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

## Monday, October 25 and Tuesday, October 26, 1999
## Rockville, Maryland

### Overview

The Secretary's Advisory Committee on Genetic Testing (SACGT) met for the second time on October 25 and 26, 1999, in Rockville, Maryland. The meeting was open to the public. The main goals of the meeting were to discuss oversight issues and finalize plans for consulting with the public on the oversight issues.

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. Dr. McCabe, SACGT Chair, formed a Working Group on Outreach to develop a plan for gathering diverse public perspectives on the oversight issues, and charged the Working Group on Oversight with developing a comprehensive document that provided background information on genetic testing and framed the central questions that SACGT had been asked to address.

During the October meeting, members were able to discuss the oversight issues and suggest modifications to the consultation document. The draft consultation document and the draft report of the Outreach Working Group were available to members of the public in attendance. On the first day, SACGT reviewed two of the four oversight issues, heard two expert presentations and comments from seven members of the public, and reviewed the recommendations of the Working Group on Outreach. On the second day, SACGT continued its review of the oversight issues, heard a panel discussion of the roles of the States, private sector and professional organizations in oversight, and voted to approve the recommendations of the Outreach Working Group and, with modifications, the consultation documents. Members also outlined a plan for the development of a summary document and decided to develop a number of additional questions to improve the public consultation process.

### Day One

**Discussion of Oversight Issue 1: Criteria for Assessing Benefits and Risks of Genetic Tests—Possible Approaches to Oversight Issues**

Dr. Burke described the major elements of one approach to addressing the first central issue—what criteria should be used to assess the benefits and risks of a genetic test. She noted that determining the analytical validity of any test is a critical first step. Assessing the benefits and risks of a test, however, mainly relates to its performance in the clinical environment. Criteria that could be used to assess a test's benefits and risks are clinical validity and clinical utility. Because clinical validity and clinical utility may vary depending upon the health condition and the population to be tested, these criteria must be assessed on an individual basis for each health condition or disease to be tested.

In discussing the suggested criteria, members provided a range of comments and engaged in discussion on a number of themes, including the continuously evolving nature of information regarding clinical validity

and utility of a genetic test. It also was noted that the fast rate at which knowledge is generated suggests that an iterative assessment process and a process for ongoing data collection may be needed.

Members agreed that more should be added to the discussion of Issue 1 about the distinction between the individual and social impact of genetic tests (the differences in benefits and risks in the use of such tests for population screening and for diagnostic purposes) and that more should be said about the social context of testing. Also, it was suggested that the document should stress the importance of not reinforcing the idea of biological distinctions and at the same time note how important it is to work to avoid stigmatization and discrimination based on genetic information.

After this discussion Allen Buchanan, Ph.D., of the University of Arizona, summarized the main points of his preliminary draft paper, "Marketing Genetic Tests: Guidelines for Policy Making." The paper presents a taxonomy of ethical issues, a survey of the current regulatory scene, and guidelines for policy. During his presentation, Dr. Buchanan emphasized the need to avoid "genetic exceptionalism" and to consider the risks and benefits associated with non-health related genetic tests. He suggested that it was important for SACGT to delve into the ethical implications of genetic tests because these had not been fully explored by the NIH-DOE Task Force on Genetic Testing.

**Discussion of Oversight Issue 2: Discrete Risk Categories of Genetic Tests—Possible Approaches to Oversight Issue 2**

Discussion of the second oversight issue -- identifying discrete categories of genetic tests based on the criteria for assessing benefits and risks -- was also presented by Dr. Burke. If clinical validity and clinical utility were used to characterize the potential risks associated with a given test, the categorization approach might involve grouping tests according to their specific levels of risk. Using this information, tests might be organized into categories such as "high risk" and "low risk." Dr. Burke noted that such a categorization would not be simple or straightforward, because it would depend upon a combination of factors, including test characteristics, availability of safe and effective treatments, and the social consequences of a diagnosis or identification of risk status.

While members acknowledged that identifying categories may be extremely challenging, they generally agreed that it would be important for the Committee to develop specific recommendations in this area, and they expressed hope that public comments would provide helpful suggestions for doing so. Members also emphasized the need for an iterative model to accommodate an ongoing process of communicating information related to risk categories. This process would be continuously refined through the ongoing collection of data at both the individual and aggregate levels.

