Highlights of the Tenth Meeting of the
Secretary's Advisory Committee on Genetic Testing
August 17, 2001
Bethesda, MD

The tenth meeting of the Secretary's Advisory Committee on Genetic Testing (SACGT) was held in public session on August 17, 2001, in Bethesda, Maryland. The Committee was briefed on the outcomes of a one-day outreach meeting convened the previous day by the Data Work Group and heard a report from the Education Work Group on genetics education of health professionals and preliminary results of a genetics education survey. SACGT was also heard brief reports on relevant activities of the National Human Research Protections Advisory Committee (NHRPAC), the Office of Human Research Protections (OHRP), and the Clinical Laboratory Improvements Advisory Committee (CLIAC). In addition, SACGT heard progress reports from its three other work groups on informed consent and IRBs, access, and rare disease testing and was updated on the status of Federal genetic discrimination legislation.

On August 16, 2001, the Data Work Group convened an outreach meeting to discuss and gather input on three areas: a template developed by FDA for the purposes of pre-market review; efforts to enhance post-market data collection efforts; and provider summaries for genetic tests. Dr. Wylie Burke, chair of the Data Work Group, briefed SACGT on the outcomes of the one-day meeting. Regarding the pre-market review template, the meeting participants agreed that the template contains the appropriate elements needed to review a genetic test's validity and utility. However, several questions were raised and concerns were expressed regarding how FDA plans to review the submitted data. The Work Group suggested that additional examples demonstrating the applicability of the template would be helpful, particularly examples of non-molecular genetic tests. Other suggestions included adding 'mode of inheritance' to the list of elements in the template and providing more guidance on the level of information required for each element, particularly for the clinical validity section.

There was general agreement that more post-market data needs to be collected, however, there are many challenges to achieving this goal. Some of the challenges include the need for multi-site research projects and longitudinal follow-up studies; the need to link laboratory results with clinical data, which is particularly challenging with regard to issues of privacy and confidentiality; and the need for broad access to data for secondary analysis and dissemination. The group recognized that the type of efforts needed would vary depending on the stage of test development.

Regarding the provider summaries for genetic tests developed by the Data Work Group, participants stated that the concept of provider summaries was a good idea but that the proposed format had a number of shortcomings. Participants were unclear as to who would develop the summaries and how they would be updated, thus calling into question the sustainability of the initiative. Furthermore, participants stated that if used as stand-alone documents, the summaries have the potential to oversimplify genetic test information and to dissociate test information from the clinical context. It was suggested that such summaries would need to be linked to more detailed information sources in order to be useful to multiple audiences. Overall, ensuring the

quality of the information will require resources for the development, dissemination, and maintenance of current data.

A general recommendation of the outreach meeting was that SACGT could serve as a champion in several different areas to advance these initiatives. For example, SACGT could promote new funding mechanisms for the different stages of post-market data collection, encourage multi-disciplinary collaborations and public-private partnerships, and emphasize the need for public access to new information. In order to help SACGT assess the current status of research on the clinical validity and clinical utility of genetic tests and determine where additional support may be needed, SACGT requested that SACGT *Ex Officio* liaisons define their agencies' roles in supporting primary and secondary data collection efforts and to identify where additional efforts may be needed. The Committee also requested that FDA be prepared to provide more details about how the agency plans to review the information provided in the template, the outcomes of the review process, and how professional organizations may be involved in the review process.

SACGT heard a number of brief presentations on various activities of interest to the Committee. Dr. Patricia Charache, liaison to the CLIAC, briefly updated the Committee on current activities of CLIAC regarding oversight of waived tests. Dr. Joe Boone, Assistant Director of Science of the Division of Laboratory Systems at CDC, updated SACGT on the status of the Notice of Proposed Rule-Making to augment CLIA regulations for genetic testing. Dr. Susan Zullo, senior advisor to the Director of OHRP, briefed the Committee on OHRP activities related to genetics and Ms. Kate-Louise Gottfried, Executive Director of NHRPAC, discussed the work of NHRPAC relating to genetics. Dr. Kathy Hudson, Director of the Office of Policy and Public Affairs at the National Human Genome Research Institute, updated the Committee on the status of Congressional efforts to pass legislation prohibiting genetic discrimination.

