Secretary's Advisory Committee on Genetics, Health, and Society
Summary of Fourth Meeting
June 14-15, 2004
Bethesda, Maryland

Committee Members Present:

Edward McCabe, M.D., Ph.D., Chair
Cynthia Berry, J.D.
Barbara Willis Harrison, M.S.
C. Christopher Hook, M.D.
Debra G.B. Leonard, M.D., Ph.D.
Brad Margus
Agnes Masny, R.N., M.P.H., M.S.N.
Joan Reede, M.D., M.P.H., M.S.
Huntington Willard, Ph.D.
Emily Winn-Deen, Ph.D.
Kimberly Zellmer, J.D.

Ex Officios/Alternates Present:

Hratch Semerjian, M.Sc., Ph.D., DOC
Martha Turner, Ph.D., DOD
Paul Miller, J.D., EEOC
Stuart Ishimaru, J.D.
Peter Gray, J.D.
Matthew Daynard, J.D., FTC
Martin Dannenfelser, HHS/ACF
Kaytura Felix-Aaron, M.D., HHS/AHRQ
Muin Khoury, M.D., Ph.D., HHS/CDC
Timothy Baker
Steve Phurrough, M.D., HHS/CMS
Judith Yost, M.A., M.T.
Suzanne Feetham, Ph.D., R.N., HHS/HRSA
Michael Carome, M.D., HHS/OHRP
Joseph Hackett, Ph.D., HHS/FDA
Elizabeth Mansfield, Ph.D.
Alan Guttmacher, M.D., HHS/NIH
Tim Leshan, M.P.A.
Howard Zucker, M.D., HHS/OASH
Robinsue Frohboese, J.D., Ph.D., HHS/OCR
Amy Turner, J.D., DOL
Sherrie Hans, Ph.D., HHS/DVA

Sarah Carr, Executive Secretary

Monday, June 14, 2004

Welcome and Opening Remarks
Edward McCabe, M.D., Ph.D.
SACGHS Chair

Dr. McCabe welcomed members and the public to the fourth meeting of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS). He noted that the public was made aware of this meeting through notices in the *Federal Register* and announcements on the SACGHS web site and listserv.

He announced changes in the membership of SACGHS. Dr. Eric Lander resigned from the Committee because of extensive commitments as Director of the new Broad Institute of MIT and Harvard. The Secretary will be appointing a new Member to take his place. Dr. Howard Zucker, Deputy Assistant Secretary for Health, has been appointed as a new *Ex Officio* member representing the Assistant Secretary for Health in the Department of Health and Human Services (HHS). Dr. Steven Gutman, Director of the Office for In Vitro Diagnostics Evaluation and Safety, is the new *Ex Officio* member for the Food and Drug Administration (FDA), replacing Dr. David Feigal. Dr. Hratch Semerjian, Acting

Director for the National Institute of Standards and Technology, is representing the Department of Commerce for Dr. Arden L. Bement, Jr.

Dr. McCabe reviewed the status of SACGHS efforts, remarking that since the March meeting some short-term actions have been taken, including the sending of a second letter to the Secretary in support of federal genetic nondiscrimination legislation.

At its last meeting, the Committee had decided that education and training of health professionals in genetics was a high-priority issue and that after additional fact-finding on genetics education efforts, it would be equipped to consider a resolution regarding these efforts during this meeting. An Education Task Force chaired by Dr. Joan Reede has been gathering data and drafting a resolution for the Committee's consideration. The Committee also decided to prepare a report describing SACGHS's mandate and priorities and providing background information on the policy considerations involved in each issue. A draft report prepared by staff, with the assistance of Dr. Emily Winn-Deen, was made available for the Committee's consideration. Additionally, the Committee identified high-priority issues that require more in-depth study. Coverage and reimbursement of genetic technologies and services was identified as the first topic on which the Committee would focus its efforts. Staff, with the assistance of Cynthia Berry, carried out additional data and information gathering and prepared a draft report.

The Committee also decided to take a short-term step by drafting a resolution expressing concern about the proliferation of direct-to-consumer (DTC) marketing of genetic tests. A draft resolution was prepared by staff with the assistance of the DTC Task Force chaired by Dr. Christopher Hook.

Two other issues also were identified as warranting in-depth study: large population studies and pharmacogenomics. The Committee's in-depth work on these two issues will begin at future meetings.

Dr. McCabe reviewed the agenda for the current meeting and noted that the meeting goals included finalizing the resolutions on education and DTC marketing and the report on the Committee's priority-setting process. He noted that given the length and complexity of the coverage and reimbursement report and the work that needs to be done to develop consensus recommendations, it is likely the Committee will need to continue to refine the report and gather additional public comment after this meeting.

Ms. Sarah Carr reviewed the rules of conduct for government employees related to conflicts of interest.

