Highlights of the Fifth Meeting of the Secretary's Advisory Committee on Genetic Testing

June 5-7, 2000
Washington, DC

Overview

The fifth meeting of the Secretary's Advisory Committee on Genetic Testing (SACGT) met in public session on June 5-7, 2000, in Washington, DC to review a second round of public comments on the preliminary conclusions and recommendations on oversight of genetic tests, develop a final report to the Secretary, and consider three new areas of interest.

During the Committee's first meeting in June 1999, Dr. David Satcher, Assistant Secretary for Health and Surgeon General, requested that SACGT assess, in consultation with the public, the adequacy of oversight of genetic tests. In the fall of 1999, SACGT drafted a document, *A Public Consultation on Oversight of Genetic Tests*, and solicited public comment in December and January. Public comments were reviewed at SACGT's February meeting and preliminary conclusions and recommendations were drafted. Public comments were solicited on the preliminary conclusions and recommendations in April and May 2000. The Committee reviewed the second round of public comments and finalized the conclusions and recommendations at the June meeting.

On the second and third day of the meeting, the Committee heard presentations on three new areas of interest: genetics education of health professionals, informed consent of family members in genetic testing research studies, and human gene patenting and licensing.

Day One

The first day of the meeting was devoted entirely to a systematic review of public comments received on the preliminary conclusions and recommendations on oversight and discussion. SACGT staff made a brief presentation summarizing public comments. Committee discussion followed the major issues as outlined by staff that were raised by public comments.

In the afternoon, the Committee heard presentations from Dr. David Feigal of FDA on clinical validity and utility and FDA's review process, an update from Dr. Muin Khoury and Dr. Robert Martin of CDC on the public-private data collection efforts, and updates from SACGT liaison members Dr. Joann Boughman from FDA's Medical Devices Advisory Committee and Dr. Pat Charache from CLIAC.

Day Two

In June 1999, SACGT members identified a number of high priority issues that they thought should be a

primary focus of their work. Many members highlighted the importance of genetics education and the need to ensure both a well-trained health care work-force and a literate public. SACGT's study of the oversight issue reinforced the view that education and training have a critical role in assuring the safe and appropriate use of genetic tests.

On June 6, SACGT began to explore one side of the education issue – the education and training of health professionals. The Committee heard presentations from a number of experts in the field who provided an introduction to the issue and to some of the initiatives planned and underway in government, in the private sector, and through public-private collaborative efforts. Presentations were made by SACGT members Dr. Puryear and Dr. Burke, and invited guests Dr. Herbert Traxler of the Health Resources and Services Administration (HRSA), Dr. Ruth Kahn of HRSA, Dr. Joan Weiss of HRSA, Dr. Jeffrey Weitzel of the American Society of Clinical Oncology, Mr. Timothy Baker of CDC, and Dr. Alan Guttmacher of the National Human Genome Research Institute. A roundtable discussion followed with all of the presenters.

During the afternoon, SACGT heard from several individuals regarding informed consent of family members in genetic testing research. At SACGT's meeting in February, the Committee briefly discussed a genetic research study conducted at Virginia Commonwealth University (VCU) that had been cited by the Office for Protection from Research Risks (OPRR) for failing to comply with human subjects regulations. According to OPRR, the investigator and the VCU IRB failed to consider that family members of twins to be surveyed in the study were human subjects whose informed consent needed to be obtained or waived by the IRB. Although the issues raised by the VCU case are broader than genetics and genetic testing, the Committee thought it would be important to learn more about the decision and how it affects or applies to research involving the development and use of genetic tests.

The goal of the informed consent session was to enhance understanding of the regulatory requirements for consenting family members and how they apply in research involving the development of a genetic test. A presentation from Dr. Jeffrey Cohen of OPRR clarified the regulatory requirements regarding informed consent of family members. The Committee also heard perspectives of research subjects and family members from Mr. Richard Curtin, Ms. Sharon Terry, and Dr. Vicky Whittemore. Dr. Jane Gitschier of the American Society of Human Genetics presented perspectives of the research community. A roundtable discussion followed with all of the presenters.

Later in the afternoon, the Committee returned to the discussion of the oversight conclusions and recommendations.

