Secretary's Advisory Committee on Genetics, Health, and Society
June 26-27, 2006 Meeting
SUMMARY AND OUTCOMES

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) held its tenth meeting on June 26-27, 2006. During this meeting, the Committee received an update from the Centers for Medicare & Medicaid Services (CMS) on the status of a Notice of Proposed Rule Making (NPRM) on a genetics specialty for the Clinical Laboratory Improvement Amendments (CLIA) program; advanced the development of recommendations on pharmacogenomics; heard presentations on the environmental components of gene-environment studies as well as gene patenting and licensing practices in the US and their subsequent effect on provision of genetic tests and services for patients, and economic impact in drug and diagnostics development; reached consensus on next steps on the Committee's study of the impact of gene patents and licensing practices on access to genetic tests and services; heard updates on the activities of two federal working groups on direct-to-consumer marketing; and were briefed by two key stakeholders regarding their consensus building efforts to advance the passage of Federal genetic information non-discrimination legislation.

Oversight Session

Ms. Judy Yost, Director of CMS's Division of Laboratories and Acute Care, provided an overview of the CLIA regulations and progress made on the development of the NPRM since the 2001 publication of a Notice of Intent. The proposed regulation would add a genetic testing specialty under the CLIA regulations and identify standards that laboratories performing genetic tests would be expected to meet. Among the areas that the proposed regulation might address are informed consent, clinical validity, proficiency testing, personnel qualifications, and reporting of research results. She also described some of the hurdles that have caused the 5-year delay in the publication of the NPRM. Without specifying an expected publication date for the NPRM, Ms. Yost indicated that it would be accompanied by a 60-90 public comment period, followed by publication of a final regulation.

Outcomes. In response to Ms. Yost's presentation, SACGHS requested that SACGHS staff prepare a briefing document describing current oversight of genetic tests by both CLIA and the Food and Drug Administration (FDA), identifying gaps in current regulations, and explaining the new CLIA genetics specialty will address these gaps. The requested information will be used to assess whether any gaps in oversight of genetic tests will remain after finalization of the CLIA genetics specialty. The Committee also stressed the need to discuss this issue in the context of its work on pharmacogenomics and patents and licensing. Additionally, during a discussion of FDA's oversight of laboratory-developed genetic tests during the Pharmacogenomics Session, the Committee requested clarification of FDA's authority to regulate laboratory-developed genetic tests. The Committee plans to revisit this issue in November once the requested information is received.

Pharmacogenomics Session

The purpose of this session was to advance the Committee's development of recommendations to accompany its report to the Secretary on issues affecting the advancement of pharmacogenomics research and its integration into clinical and public health practice. The Committee's discussion centered on the review and refinement of draft recommendations discussed at SACGHS's March 2006 meeting and additional draft recommendations developed since then. The Committee agreed it would be important to focus its recommendations on issues specific to pharmacogenomics. It was most inclined toward the draft recommendations addressing the issues of health outcomes research, education of health providers and the

public, co-development of drugs and pharmacogenomics products, and drug dosing informed by pharmacogenetic test results. Dr. Emily Winn-Deen, SACGHS Pharmacogenomics Task Force Chair, reported that the Task Force would further develop these recommendations based on the Committee's input during an in-person Task Force meeting planned for September. She also reported that the pharmacogenomics literature review would be transformed into a draft report, and the revised draft recommendations would be incorporated into it accordingly. In addition to discussing the draft recommendations, the Committee heard from Dr. Janet Woodcock, FDA's Deputy Commissioner for Operations, on the Critical Path Initiative, which seeks to improve, accelerate and facilitate the medical product development process by applying new technologies, including pharmacogenomics.

Outcomes. The Committee was asked to email staff their comments on the literature review. Also, because Dr. Winn-Deen's term is ending, the Committee designated Dr. Kevin FitzGerald as the new Pharmacogenomics Task Force chair. Dr. Winn-Deen agreed to continue to serve on the task force in an ad hoc capacity.

Large Population Studies Session

At its March 2006 meeting, SACGHS reviewed the first draft of its report, *Policy Issues Associated with Undertaking a Large U.S. Population Cohort Project on Genes, Environment and Disease*. The Committee agreed then to move forward with a request for public comments on the draft report. The draft report was released for public comment on May 22, 2006. At this meeting, the Committee received an update on the status of efforts to disseminate the public comment solicitation. Efforts were made to reach many stakeholders and interested publics as well as the general public. The solicitation was disseminated through targeted mailings, listserv postings and notification of selected media organizations. The request also was announced in the Federal Register and a publication of the National Institutes of Health (NIH) that reaches a broad swath of the scientific community. The public comment period continues through July 31, 2006.