**Public Comment Period**

Seven members of the public presented comments to SACGT on a variety of topics. The presenters were Penelope K. Manasco, M.D., GlaxoWellcome, Inc.; Rodney Howell, M.D., FACMG, American College of Medical Genetics; Tom S. Frank, M.D., Myriad Genetic Laboratories; James R. Allen, M.D., M.P.H., Association of State and Territorial Health Officials; Judith L. Benkendorf, M.S., C.G.C., Human Genome Education Model Project; Joseph L. Graves, Ph.D., Arizona State University West; and Michael Watson, Ph.D. Dr. McCabe also provided opportunities at other points during the two day meeting for members of the public in attendance to make brief comments or pose questions.

**Report of the SACGT Working Group on Outreach**

First, Dr. Lewis presented a brief introduction to the efforts of the Working Group on Outreach in working toward soliciting broad and inclusive public comment on oversight of genetic testing. Next, Vence Bonham, J.D., Co-Investigator, Communities of Color and Genetics Policy Project, University of Michigan School of Public Health/Michigan State University/Tuskegee University, spoke on the process of engaging minority communities in a discussion of issues involving genetic testing, a process termed community engagement. Mr. Bonham stressed the importance of maintaining long-term relationships with groups from which input is sought, rather than simply implementing a one-time information seeking process. He also discussed how misuse of research information is a significant concern among many ethnocultural groups in this country, and how important it is to use a variety of formats in public consultation meetings to solicit comments from a wide range of groups. Mr. Bonham agreed to Dr. McCabe's request that he consider engaging participants of the Communities of Color and Genetics Policy Project in discussions of the SACGT oversight issues and sharing any other views and perspectives from the Project that might be relevant to the issues before the SACGT.

Members then reviewed, and ultimately approved, the five proposed outreach strategies recommended in the outreach document (solicitation in the Federal Register, a targeted mailing, a website consultation, at least one public consultation meeting, and a retrospective review and analysis of the literature). A concerted effort will be made to solicit opinions from as many diverse communities and groups as possible. SACGT agreed that a Spanish language version of the summary document would be produced and disseminated. The difficulties involved in holding more than one public consultation meeting were discussed, and members agreed that the logistics of holding a number of meetings in this short period are not trivial. The Committee decided that the most feasible plan was to hold a one-day meeting on January 27, 2000, and then consider the possibility of holding other meetings in other parts of the country at a later time. Dr. Boughman made arrangements to reserve a meeting facility for the January meeting at the University of Maryland, Baltimore Campus. It was also agreed that a steering committee, chaired by Dr. Lewis and composed of other SACGT members and ad hoc experts, would be established to help plan the January meeting.

## Day Two

The morning began with a panel on "The Role of the States and Private Sector Organizations in Oversight of Genetic Tests" that included Ann Willey, Ph.D., Director, Laboratory Policy Development, New York State Department of Health; Rod Howell, M.D., President, American College of Medical Genetics; Walter W. Noll, M.D., Member, College of American Pathologists Molecular Pathology Committee and CAP-ACMG Joint Committee on Biochemical and Molecular Resource Committee; and Dale H. Altmiller, Ph.D., Member, Area Committee on Molecular Methods and Chairholder, Subcommittee on Molecular Genetics, NCLLS.

This was followed by a discussion among the panel and SACGT members that focused on the nature of voluntary oversight for genetic testing and how genetic testing would be dealt with in the future, when the number of available tests is likely to expand significantly. In addition, members of the morning panel met during lunch with several representatives from agencies involved in the regulation/oversight of genetic testing—CDC, FDA, and HCFA—to discuss existing guidelines and guidelines in development. The representatives of the various groups decided to convene a meeting prior to the SACGT public consultation meeting in January to discuss in detail the role their respective groups may play in the future of oversight of genetic testing. This meeting will serve as a forum for representatives of private sector professional organizations (e.g, American College of Medical Genetics, College of American Pathologists,

and NCCLS), state public health agencies and federal regulatory agencies to formulate additional strategies for providing quality assurance of genetic tests.

Dr. Charache presented a summary of current CLIAC recommendations and the issues involved in these recommendations. She also informed members that CDC will be issuing a Federal Register announcement requesting public comments on a notice of intent to augment CLIA regulations with specific genetic testing provisions. She suggested that SACGT could be helpful to the process by expressing support for the overall goal of the proposal and urging its rapid clearance and publication. The Committee voted to support the goal of the notice of intent and agreed that a letter of support from SACGT should be sent to the Surgeon General.