Day Three

On June 7, 2000, SACGT held a session on *Human Gene Patenting and Licensing Practices and Access to Genetic Tests*. The goal of the session was to learn how patents and licenses work, how they enhance the public good through the development of products and diagnostic services, and concerns that have

emerged regarding the impact of patents and licenses on the cost, accessibility, and quality of genetic tests.

The session was divided into three panels. The first panel was entitled *The Basics of Gene Patenting, Licensing, Technology Transfer, and Commercialization*. The panel consisted of Ms. Lila Feisee from the U.S. Patent and Trademark Office; Mr. Stephen A. Bent from the law firm Foley and Lardner; Mr. Jack Turner from the technology transfer office at the Massachusetts Institute of Technology; and Mr. Charles E. Ludlam from BIO. Presentations in this panel provided a foundation of basic information about gene patents and licensing policies and practices. In the second panel, entitled *Emerging Concerns about the Impact of Gene Patenting and Licensing on Genetic Testing: Clinical, Ethical, and Patient Perspectives*, perspectives were presented from the clinical, ethical, and patient communities about the impact of gene patenting and restrictive licensing practices on access, quality, and cost of genetic tests Members of the second panel consisted of Dr. Michael Watson, a laboratory director from Washington University; Dr. Ellen Wright Clayton, a clinician from Vanderbilt University; Dr. Judith Tsipis, a consumer from the National Tay-Sachs and Allied Diseases Association; and Dr. Jon Merz, a bioethicist from the University of Pennsylvania. The third panel, entitled *The Importance of Gene Patenting and Licensing: Industry Perspectives*, consisted of Dr. Tom S. Frank from Myriad Genetic Laboratories; Dr. James H. Davis from Human Genome Sciences, Inc.; Mr. Lee Bendekgey from Incyte Genomics; and Dr. Christopher M. Palatucci from Athena Diagnostics, Inc. Presenters in this panel provided perspectives from gene-based companies about the benefits of patents and licenses. After lunch, a roundtable discussion was held with presenters from the gene patenting and licensing session and Committee members.

In the afternoon, the Committee reviewed final changes to recommendations in the oversight report and then approved the report unanimously. Among other changes made to the report were the following: Clarification of the definition of a genetic test. Many public comments urged the Committee to articulate the scope of the tests to be covered by their recommendations through the development of a precise definition of a genetic test. The following is the revised definition of a genetic test:

A genetic test is an analysis performed on human DNA, RNA, genes, and/or chromosomes to detect heritable or acquired genotypes, mutations, phenotypes, or karyotypes that are causing or are likely to cause a specific disease or condition. A genetic test also is the analysis of human proteins and certain metabolites, which predominantly used to detect heritable or acquired genotypes, mutations, or phenotypes. The purposes of these genetic tests include predicting risks of disease, screening of newborns, directing clinical management, identifying carriers, and establishing prenatal or clinical diagnoses or prognoses in individuals, families, or populations. Tests that are used primarily for other purposes, but may contribute to diagnosing a genetic disease (e.g., blood smear, certain serum chemistries), would not be covered by this definition. Tests conducted exclusively for forensic identity purposes also are excluded from the definition.

SACGT strengthened the recommendations regarding genetic counseling and education and informed consent, requiring both for genetic tests of high scrutiny.

SACGT made some modifications to their preliminary recommendation that FDA should be involved in the review of all new genetic tests. For example, they more explicitly stated that FDA should correlate the leve of review with the level of scrutiny required by a test and also made a commitment to form a working group to further develop the classification criteria and methodology described in Issue 2. They also added language to emphasize and define more fully the need for FDA to use flexible review mechanisms developed in association with other relevant agencies and organizations and to urge that modeling of the review processes should occur before implementation for a variety of tests of different scrutiny levels, including an analysis of cost and potential delay in test availability.

After members have an opportunity to review the final version,SACGT will submit the final oversight report to Dr. Satcher for transmittal to the Secretary.

In a discussion of next steps, members determined that more time and consideration was necessary before a decision could be made about where to focus their next effort. At the sixth SACGT meeting in August, the Committee will devote time to a discussion of topics presented at the June meeting and plan a course of action for future projects.