many other specific projects. NCHPEG supports the view that genetics helps build bridges between health professionals with an interest in risk factors at the population level, e.g., epidemiologists and public health practitioners, and health professionals focused on risk factors in individuals, e.g., the primary care provider. NCHPEG also believes that genetics education for the health professional and the public must be congruent.

Mr. McInerney explained that the first critical challenge NCHPEG faces is not in conveying specific genetics content but rather in addressing how genetics can be utilized in actual practice. This will require people to focus less on genetic disease and more on the role that genes play in the expression of disease, to look at "health care through a genetic lens."

One key theme that is apparent when looking at the current activity in this area is the lack of coordination. Many guidelines promulgated by various societies are played out in different ways in different institutions. While a variety of approaches allows for considerable flexibility by specialty, there may be a need for an overarching mechanism that specifies what basic content is needed. Several practical constraints, including variation in state-by-state regulation of practice and varying levels of education, may make a single approach unfeasible.

Genetic Counselor Training Program Capacities and Needs
Robin Bennett, M.S., CGC
Past President, National Society of Genetic Counselors

In June 2003, SACGHS asked the National Society of Genetic Counselors (NSGC) to explain what is needed to increase the number, diversity, and quality of training of genetic counselors. In response to this request, Ms. Bennett reviewed the field of genetic counseling and the current status of genetic counseling programs and offered suggestions for expanding existing programs and developing new programs. Currently, there are 25 genetic counseling training programs in the U.S.; 11 new programs are under consideration. It is likely that certain geographic regions will always lack such programs since most programs are associated with an academic medical center. Although there has been a steady increase in the number of genetic counselors, and a steady rise in the number of jobs, Ms. Bennett indicated that it is difficult to predict the demand for genetic counseling. Generally, the counseling community has been able to meet the demand.

There are several limits on genetic counseling training programs, including a lack of quality field placements, the lack of funding for programs, and limited scholarship opportunities. NSGC has tried to address these problems by increasing the capacity of existing training programs and enhancing existing programs by adding additional training sites, adding faculty, developing training grants for students, increasing the number of programs, and increasing access to expert training (for example through web based teaching). Additional funding for genetic counseling training programs would help toward increasing the diversity of the field, client access, and access to genetic counselors by other health professionals.

Roundtable Discussion
Facilitator: Reed Tuckson, M.D.

Dr. Tuckson began the roundtable discussion by focusing attention on questions related to certification and competence, i.e., how do we decide who is competent to provide which services? Dr. Boughman responded that the question of competence is generally decided based on the definition of the service being provided. The coding process for reimbursement of these services will not specify which provider performs the services ("as done by"), since the code itself focuses on the activity, and not the process by

which the activity is accomplished. Ms. Bennett added that to date legislative proposals at the State level on licensure generally have stipulated that genetic counselors providing services must work under the supervision of a physician which, given the complexity of genetic testing, counselors view as appropriate. Dr. Boughman noted that other health care professionals can in fact provide genetic counseling services as well (such as pathologists who go through a special year long training process). Although there are many professionals who are capable of doing some genetic counseling, a certain level of competency is needed to provide these services effectively. Ms. Bennett suggested that in general when genetic testing is involved, only medical geneticists and genetic counselors should provide counseling. Ms. Robinsue Frohboese pointed out the lack of diversity among providers of genetic services as well as among the research and clinical populations. This is a major issue as is trying to assure that genetic services reach underserved and minority populations.

Several points regarding responsibility for genetics education and training were made throughout the discussion. Dr. Boughman pointed out that only 78 of 175 medical genetics residency slots are filled due to lack of funding. Dr. Willard pointed out that any hospital can add new residency slots, but in order to do so, it must self-fund them. Dr. Winn-Deen suggested that if there is a defined need in a state for more genetic counselors, then the state must respond to that need by funding efforts to establish genetic counseling training programs within the state. When Ms. Bennett pointed out that genetic counselors volunteer their time to teach and mentor students, Dr. Willard suggested that support for teaching and mentoring should be a matter of academic and hospital leadership, not a Federal responsibility. Dr. McCabe noted that HRSA considers genetic counselors to be allied health professionals and, as such, genetic counseling programs qualify for funding under Title VII.

SACGHS Discussion of Next Steps and Plans for the March 1-2, 2004 Meeting

The Committee decided that it was important to engage in a systematic process to determine the priority issues that should be the focus of SACGHS' work over the next year. Dr. McCabe established a Task Force to carry out the issue identification process and to help plan the agenda for the March meeting. The Task Force's work before the March meeting involves the following key steps:

1) Review of the possible issues;
2) A straw vote of the members identifying the three to five top priority issues that they wish to see SACGHS address;
3) Identification of the ten most often cited issues or questions; and
4) Development of background information on each of the top ten issues.

At the March meeting, the Task Force will present the top ten issues, and from these ten issues, the Committee will identify two to three issues that will become the Committee's priorities for the next year. The Committee will also identify those issues that may only require monitoring, rather than action. Cynthia Berry, Debra Leonard, Barbara Harrison, Reed Tuckson, Hunt Willard and Emily Winn-Deen volunteered to serve on the Task Force.

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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.

Edward R.B. McCabe, M.D., Ph.D.
SACGHS Chair

Sarah Carr
SACGHS Executive Secretary