The chairs of the remaining work groups reported to the Committee on their groups' progress. Ms. Mary Davidson and Dr. Michael Watson, Co-chairs of the Rare Diseases Work Group briefly outlined the framework of a white paper to be completed in early 2002. The paper will focus on several areas including current standards and definitions of rare diseases, marketing and development incentives for rare disease testing, access issues related to rare disease testing (in conjunction with the Access Work Group), and technical assistance for rare disease testing laboratories.

Dr. Judith Lewis, Chair of the Access Work Group, discussed the group's efforts to address reimbursement and health disparities issues as they relate to genetic testing. Dr. Lewis reported on the progress of two documents, one on billing and reimbursement for patient education and counseling services for genetic testing and the second on guiding principles for health care payers regarding coverage of and reimbursement for genetic testing services. Dr. Lewis also stated that the group, in conjunction with the Data Work Group, would be planning presentations on how population data are collected, organized, and reported to provide clarification on the health disparities issues as it relates to genetic testing.

Dr. Barbara Koenig, Co-Chair of the Informed Consent/IRB Work Group, reported on the status of her group's current projects. After reviewing the group's mandate and six specific charges, Dr. Koenig updated the Committee on the progress of two current projects--the development of

an information brochure on genetic testing for the general public and the formulation of principles of informed consent in clinical and public health practice. Thus far, the group has discussed the benefits, limitations, and challenges of informed consent for genetic tests and agreed that a pragmatic approach to the development of principles or standards is critical. At a work group meeting on May 4, the group drafted a basic framework of the dimensions of informed consent (information disclosure, information comprehension, directiveness/personal choice, and documentation) across four levels (minimal to high). Meetings and conference calls over the summer of a small subgroup led to refinements in the framework and the development of an approach to categorizing tests according to level of consent needed. In addition to finalizing the information brochure at a meeting September 12, the work group will continue working on the framework and classification concept with the goal of presenting recommendations in November.

The afternoon was devoted to a session on genetics education of health professionals. Dr. Joann Boughman, Chair of the Education Work Group, introduced the session and updated the Committee on the activities of the work group. At the May meeting, SACGT had agreed that an Education Summit should be convened to gather input from educators, health professionals, consumers, and other interested parties to help identify gaps in genetics education of health professionals and determine what role, if any, SACGT could play in addressing these gaps. The Committee requested that background data be gathered in preparation for the Education Summit and presented to the Committee at this meeting. Dr. Susanne Haga, SACGT staff, presented an overview of genetics education of heath professionals, summarizing past recommendations of other Federal committees, curricula of medical and nursing programs, workforce issues, and genetics content on national board examinations. Presentations followed on the preliminary results of a survey administered by the National Coalition for Health Professional Education in Genetics (NCHPEG). Dr. Priscilla Short, Program Director of Science, Technology, and Research at the American Medical Association, presented the NCHPEG survey results for the healthcare and non-profit sector and Mr. Joe McInerney, Executive Director of NCHPEG, presented the survey results for the private sector.

Following the presentations, Dr. Boughman led a discussion to define the focus, content, and structure of the proposed Education Summit. It was determined that the summit should be inclusive of consumers and all health professions and disciplines in order to gain a complete picture of current activities in genetics education, identify gaps, and develop recommendations to address the gaps. It was suggested that the Education Summit be structured similarly to the public consultation meeting convened by SACGT in January 2000 to facilitate the involvement and participation of key players in the education arena. It was decided that the summit should be held the day before SACGT's November meeting on November 14, 2001, and that the full Committee should be in attendance.

At the November meeting, the Committee will also hear a progress report from the agencies regarding its request for information on resources devoted to projects focused on understanding the clinical validity and clinical utility of genetic tests, a progress report from FDA on the steps towards implementation of oversight for genetic tests, a presentation and discussion on the draft report of the Informed Consent/IRB Work Group, and a session convened by the Rare Diseases

Work Group to gather information on various issues related to the development, translation, and maintenance of tests for rare diseases.