Update on Genetic Nondiscrimination Legislation and the Activities of the Coalition on Genetic Fairness
Joann Boughman, Ph.D.
Executive Vice President
American Society of Human Genetics (ASHG)

Dr. McCabe introduced Dr. Boughman and remarked that protection against genetic discrimination has been a top priority for both the Secretary's Advisory Committee on Genetic Testing (SACGT) and SACGHS. He said that the Committee was encouraged when S.1053 was unanimously passed in the Senate last October, 95 to 0, and that following the March SACGHS meeting, the Committee wrote a second letter to the Secretary urging that continued pressure be applied to facilitate passage of the Genetic Information Nondiscrimination Act in the House.

Dr. Boughman began by emphasizing that there has been no real movement of this legislation, but that because the session is not over, efforts could still be made to work with those on the Hill and to

encourage constituents to contact and discuss this issue with their representatives. She discussed the status of the bills that were introduced to the House, noting that the best hope was to get S.1053 to committee and then to the floor, or directly to the floor. Otherwise, the goal would be to get H.R.1910 through some committees so that the wording and concepts in S.1053 could be used in a compromise. She noted that H.R.3636 is a shell bill that would be complicated to negotiate through the process.

She described several meetings with advocates and the Chamber of Commerce and noted an editorial published in the *Washington Post* recommending passage of the legislation. In addition, the American Society of Human Genetics (ASHG) and the Coalition for Genetic Fairness have continued to meet with members, staff, the leadership of various agencies and key congressional committees to try to initiate movement. She described other efforts made to keep the Act on the agenda and stated her hope that at today's meeting participants would state a renewed commitment to moving the legislation forward.

Dr. Boughman reminded Members that the White House has expressed support for S.1053 and that any comments made to the Secretary should be set within this context. She also remarked that a focus in Congress on other high priority issues has been an important factor in the legislation's lack of progress, and it is not clear whether there are additional specific actions that would be effective at this point. Mr. Leshan of the National Institutes of Health (NIH) said that he believes it is important that the legislation is passed and that the agency will continue to provide assistance in this area.

Dr. McCabe noted that government employees and special government employees are precluded from lobbying and that Dr. Boughman's update represents the views of ASHG and the Coalition for Genetic Fairness, while the Committee's role is to discuss how it can provide advice to HHS and the Secretary. He said that at the March meeting there was discussion regarding the possibility that further action may be required in the event that legislation is not passed during this session. Such action could take the form of organizing briefings from individuals who have faced genetic discrimination, who paid out-of-pocket to keep their genetic information out of their medical records, or who chose to forego treatment because of fear of genetic discrimination. Dr. Boughman and Dr. McCabe outlined the progress that has been made in obtaining patient stories.

The Committee agreed that, should the House bill not pass, it would be worthwhile to have at the October meeting remarks by individuals who have been the subject of genetic discrimination, and it was agreed to begin planning for these briefings at the end of the current meeting. A task force was formed for this purpose that would include ASHG and the Coalition for Genetic Fairness, which have already initiated efforts in this area. The Task Force included: Ms. Barbara Harrison, Dr. Debra Leonard, Ms. Agnes Masny, and Dr. Winn-Deen. The Task Force would also call upon ad hoc members.

Session on Genetics Educations and Training

Presentation on Information Gathered on Efforts in Genetics Education and Training
Joan Y. Reede, M.D., M.P.H., M.S.
Chair, Education Task Force

Dr. Reede reviewed the Education Task Force's information-gathering efforts over the last three months. During the discussions of genetic education and training at the March meeting, the Committee decided to establish a task force to collect information on the genetics education and training activities of various health professional organizations, to organize and facilitate a roundtable discussion to be held during the June meeting, and to draft a resolution to the Secretary on genetics education and training outlining key recommendations in this area. Dr. Reede said that the Committee's request to hear formally from

professional organizations in the private sector on their activities in genetics education and training was meant to serve as a follow-up to a previous survey of federal agencies on their activities. The recent survey was to be used to inform the resolution to the Secretary and to ensure that any recommendations made to the federal government would be complementary to activities already ongoing in the private sector.

The Task Force solicited information from organizations representing multiple specialties and disciplines in three categories: genetic-specific organizations, organizations involved in the education of health professionals, and health profession organizations, with the latter category including groups that represent generalists, specialists, and specific constituencies. From a total of 26 organizations sent surveys, fifteen responded to questions regarding current initiatives and activities that relate to genetics education and training.