The Committee also heard two presentations on policy issues associated with assessing environmental components of gene-environment studies. In his presentation, Dr. David Schwartz, Director of the National Institute of Environmental Health Sciences, explained how assessing environmental exposures can enhance our understanding of the etiology of complex disease and discussed gaps in current knowledge that limit the ability to make definitive conclusions about exposure and disease. Dr. Schwartz described a new NIH initiative, the NIH Genes and Environment Initiative, which aims to identify measures of biologic impact in an effort to move beyond exposure-disease correlations toward an understand of mechanistic linkages between exposure and disease. He noted that the policy considerations associated with environmental exposure assessment are similar to those for genetic assessment and include the need to ensure privacy and confidentiality of data; engage the public before, during and after the study; develop clear policies for data access and sharing and dissemination of results; and gauge public perspectives in advance.

Dr. John Hewitt, Director of the University of Colorado's Institute for Behavioral Genetics, focused on social and behavioral influences on gene expression and health outcomes and the factors that can complicate their measurement. For example, the relative influences of the environment and genes may change with the onset of a behavioral condition and over its course. Also, the design and analytic strategy of a large population study must account for the possibility that the individual's selection of their environment could be determined at least in part by their genes, which would mean that genetic and environmental contributors to disease would not be independent. Dr. Hewitt also suggested that one way of addressing the complications of studying the environmental influences on health outcomes would be to

carry out studies on a subsample of identical twins. Since twins have the same genetic makeup, the environmental influences on their health will be more apparent.

Outcomes. Information from the two presentations and Committee discussion will be incorporated into the next iteration of the draft report. The Large Population Studies Task Force will be reviewing public comments and modifying the draft report accordingly. At the November meeting, a final draft report will be presented along with a summary of the public comments. With the Committee's approval, the final report will be developed and transmitted to the Secretary.

Patents and Access Session

In March 2004, when SACGHS identified the impact of gene patents and licensing practices on access to genetic tests and services as a high-priority issue, the Committee agreed to postpone its decision on whether to undertake an in-depth study of the issue until publication of the National Academy of Sciences (NAS) report, *Reaping the Benefits of Genomic and Proteomic Research; Intellectual Property Rights, Innovation, and Public Health*. At the last meeting, SACGHS reviewed the NAS report and concluded that the recommendations put forth in NAS report sufficiently addressed intellectual property concerns in the research realm; however, there was still a need to examine the impact of patents and licensing practices on clinical practice. At this meeting, the Committee heard from three experts who explained the nature of gene patents and licensing and their effects on the ability of clinical laboratories to provide genetic tests and services for patients, presented data on the effect of patents and licensing practices on access to genetic tests and services, and reviewed the role and economic impact of gene patents in drug and diagnostics development.

At the June 2006 meeting, Dr. Debra Leonard, SACGHS Patents Task Force Chair, presented some basic information about gene patents and licensing and reviewed barriers she has faced as a clinical pathologist and head of a clinical laboratory in providing genetic testing for her patients due to patents and exclusive licensing practices. Dr. Mildred Cho, Associate Director of the Stanford University Center on Bioethics, reviewed findings of a study on the impact of licensing practices on laboratories' ability to develop and perform genetic tests. She also presented results of a survey of clinical laboratory directors on the effect of licensing practices on patient access to clinical genetic tests and services. Dr. Mark McCamish, Chief Medical Officer for Perlegen Sciences, described the role of patents in supporting research and development of drugs and diagnostic products. His presentation emphasized the need for patent protections for validated genetic- and proteomic-based products.

Outcomes. After hearing from the panelists, the Committee agreed that further study of the effects of gene patents and patent licensing practices on clinical practice was warranted. The Committee expressed particular interest in determining whether there are effects in three areas: patient access to and use of genetic/genomic services; healthcare delivery and financing; and quality of genetic/genomic services. In investigating the effects, SACGHS will consider both single and complex gene diseases; legal and legislative issues; industry, healthcare provider, and patient perspectives; economic considerations; and the processes of granting and licensing of medically-relevant patents. In its assessment, the Committee will employ presentations, public discussions and deliberations with a broad array of stakeholders, and other fact-finding methods. Should its inquiry lead to the development of recommendations, the Committee indicated that every effort would be made to be balanced and attentive to negative ramifications within and outside of clinical practice.

