The development and use of genetic technologies raises a broad range of human health and societal issues. In 2002, the U.S. Secretary of Health and Human Services chartered the Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) to explore, analyze, deliberate and provide advice on these issues. SACGHS’ membership is diverse and multidisciplinary. Its 13 members include experts in molecular biology, genetics, medicine, nursing, public health, health insurance, law, ethics, and consumer advocacy. The Committee’s activities are informed and guided by representatives of 16 federal agencies and departments. This breadth of expertise and input is critical to the Committee’s ability to provide reasoned assessment of the wide range of issues it may potentially consider.

At its inaugural meeting in June 2003, SACGHS heard presentations on current and future applications of genetic technologies and the wide range of issues raised by the development of those technologies. A major goal of the meeting was to identify high priority issues for further deliberation and study. The Committee expressed serious concerns about the potential misuse of genetic information by health insurers and employers and took a strong stand in support of federal legal protections against genetic discrimination in these settings. The Committee also identified oversight of genetic technologies, genetics education and training of professionals, and the adequacy of the genetics workforce as areas of interest, and requested briefings on these topics. Informational presentations and discussions on these topics thus formed the central elements of the agenda for SACGHS’ second meeting in October 2003. Time was also taken to explore how two other countries, United Kingdom and Australia, are addressing issues related to genetic technologies. This article provides a summary of the topics discussed at the October meeting and outlines the Committee’s next steps in its priority setting process.

Oversight of Genetic Technologies, Marketing and Laboratories
Federal oversight of genetic technologies, marketing and laboratories is handled by several Federal agencies and regulatory schemes. The Centers for Medicare & Medicaid Services (CMS), Centers for Disease Control and Prevention (CDC), Food and Drug Administration (FDA) and the Federal Trade Commission (FTC) each have jurisdiction over aspects of the development, use and provision of genetic technologies for health purposes and all share responsibility for the oversight of genetic technologies. The Committee heard briefings from agency officials involved in the management of these programs.

The first group of presentations focused on the oversight of clinical laboratories, which CMS, CDC and FDA jointly administer through the Clinical Laboratory Improvement Amendments (CLIA). CLIA regulations are aimed at ensuring the quality of laboratory testing, and they cover all laboratory testing performed on humans in the US for health purposes.

The main provisions of CLIA, which require laboratories to meet five quality standards relating to personnel qualifications and responsibilities, quality control, patient test management, proficiency testing and quality assurance, were reviewed by Ms. Judith Yost, Director of CMS’ Division of Laboratories and Acute Care. CLIA’s focus is on assuring the analytical validity of laboratory tests, not their clinical validity or utility. Ms. Yost emphasized that laboratories providing genetic tests are already covered by CLIA. However, since CLIA does not have a molecular specialty, explicit standards for molecular tests are not specified in the regulations.

In 1997, a DHHS advisory committee, the Clinical Laboratory Improvement Advisory Committee (CLIAC), proposed that CLIA be augmented to provide specific requirements for laboratories conducting genetic tests in order to provide greater assurance of quality. Dr. Joseph Boone, CDC’s Associate Director for Science in CDC’s Division of Laboratory Systems, noted that when the CLIA regulations were first promulgated, the genomics revolution was not yet in full swing. With the ensuing development of molecular approaches to testing, CLIAC undertook a comprehensive analysis of the regulations to assess what additional specific requirements were needed to address this growing component of the diagnostic field. As a first step toward adding a genetic testing specialty, CDC issued a Notice of Intent in 2000 seeking public comment on the proposal.[1] As a second step, a Notice of Proposed Rule Making will be issued for public comment in the near future.