Genetic-specific organizations: For this group, the Task Force wanted to increase its understanding of current initiatives and activities that relate to diversity in the genetics workforce, that enhance genetics and genomics curricula, and that promote the incorporation of genetics and genomics content into licensure and certification. It was found that genetic-specific organizations are involved in many activities that target the education pipeline, as well as the recruitment of individuals from diverse backgrounds into genetic counseling. These efforts also target minority health professionals and at the organizational level encourage diversity through, for example, the organization of diversity standing subcommittees. Genetic-specific groups appear to be leaders in the development of genetics curricula and are actively engaged in dissemination and outreach efforts to other health professionals. A major effort on the part of these organizations has been the development of what are known as the core competencies in genetics. Those surveyed remarked that educational materials should conform to the trend of electronic medicine. Barriers include a lack of appropriately prepared faculty for genetics training and education and a need for more courses to train faculty to teach genetics content.

These groups have the potential to change behavior and increase the integration of genetics and genomics knowledge through licensure, certification, and accreditation. Although many of the respondents did not report a great deal of activity in these areas, some activities that were reported included providing credentialing programs in genetics, using core competencies to help inform discussions or programs related to licensure and certification, and conducting biannual genetics review courses. Different types of specific policy solutions are needed in this area, partly because of the variety of organizations involved, including state governments, specialty professional societies, the federal government (through the Clinical Laboratory Improvement Amendments [CLIA]), and private organizations (the Joint Commission on Accreditation of Healthcare Organizations [JCAHO]). The overall recommendation from this group of respondents was that the HHS Secretary should actively support a wide variety of endeavors based in or funded by any of the HHS-based agencies and should seek partnerships with other relevant federal agencies.

Organizations involved in the education of health professionals: Survey questions for this group were designed to gauge where the issue of genetics/genomics stands in health education organizations that have a more general focus. The task force was trying to focus on the need for the integration of genetics and genomics into curricula as well as identification of barriers to this integration and current ongoing activities and initiatives. In their responses to the survey, these organizations said that there is a need to acknowledge that genetics education is for both generalists and specialists. Many cited a need for improved access to knowledge that will help providers evaluate the validity, efficacy, and safety of products, tying in with the Committee's interest in and focus on DTC marketing and oversight. Genetics needs to be redefined and recast as an inherent and overarching part of health communication between all

health professionals and the public about how genetics affects health requires improvement, and tools for lifelong learning are needed.

Barriers to teaching genetics and genomics include the lack of models using clinically relevant examples and insufficient numbers of trained faculty broadly competent in genetics and genomics. It is expected that there will be a lack of uniformity in the rate of integration of genetics into the various medical specialties, which in turn will make it difficult to determine who should be learning what and when. Other barriers include difficulty in motivating students to learn about genetics based on the promise of the subject's future importance, an overcrowded curriculum that is already struggling with issues of basic science and clinical practice, and the fact that genetics is still considered by many to be an esoteric field.

This group of organizations emphasized that schools and professional organizations must assume the primary role in providing leadership in preparing the next generation of health professionals in genetics. Current activities of health professional educational organizations include integrating the core competencies that have been identified by the National Coalition for Health Professional Education in Genetics (NCHPEG) into their entry-level competencies for their specific disciplines. Continuing education is needed to train the trainer, or to help clinicians determine when to refer, how to obtain information about genetics research studies, and how to discuss research options with patients. More funding is needed to support training and education in genetic technologies and to facilitate the incorporation of new knowledge and skills. One organization is sponsoring relevant legislation that would help support curricular development in the area of genetics and genomics.

Health profession organizations: The Committee also gathered information from umbrella organizations that focused on specific health professional disciplines in order to provide useful information on the relative importance of genetics to organizations grappling with many other equally important issues. Such information also may be an indicator of how genetics and genomics are perceived within the health care system generally. These groups were asked to characterize the need to integrate genetics and genomics and about what types of activities or initiatives are currently ongoing. They also were asked about partnerships, interdisciplinary efforts, educational products, and outcomes. They were asked to note specific steps they were taking to improve diversity and to identify recommendations they would like the Committee to make.

This group responded that all health professionals need a strong knowledge base in genetics and genetics testing and that identifying clinically relevant examples of genetics and genomics would help address the need to understand how genetics interfaces with practice. Professionals need help in becoming familiar with up-to-date advances in genetics. In terms of current activities, the health professional organizations are undertaking a broad array of activities related to genetics education and training. Several involve partnering with others in such efforts, and most report that their activities are interdisciplinary in nature. Most of the organizations measured their outcomes and evaluation based on continued or increasing interest in issues related to genetics and genomics. Current activities related to increasing diversity include community outreach, career development, education, research, advocacy, and the development of organizational position statements.

Concerns included a need to examine the science underlying issues of race in medicine and to integrate this understanding into genetics education and training. Issues of race in genetics necessitate special consideration and treatment in the educational setting, and awareness of these issues is an important part of a health practitioner's cultural competency.