The Committee also identified some of the key stakeholders that would have important perspectives to consider, namely patients who have experienced difficulty obtaining clinical genetic tests; patient advocacy groups who have become gene patent holders; healthcare providers (e.g., clinicians, nurses, genetic counselors) who have had difficulty procuring genetic tests and services for patients; public health

programs that may have encountered barriers to integrating genetics due to patents; economists who concentrate on patents and their impact on access to health services; academic technology transfer programs; biotechnology and pharmaceutical industry representatives; representatives of the U.S. Patent and Trademark Office; and patent officials from Europe, Japan, and Australia. The Patents Task Force will refine the work plan and organizing an in-depth session for the November meeting. Also, because Dr. Leonard's term is ending, the Committee designated Dr. Jim Evans as the new Patents Task Force chair. Dr. Leonard agreed to continue to serve on the task force in an ad hoc capacity.

Direct-to-Consumer Marketing Session

To date, the Committee has written two letters to the Secretary conveying concerns about the impact of genetic tests marketed directly to consumers without the involvement of a health provider. In response to the Committee's concerns, two interagency work groups were formed. At this meeting, the Committee was updated on the progress of these two work groups.

The first work group, composed of staff from the Federal Trade Commission (FTC), FDA, Centers for Disease Control and Prevention (CDC), and NIH, has been assessing the scientific accuracy of claims made by companies advertising genetic tests on the Internet. The group also drafted a consumer alert that advises consumers to speak with a healthcare provider when using genetic tests marketed direct to consumers. The alert also will refer consumers to validated information from CDC, FTC and FDA. The alert is currently under review at FDA and FTC; publication is expected later this summer. SACGHS commended the efforts of this work group and plans to post the consumer alert on its website once it is published.

The second work group, composed of staff from FDA, CDC, NIH, and the Health Resources and Services Administration, has been exploring mechanisms for collecting data on the public health impact of DTC marketing of genetic tests. At this meeting, Dr. Scott Bowen, who served as CDC's ex officio during the meeting, explained how CDC is currently collecting data from consumers through the HealthStyles Survey and primary care physicians through the DocStyles Survey on their knowledge about genetic tests and their sources of information. Physicians also are asked about the types of questions regarding DTC genetic tests they receive from their patients. Additionally, at the state level, three states have added questions to their Behavioral Risk Factor Surveillance Systems surveys regarding DTC genetic tests. Preliminary results from these surveys are expected in mid-2007.

Genetic Discrimination Session

In March 2004, SACGHS identified genetic discrimination as its highest priority issue. Since then, the Committee has written three letters to the Secretary urging the passage of Federal genetic nondiscrimination legislation. The Committee also documented public fears and concerns about genetic discrimination and commissioned an analysis of the adequacy of current legal protections in this area. At every meeting, SACGHS is briefed on the progress of the pending legislation in Congress. At this meeting, SACGHS heard from Mr. Michael Eastman, Director of Labor Policy at the U.S. Chamber of Commerce, and Ms. Sharon Terry, Chair of the Coalition for Genetic Fairness, on their efforts to reach consensus on the unresolved issues surrounding H.R. 1227.

Mr. Eastman explained the Chamber's historical position that any Federal law be narrowly drafted to ensure that it does not invite frivolous lawsuits or impose undue burdens on employers as they implement a new law. The Chamber's concerns with the current legislation focus on provisions relating to the appropriate scope of relief to true victims, preemption of State law, and the extent to which the bill could affect the collection by and flow of information to employers. Through several meetings, the Chamber

and the Coalition for Genetic Fairness reached consensus on a set of eight common ground principles that address many of the Chamber's concerns about the bill. In sum, they were:

- Fear of genetic discrimination has an impact on the use of genetic tests and services and participation in clinical research.
- Legislation should create a single national standard and provide the same protections, obligations, remedies, enforcement and exceptions from state-to-state thus pre-empting state law and target discriminatory conduct rather than the processing of health information.
- Any enforcement process needs to screen for unwarranted claims in order to minimize frivolous lawsuits.
- Remedies should provide injunctive relief and equitable relief. In egregious cases, additional remedies would be in order.
- The definition of "family" should cover three generations and include grandparents, parents, siblings, spouse, and children as well as adopted children.
- The Employers should not be punished for inadvertently acquiring genetic information or for acting on genetic information in order to be in compliance with other laws (e.g., Family and Medical Leave Act).
- An independent review of the legislation within six years of enactment should be carried out to assess unforeseen consequences. If an independent review is authorized, a sunset clause is not necessary.
- A communication and education campaign to inform the public about the law and their rights and responsibilities under it will be needed.

Ms. Terry explained that the legislation is currently pending in three House Committees--Education and Workforce, Energy and Commerce, and Ways and Means. She indicated that the Energy and Commerce and Ways and Means Committees have expressed agreement with the common ground principles. The Education and Workforce Committee has indicated that those principles will serve as a guide as it considers the pending bill. Ms. Terry expressed hope that this process would occur within this legislative year.