A second group of presentations featured FDA officials and focused on FDA’s role in regulating in vitro diagnostic devices. Genetic test kits and analyte specific reagents (ASRs), the active ingredients in tests, are considered in vitro diagnostic devices. Manufacturing quality standards, labeling and, to a lesser extent, advertising of devices are also under FDA’s purview. Dr. David Feigal, Director of FDA’s Center for Devices and Radiological Health, noted that genetic tests developed in-house and offered as a service (so-called “home brew” tests), which account for the vast majority of the genetics tests done yearly in the US, are primarily regulated by CLIA, not FDA. Home brew tests that include ASRs are subject to some FDA regulation through a rule
finalized in 1997 known as the ASR rule. Dr. Steven Gutman, Director of FDA’s Office of In Vitro Diagnostics, explained that the ASR rule classifies most ASRs as Class I devices; Class I devices are required to meet certain general requirements, such as following quality systems regulations and restricting sales to laboratories designated as high complexity under CLIA, but are not subject to premarket review by the agency. Dr. Gutman reported that the FDA is planning to revisit the ASR rule to determine whether the current classification system is appropriate and that the rule provides sufficient safeguards on ASRs. In addition, it is becoming apparent that some new genetic technologies may not fit the definition of an ASR due to their complexity and intended uses. The agency does not, however, intend to classify genetic tests per se differently from other tests; the classification will be based on an assessment of the risk posed by the test.

FDA’s use of pharmacogenomic tests in the drug review process was described by Dr. Lawrence Lesko, Director of FDA’s Office of Clinical Pharmacology and Biopharmaceutics. Currently, most pharmacogenomic tests have not been sufficiently validated to be included in the FDA drug approval process. Dr. Lesko indicated that the agency is endeavoring to promote broader use by expanding its own knowledge and understanding of the technology. The agency is requesting voluntary submissions of pharmacogenomic data and developing guidance for industry on the submission of the data. Dr. Lesko indicated that the processes of drug development and relevant pharmacogenomic test development ideally should occur concurrently.

The oversight of the marketing of genetic tests is the responsibility of both the FTC and FDA. FTC monitors advertisements for unfair or deceptive acts or practices and is authorized to take action against those who deceive or cause injury to consumers. To maximize its impact with limited resources, Mr. Matthew Daynard, Senior Attorney in FTC’s Advertising Practices Division, explained that the agency focuses its attention on products or services that present significant safety concerns, make claims about serious diseases, and are being marketed on a nation-wide basis. FDA is primarily responsible for assuring that labeling is accurate and promotional material does not promote "off-label" use. The ensuing discussion highlighted that fact that home-brew tests not using ASRs fall into a regulatory gap since FDA does not monitor labeling and promotion of these tests. While FTC could regulate the advertising of home-brews marketed as services, it has not done so up to this point.

The Committee did not draw any firm conclusions about the adequacy of current government oversight of genetic tests and its priority as an issue for further Committee study is still being explored.

**Genetics Education and Training and Workforce Analysis**

As more genetic technologies become available for clinical use, efficient, appropriate and equitable integration of genetics into health care and public health becomes even
more critical. Achieving appropriate and optimal use of genetic technologies depends heavily on the adequate education and training of health professionals in genetics.

The session on genetics education, training and workforce was introduced by Dr. Joann Boughman, Executive Vice President of the American Society of Human Genetics (ASHG) who framed the issue, reviewed the recommendations of prior studies of the issue, and highlighted some of the progress made to date in this area. She concluded by noting that educating health professionals to be able to successfully integrate genetics into their practice poses an enormous challenge.

This session’s presentations reviewed Federal and private sector efforts currently underway in the Federal government and the private sector to enhance genetics education and training of professionals. The results of a survey of SACGHS’ 16 ex officio agencies and departments on the number, nature and outcome of federal efforts to enhance genetics education and training and to analyze genetics workforce needs were presented by Dr. Sam Shekar, Associate Administrator of the Bureau of Primary Health Care at the Health Resources and Services Administration (HRSA). Currently, there are more than 180 relevant activities being conducted within seven of the reporting agencies with over $102 million dedicated to these efforts. Federal efforts generally address the need for facilitating the translation of genetics into practice and emphasize access through this translation and appropriate integration.

Information on the many efforts being undertaken in the private sector by both organizations and professional societies was presented jointly by Dr. Boughman and Mr. Joseph McInerney, Director of the National Coalition for Health Professional Education in Genetics (NCHPEG). Professional sector efforts to educate physicians have focused on curricular changes, continuing medical education, combined residencies and licensing exam changes, and generally require a commitment from top leadership. Dr. Boughman also described some of the barriers to enhanced education and training in genetics. These include an overcrowded curriculum, the inherent complexity of the probability and risk subject matter, and a perceived lack of necessity or relevance of genetics to medicine and health care. These gaps continue to grow, in spite of focused efforts to close them, because of the rapid increase in genetic knowledge. Although many health professional groups are seeking help in the form of curriculum guidelines, course content, and workshops, the heterogeneity of the educational levels of the practitioners (MD, RN, BA, MS, etc.) and state laws regulating scope of practice, among other issues, can sometimes pose challenges to the development of suitable educational materials.