For many physicians, genetics does not have immediate, daily, and clinical applicability. Such comments underscore the need to understand how genetics interfaces with practice and the barriers caused by the difficulty in finding case examples or models in genetics and genomics. Genetics education must be represented throughout the entire continuum of medical education, and tools are needed that facilitate lifelong learning. Educational programs must have a focus on pediatrics and genetics and recognize that physicians prefer interactive learning with case studies to didactic approaches. The effect of the nursing shortage on patient education and informed consent also was raised.

This group recommended that ongoing, continuing education should be the responsibility of the licensing agencies and professional organizations and that the development of profession-specific materials should be left to the health professions. Providing support for genetic education programs would be an appropriate role for the government. Funding is needed for new programs that feature educational practices known to change physician behavior. Efforts in genetics education must extend to related areas of molecular medicine, and benchmarks should be instituted to inform and assess the bidirectional impact of translational research—that is, from bench to bedside and bedside to bench. Education and training should address population-based genetic variation and its utility in the emerging era of individualized medicine.

Draft Resolution. The goals of the Education Task Force were to draft a resolution on the issue of education and training based on consensus, to convey this consensus to the Secretary, and to make recommendations or outline possible steps that would address the concerns raised during the discussion and fact finding activities. The Task Force's conclusions were that genomics can improve health, that adequate education and training in genetics and genomics are essential to integrating genetics into the health care system, and that access is contingent on effective integration. Additionally, the health education organizations identified the need for inventoried, widely relevant clinical applications; educational models that use such applications; and a broadened focus from genetics to genomics. Also needed are appropriately trained faculty and training programs that address genetics/genomics and public policy.

Dr. Reede described the issues that were the focus of the draft resolution. First, genetics is important and special because it is relevant to all areas of medicine and health care, and thus it should be integrated throughout all stages of learning, in all settings, and throughout all disciplines. Second, there is a need to support programs that enhance diversity in and the cultural competency of those in the health professions. Third, there is a need to engage other stakeholders in the process of cataloging genomics applications to clinical medicine and public health. This third recommendation addresses the concern about the lack of clinically relevant genomics and genetics applications. Fourth, there is a need to support programs that "train the trainers" in genomics and genetics education, which would address the need for more professionals trained to teach genetics. Fifth, there is a need to promote communication among faculty to enhance the use of genomics educational models. Sixth, there is a need to encourage the incorporation of genetics and genomics into the certification and licensure processes.

Roundtable with Selected Organizations on Their Efforts in Genetics Education and Training
Joan Y. Reede, M.D., M.P.H., M.S.
Chair, Education Task Force

During this roundtable, representatives of eight organizations explored in greater depth the efforts of key professional societies and educational organizations to enhance the knowledge of health professionals in genetics and genetic technologies and identified what steps are needed to advance these efforts. Participants discussed their key issues, concerns, and problems with respect to genetics education and

training from the perspective of their organizations. They also discussed gaps in the current education of genetics health professionals and made suggestions for addressing the lack of trained faculty in genetics.

Participants in the roundtable included:

Dawn Allain, M.S., CGC
President
National Society of Genetic Counselors

Joann Boughman, Ph.D.
Executive Vice President
American Society of Human Genetics

Toby Citrin, J.D.
Association of Schools of Public Health
Office of Community-Based Public Health
University of Michigan
School of Public Health

Georgia Dunston, Ph.D.
National Medical Association
Professor
Department of Microbiology
Howard University College of Medicine

Felissa Lashley, Ph.D.
American Association of Colleges of Nursing
Dean
College of Nursing
Rutgers, The State University of NJ

Judith Lewis, Ph.D., R.N.
International Society of Nurses in Genetics
Associate Professor
School of Nursing
VA Commonwealth University

Joseph McInerney, M.S.
Executive Director
NCHPEG

Michael Whitcomb, M.D.
Senior Vice President
Medical Education
AAMC

Mr. Citrin, representing Association of Schools of Public Health (ASPH), discussed the findings of a survey of the web sites of all 35 schools of public health to determine the extent to which genomics has found its way into programs or curricula. The survey found that 10 of these schools have genetics programs, and 12 offer courses, although few are identifying ethical, legal, or social issues in their curricula, and the large majority of schools of public health have not incorporated genetics or genomics information into the content of public health education. Furthermore, most of the schools that say they are teaching genetics are offering specific, discrete courses that are typically electives. Therefore in general, students in schools of public health today are receiving little or no formal genetics education. In his view, the major barriers to incorporating genetics/genomics into curricula are a lack of faculty able to incorporate genomics into the broader view of causation of health and disease, an already full curriculum, and a lack of a requirement to incorporate genetics/genomics into the curriculum. In addition, most schools do not require ethics training for a master's degree in public health.