Noting that the vote on supporting the publication of the notice of intent was the first occasion for formal vote-taking by SACGT, Dr. McCabe proposed that SACGT develop a procedural approach to voting on issues. He suggested that if unanimity was not reached on any particular issue, positions could be framed in terms of minority views. It was agreed that these could be framed as philosophical differences and concerns and that the Committee would strive to reach consensus to the greatest extent possible.

**Discussion of Oversight Issue 3: Options for Oversight of Genetic Testing—Possible Approaches to Oversight Issue 3**

Mr. Hillback began the discussion of Issue 3 -- options for oversight of genetic tests -- by noting the importance of thinking about the concept of oversight in the broadest sense possible not as only a matter of laboratory quality assurance. He went on to describe a "flow versus batch" approach to oversight of tests that might be effective in taking account of the iterative nature of knowledge generation. He also described how a consortium of government, industry, and private sector partners might be an innovative approach to oversight and data collection. Ms. Beardsley outlined the several options SACGT had identified for oversight of analytical validity and clinical application of genetic tests. These included: strengthening CLIA regulations, which could require any laboratory that provides tests upon which clinical decisions are based to demonstrate the test's validity to outside reviewers; expanding FDA medical device regulations to include all or some laboratory-based genetic tests; forming an interagency government review board to review tests before their clinical and public health use; forming government and private sector partnerships for the purpose of developing standards and guidelines, including guidelines for the review of research protocols; and/or developing government and private sector partnerships to review tests prior to their use.

During discussion of Issue 3, a suggestion was made that rather than presenting options for oversight, the consultation document should discuss principles of oversight and provide examples. Members agreed that there is a need to be more explicit about the elements that go into each option and that the tables displaying the options should not be included in the document because they appear to limit and over-simplify the options. It was also suggested that the document should explain what should be included in regulation/oversight, rather than identifying which agency/entity should provide the oversight.

**Discussion of Oversight Issue 4: Appropriate levels of Oversight for Categories and Data Collection, Evaluation, and Dissemination—Possible Approaches to Oversight Issue 4**

With regard to SACGT's approach to Issue 4a -- appropriate level of oversight by category -- Ms. Beardsley pointed out that it would be premature for SACGT to put forward any specific approaches until after public input had been received. With regard to 4b -- a process for data collection, analysis, and dissemination,

Ms. Beardsley discussed four possible approaches to data collection and three possible approaches to dissemination. She pointed out that the SACGT focused on the dissemination of scientific data but that, in light of discussions during the previous day's meeting, more emphasis may be needed on information disseminated through marketing of tests. During discussion of Issue 4, members agreed to rearrange the order of questions 4 and 3 and a suggestion was made that the Committee step back from the examples and instead focus on the issues and lay out the discussion that led to their development.

Dr. Khoury reported on efforts underway by a DHHS interagency group to explore a public/private partnership for data collection in genetic tests. He emphasized the importance of a standardized data format and noted that a model for data collection would be useful if it is perceived as an independent entity.

**General Discussion of the Oversight Documents**

In response to comments by several members that the current long consultation document and executive summary may not be understandable to the average individual, Dr. Boughman suggested that public review and comment on the issues might be facilitated if the major issues and some additional questions were put forward in a separate stand-alone document that would supplement the current long version and the summary. The Committee agreed that identifying additional questions would be important. Dr. McCabe said that he, along with Ms. Beardsley and Dr. Lewis, would work with staff to articulate additional questions and revamp the consultation documents along the lines agreed to by SACGT at the meeting. Dr. McCabe said he hoped to be able to hold a conference call by November 9 to review and discuss the documents and that the group as a whole would review the revised documents before the consultation process is launched.

**Future Directions**

Before adjourning, SACGT members discussed next issues that the Committee may take up after the current assignment is completed. The issue of the training of health care providers in genetics and the promotion of public understanding of genetics is a high priority for SACGT, and members noted that training is related in important ways to the oversight issue.

Members also expressed concern about whether gene patenting and restrictive licensing practices are having an adverse impact on availability, cost, and quality assurance of genetic tests. During the meeting, the American College of Medical Genetics reported on survey results indicating that 25 percent of its members had discontinued offering certain genetic tests because of the patent/licensing complexities.

The issue of confidentiality of genetic information is also a serious issue, especially with regard to social concerns of discrimination and stigmatization. SACGT members were aware that a proposed HIPAA privacy regulation would shortly be released by DHHS for public comment. Members indicated an interest in reviewing the proposed regulation. Members also expressed continuing interest in being briefed on the Administration's efforts to address genetic discrimination in health insurance and employment, a briefing that had been scheduled during this meeting but was canceled.