The balance between specialists and generalists, the role of the genetic specialist in the future, and the best model to realize the most effective workforce are all issues of concern when considering the genetics workforce, said Dr. Judith Cooksey, University of Maryland School of Medicine, who is the principal investigator of a workforce study on genetic services and the genetics workforce, funded by HRSA and the NIH. Dr. Cooksey’s study has preliminarily determined that most genetic services relate to counseling, testing and test interpretation and the number of genetic specialists is quite
small, but it is not clear whether this is adversely affecting the delivery of genetic services. The study aims to identify the factors that drive the organization and delivery of clinical genetics services, with the ultimate goal of defining the most efficient and effective delivery model.

The role of the genetic counselor, an important component of the genetics workforce, is evolving as genetics becomes more integrated into medicine, public health and health care. Ms. Robin Bennett, Past President of the National Society of Genetic Counselors, presented information on what is required by graduate programs in genetic counseling to increase the number, diversity and quality of training of genetic counselors. Specifically, there is a need for additional programs, student as well as supervisor stipends, additional faculty, diversity scholarships, and increased access to expert training. There is currently an overall lack of funding for programs as well as limited scholarship opportunities for genetic counseling students.

Overall, the presentations provided the Committee with a deeper understanding of the broad scope of current genetics education activities being undertaken through an array of approaches by a wide variety of organizations. They also made it clear that addressing the gaps in genetics education and training and workforce analysis will be a significant challenge. The Committee is continuing to explore what priority to place on genetics education and training as an issue for further Committee study.

International Approaches to Genetics Issues

A number of other countries, including the United Kingdom (UK) and Australia, have established advisory bodies to respond to concerns in a manner suitable to their population needs and social and political structure. The global nature of genetics issues and product markets encourage countries to share information, communicate about and collaborate on solutions, and learn from each other’s experiences in addressing similar problems. In an effort to build bridges between the US and other nations that are grappling with similar questions, SACGHS invited representatives of the UK Human Genetics Commission (HGC) and the Australian Law Reform Commission (ALRC) to report on the work of their committees and relevant policy developments in their countries.

The HGC was formed in 1999 to provide the UK government with advice on the ethical, legal, economic and social implications of advances in human genetics. Its work products include *Inside Information*, a report on the storage, protection and use of personal genetic data, and *Genes Direct*, which offers recommendations on the provision of genetic services direct to the public. ALRC was established in 1975 as an independent federal statutory corporation that provides advice to the Australian government on federal laws and legal processes. The ALRC recently completed a comprehensive review of the protection of human genetic information in Australia, entitled *Essentially Yours*.
The presentations identified a range of issues that are of shared concern. Genetic testing without the involvement of health providers is a concern shared by both the US and UK. This issue will likely benefit from international collaboration because the global reach of the Internet will make national laws that stop at countries’ geographic borders unenforceable and largely irrelevant. Other areas of mutual concern are privacy and confidentiality of genetic information, the adequacy of health professionals’ and the public’s knowledge about genetics, and genetic tests of questionable validity. Genetic discrimination also is an issue in all three countries. Whereas the concern in the US focuses primarily on health insurance, in the UK and Australia, two nations with universal health care systems, the focus is on life insurance.

Information about the HGC and ALRC, including copies of their reports, can be found at http://www.hgc.gov.uk and http://www.alrc.gov.au, respectively.

Conclusion

At the end of the October meeting, SACGHS determined that an inter-meeting task force should be organized to help the Committee identify its priority issues. At the next meeting of the Committee, in March 2004, the Committee will decide on its priority issues and develop a work plan to address these issues over the coming year. In taking these next important planning and priority setting steps, SACGHS will be well-positioned to advise the Federal government on key issues raised by the development and use of genetic technologies.