Mr. Citrin noted that ASPH has had a role in advancing knowledge of genomics in public health and furthering the interaction between schools of public health and the world of public health practice. Genomics—which is in an early stage of expanding in public health practice—presents a wonderful model for schools of public health for connecting public health to practice. He noted the role that the Centers for Disease Control and Prevention (CDC) has had in supporting schools of public health in both teaching and practice.

Dr. Lashley described the efforts of the American Association of Colleges of Nursing to ensure that genetics is recognized as an essential part of the nursing curriculum. This has included an attempt to incorporate knowledge of genetics in its 1998 Essentials of Baccalaureate Education document. Barriers

include the intensity and density of the curriculum and the number of other topics competing for space. She also said that much of the genetics material covered in nursing programs is focused on providing information in specialized, disease-related areas rather than in providing a broad look at the influence of genetic and genomics across the curriculum. Also, there is a lack of qualified faculty to teach genetics material.

Dr. Lewis of the International Society of Nurses in Genetics (ISONG) said that ISONG works with its nurse members to ensure they have an appropriate knowledge base in genetics and with nonmembers to ensure a broad knowledge of basic genetics. The presence of many competing priorities in the curriculum was again cited as a barrier. Certification and credentialing bodies may be part of the solution by providing mandates, which has been found to be an effective strategy in other areas. It is important to convince curriculum committees that this material is important. Another critical task is determining the best model for learning. Several programs are available that might serve as models in terms of nursing professionals and genetics. A mixed model is useful for accommodating different learning styles.

Dr. Whitcomb of the Association of American Medical Colleges suggested that the barriers in medical schools were not necessarily related to an overcrowded curriculum, but rather to two fundamental problems that transcend all medical education. The first is the need to integrate content across the curriculum in a way that makes it relevant, important, and learnable for students and residents. He remarked that there has been a shift in medical education reform over the past decade toward a more integrated curriculum, as opposed to the discipline-specific, departmentally-controlled courses that have been typical of most medical school curricula. The second barrier involves the need to provide students and residents with the opportunity to be exposed to patients in order to learn how to apply their knowledge in an actual patient care environment.

Mr. McInerney said that the barriers mentioned by the other participants are also applicable to NCHPEG's efforts. The vast majority of NCHPEG's more than 145 member organizations in the United States and abroad represent nonphysician allied health groups, commercial organizations, and consumer groups. In addressing the needs of NCHPEG's diverse membership, the greatest gap in changing current practice is the lack of evidence and educational materials that show that genetics makes a difference in patient outcomes. In addition, although the core competencies have been effective and many organizations are beginning to incorporate them into the development of their own curricula, more guidance is needed about what to teach. He noted that NCHPEG has on its web site a set of core principles in genetics that most health processionals should be able to understand. Other serious gaps include the issues of diversity and cultural competence. Also, there may be constituencies within the health professions that NCHPEG does not currently reach and that should be involved in the development of its materials.

Mr. McInerney emphasized that from his experience with the NCHPEG membership, it is apparent that all health professionals, not just physicians, prefer interactive learning with case studies. He discussed a program NCHPEG has completed in conjunction with colleagues from the dental community that was developed around case studies (with some core basic genetics) to try to get dentists and dental hygienists to think differently about genetics and patient care.

Dr. Dunston remarked that the National Medical Association (NMA) is the largest and oldest national organization representing the interests of African Americans, with more than 25,000 physicians and their patients as members. NMA is dedicated to keeping its members abreast of the many rapidly occurring advances across the various medical specialties, including genetics and genomics, and is sensitive to

genetics as a subject that can destabilize a community for numerous reasons. Dr. Dunston emphasized the importance of learning how to engage the community in such a way that the prospect of benefitting from genetics is conveyed. The education of the community, as well as medical professionals, is of paramount importance, as is demonstrated in continuing medical education (CME) as well as community outreach, which often is complemented by a focus on faith-based organizations. She noted that the inclusion of African people brings with it the challenge of incorporating and understanding variation and diversity in biology into medical practice as strengths rather than as bases of disease. The question, she said, becomes one of determining how to begin to change the concept of medicine when the gene is not the pathological agent but rathera means by which we can analyze and understand disease.

Dr. Boughman discussed the resources of ASHG, the umbrella organization of genetics professionals, that could be used to bring to the table individuals who have a knowledge base in genetics and who can help answer questions. She said that we may be close to the point of "genetics readiness", as more people are beginning to identify genetics as interesting and important. The tasks are now for geneticists to become ready to teach, for organizations and communities to be more specific about what they need, and finally for the appropriate models and materials to be developed and disseminated.

Ms. Allain said that over the last year the National Society of Genetic Counselors (NSGC) joined the Health Profession Network, an organization of allied health provider professional organizations, as well as the Association of Schools of Allied Health Providers in an effort to be a resource for the integration of genetics curricula into allied health provider education and for the development of ongoing education of these professionals. To move forward, organizations need evidence-based educational activities and the development of case studies that are based around specific medical disciplines.

Roundtable discussion points:

- Several participants noted that some of the best ways to get the word out to practitioners about the availability of educational materials are through licensure, certification, and accreditation, as well as through specialty organizations and their programs and conference updates. It was suggested that licensure and certification is one way to drive curricula and that professional societies are an extremely important vehicle for reaching out to health professionals.

- Dr. Boughman remarked that there has been a formal shift to require CME as a condition of maintaining certification. This may provide a window of opportunity for the inclusion of genetics information. Dr. Whitcomb remarked that there is no national mechanism to mandate continuing education and that licensure is granted by the states, not all of which have continuing education requirements. Of the states that do have requirements for re-licensure, few mandate the specific content to be covered.

- Dr. Lewis commented that patients are the most valuable source of education and that greater partnership between the clinician and the patient (and the patient's family) is needed. Dr. Boughman also emphasized the importance of consumers' role in sharing genetic information with their health care providers.

- A valuable strategy would be the identification of a mechanism through which existing information and materials could be shared and health care providers at the forefront of the evolution of genetic science could be identified.

- Mr. Brad Margus suggested that it is important to identify how the system is broken. For example, a physician should be able to recognize within a reasonable period that there could be some genetic basis for the problems a patient is having. Professionals also must be able to assess and evaluate how worthwhile and valid a technology may be in order to avoid wasting valuable health care resources.

- Mr. Citrin said that more professionals are needed who understand how to inform the public about genetics. How professionals learn about genetics and how they inform the public about its meaning will influence whether this new technology will create additional health disparities or reduce them.

- Dr. Kaytura Felix-Aaron, Agency for Healthcare Research and Quality (AHRQ), provided information on AHRQ's efforts in funding the information technology infrastructure in small community hospitals and in rural settings and in demonstrating the value of health information technology to health care.

- Dr. McCabe remarked that premedical education should impart to students an understanding of the importance of genetics. Some of the revamping of medical school curricula is treating genetics as a thread that runs throughout the medical curriculum rather than as a specific topic. Genetics must be made fundamental, exciting, and case-based. The politics of curricula and arguments involving the crowding of the curriculum need to be addressed. It also is important to consider the cost-benefits of genetics education and to ensure that students understand that this knowledge will prevent medical/legal mistakes and errors.

- Dr. Reed Tuckson suggested establishing a relationship between this effort and the Secretary's National Health Information Infrastructure Task Force so that SACGHS can help support the availability of the best evidence-based science to clinicians in an easy and accessible way. Ms. Carr noted that this would be an opportune time to make such a suggestion to the Secretary because the President has asked for a strategic plan on the improved use of health information technologies. She remarked that it would be helpful for the Committee to be specific about how this technology and genetics would dovetail.

- Dr. Sherrie Hans, Veterans Health Administration, suggested interfacing with the Secretary's health information technology initiative by proposing the elements of genetic knowledge that should be included as basic components of the electronic health record. This could include family histories. It also could be helpful to look at other kinds of tests and knowledge that should be included in the basic record and to look at what HHS and others are recommending. Looking forward, it also is important to think about bedside medical record programs and their interactive capability of those programs, as well as the kinds of daily prompts and guidelines that would be helpful to a practitioner.

- Dr. Suzanne Feetham, Health Resources and Services Administration (HRSA), discussed the importance of communicating about resources that are available to support the integration of genetics into practice, research, and education, and she discussed HRSA's efforts and resources in this area and in the areas of cultural competency and workforce diversity. These resources, along with agency mechanisms for working with other federal and nonfederal partners, could be of use in addressing some of the issues raised in the draft resolution.

- Mr. McInerney discussed how providers are thinking carefully about how genetics manifests itself in their own practices and then selecting core competencies. He said that both professionals and the public must be well educated and that education must rest on the same conceptual base and assumptions about genetics.

- Dr. Dunston remarked that public education is an absolute necessity and that consumers must be engaged in understanding genetic knowledge and in participating in genetic research. She underscored the importance of research that engages the public, including both healthy and affected individuals. Having a public that is excited about the power of science can help us understand disease in a way that will lead to new approaches to prevention and health promotion and to solutions about how to protect privacy and confidentiality.

- Dr. McCabe emphasized that consumer/patient involvement is important, because in order to utilize the power of genetics, large population studies will be needed. Until patients feel that their genetic information in their medical records is safe, they will not be willing to participate in research. This highlights the importance of inclusive genetic nondiscrimination legislation.

Members discussed the resolution and recommendations and together worked to refine their structure, meaning, and wording. A revised version was presented and discussed further on the second day of the meeting.

Report on the Inaugural Meeting of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
Reed V. Tuckson, M.D.
SACGHS Liaison to ACHDGDNC

Dr. Tuckson serves as a nonvoting liaison to Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC), which is charged with providing advice and recommendations to the Secretary concerning grants and projects authorized under Section 1109 of the Public Health Service Act. ACHDGDNC also is charged with providing technical information to develop policies and priorities for this program that will enhance the ability of state and local health agencies to provide screening, counseling, and health care services for newborns and children having, or at risk for having, heritable disorders.

Dr. Tuckson said that the first meeting included discussion about the state of the art and the challenges involved in screening newborns for genetic disorders and that the committee decided to focus on a few key areas in which it could make a significant impact. Its preliminary goals included recommending a uniform panel of conditions for newborn screening, assessing the capacity needs of states' newborn screening programs and whether and how they should implement them (realizing that there is an extraordinary amount of variability in how states require, mandate, and conduct newborn screening activities), and making recommendations on parent education, notification, and informed decision making.

The committee also plans to study newborn screening tests and to use its work product to develop recommendations on a core set of tests to be adopted nationwide by refining and extending a study that HRSA commissioned with the American College of Medical Genetics (ACMG). The committee will look at the costs of individual genetic tests and the costs of tests at the state level, including administrative costs. Finally in this area, the committee will carefully consider state-specific issues, such

as workforce needs, assessment of technologies and their translation into practice, and public education.

Furthermore, recognizing that the addition of new screening tests is likely to accelerate, the committee plans to develop a system or a process by which it will consider whether and how these new tests should be integrated and incorporated into the diagnostic panoply available for newborn screening. The committee is interested in the research needed to improve newborn screening and will use the ACMG study to help identify gaps in knowledge that would then be the focus of research initiatives. It also will encourage the creation of a working group on interagency coordination. Dr. Tuckson noted that the committee was impressed by SACGHS' strategic planning effort and priority-setting process and was given information on the process and lessons learned. A summary of the meeting will be made available.

Public Comments Session

Kathleen Rand Reed, B.A., M.A.A.
The Rand Reed Group

Ms. Rand Reed, an anthropologist and an ethnomarketer who has looked at genetic issues in communities of color, particularly in hypersegregated communities, discussed a poster that she presented at an NCHPEG meeting entitled "U.S. Prison Policies, the Baby Daddy, and Genetics in the Hood," the key components of which were: the removal and incarceration of young black males from hypersegregated inner-city neighborhoods and some rural towns; the high incidence of teenage mortality from violence, poverty, and poor health; the resulting skewing of the male to female ratio; and the phenomena of multiple matings, which often result in high consanguinity levels. In discussing the matings and familial relationships, she noted there could be possible increases in autosomal recessive disorders in this population due to half-sibs mating. She said that some leaders of genetic organizations have indicated that a major obstacle to addressing this issue is the fact that genetic counselors are hesitant to work with this population for fear of being accused of supporting or encouraging eugenics and that the Committee should consider constructing bridges that allow for open discussion to occur that would help protect such communities.

Another issue involves the increasing numbers of lawsuits initiated by minorities regarding reactions to prescriptions drugs. This is calling attention to a scientific and legal gap that causes injury to both the minority patient and physician. Pharmaceutical manufacturers have an automatic defense through the learned intermediary doctrine, which says that the prescribing physician or institution is liable to the patient but not the manufacturer. She noted that SACGHS already has identified a flaw in this process, which is that the pharmaceutical industry has little incentive to do pharmacogenomic studies on already marketed drugs or generic drugs, thus undermining the development of new information related to different populations and having implications for the diverse representation of the population in clinical trials. This is an important issue in the area of health disparities and one that the Committee should review.

Dr. Leonard commented that in Japan there are requests that all drugs on the market be pharmacogenetically analyzed for the Japanese population. She asked Ms. Reed if she would advocate that the federal government mandate this for all ethnic populations in this country. Ms. Reed mentioned that literature searches can provide a wealth of information on this topic and she agreed that pharmaceutical manufacturers of the drugs that are already on the market should begin to think about revamping.

Dr. Leonard suggested that through FDA, drug manufacturers could include in information packets what

ethnic populations were included in drug studies. This also may help physicians identify adverse reactions in other populations that have not yet been studied. Ms. Reed responded that some of that information was being included in DTC advertising.

Andrew Faucett, M.S.
Member, Board of Directors
American Board of Genetic Counseling

Mr. Faucett explained that knowledge about genetics and its social and ethical implications is becoming increasingly essential for many health professionals and must become an integral part of the medical curriculum. The draft resolution on genetics education and training of health care professionals developed by SACGHS clearly acknowledges this need and makes several solid recommendations related to integrating genomic concepts into the health care system. However, the American Board of Genetic Counseling (ABGC) advocates for a distinct and separate focus on the education and training of genetics health care professionals to be included in the resolution, because the demand for genetic counseling services will continue to increase, and without more training of genetic counselors there will not be enough of them to provide care and to educate other health care providers. He emphasized the need for adequate financial resources for training for the entire health care system and for specialized training in genetics.

He noted that the recommendations in this area will be successful only if there is an adequate genetics workforce to implement them. He also emphasized that if genomic medicine is going to be equally accessible and practiced in a culturally sensitive manner, individuals from minority populations must be recruited into the genetic counseling profession, and programs must be established in less-populated areas of the United States. He strongly urged the Committee to make specific recommendations that would support the continued training of individuals in the field of genetic counseling, encourage increased diversity in the profession, and encourage those uniquely trained individuals to demonstrate their competency through certification and licensure.

Barbara Handelin, Ph.D.
Handelin Associates

Dr. Handelin noted that she has worked with the Institutional Review Board community in improving the oversight of genetics research involving human subjects, especially research that has been sponsored by private industry, and is routinely amazed by how often the genetic basis of a set of symptoms is not considered in a differential diagnosis. She discussed the reasons this may occur and emphasized that there is a reluctance to consider alternative diagnoses because of a lack of time to incorporate new ideas, new concepts, and new knowledge. She emphasized the importance of promoting the development and implementation of information technology systems that can support routine clinical practice, including those that can suggest genetic etiologies or contributions to common disease in everyday practice, even though a genetic etiology is an uncommon explanation for many presenting symptoms. However, she cautioned that because such practice tools have been rejected by many physicians, using them may have to become a requirement.

She emphasized the critical importance of conducting large population studies across the United States that include the various strata of the country's diverse population groups in order to understand population variability. She asked the Committee to focus on finding ways to identify the main barriers to the ethical and productive conduct of large studies and noted that the simple promise of guaranteed health insurance and health coverage will go a long way toward encouraging people to participate in such

large research protocols. She added that it is paramount that a system of clear consequences be implemented for wrongful acts regarding privacy and confidentiality, and she reminded the Committee about the importance of focusing on the principles of respect for persons, beneficence, and justice in the course of its work.

Session on Coverage and Reimbursement

Overview of the Draft Coverage and Reimbursement Report
Cynthia E. Berry, J.D.
Chair, Coverage and Reimbursement Task Force

Ms. Berry presented an overview of the draft coverage and reimbursement report and proposed recommendations to improve coverage and reimbursement for genetic technologies, thereby improving access to them. Dr. McCabe said that the goal for this session would be to discuss the draft and develop recommendations for the report to the Secretary, although it was likely that this work would not be finalized today.

Discussion of Draft Report and Development of Recommendations

Dr. Stephen Phurrough, Centers for Medicare & Medicaid Services (CMS), and others emphasized the need for strong evidence to serve as a basis for making coverage decisions for genetic technologies (especially for the Medicare population) and the need to remove the Medicare screening/preventive services exclusion. It was noted that although the Committee could encourage legislation in this area, it remains a difficult barrier to surmount and would require significant support from others. The possible role of the U.S. Preventive Services Task Force (USPSTF) in developing the evidence base was discussed, as was the need for a process for conducting technology assessments for genetic technologies when they emerge from the research stage. It was noted that the development of the evidence base is the most important link missing between research and practice and that an orderly process of transition that uses existing regulatory oversight mechanisms, along with public and private partnerships to help identify what is ready for use and what is not, will be required. To help improve coverage decisions, a suggestion was made to recommend that the Secretary task the appropriate agencies to conduct a technical assessment that identifies current genetic technologies, the evidence base for them, the gaps that remain, and the knowledge needed to fill them, including the need for clinical trials.

Members discussed the possibility of the Secretary commissioning such a study through one of the HHS agencies, such as AHRQ and its USPSTF or the Evidence-based Practice Centers (EPC) program. Dr. Winn-Deen noted that it might be worthwhile for SACGHS to look at a guidance document that a NCCLS group is developing on clinical utility.

Members discussed the importance of focusing its recommendations on the areas of coverage and reimbursement that have the greatest likelihood of producing an impact. Cost issues should be framed for the Secretary, with some focus on royalty fees, as well as information about who pays for and who receives the benefits of preventive medicine. One suggestion was to add information on the economics of evidence-based coverage decisions.

There was discussion of using the SACGT-developed definition of genetic tests in this report. That definition focuses on heritable germline variations, or mutations that cause disease risk, but also includes somatic mutations and potentially even infectious disease applications. There was general agreement that SACGHS should use SACGT's work in this area in some form, but that there may be instances when

the definition of genetic technologies should include all types of genomics beyond heritable and other times when somatic and germline should be distinguished. It was agreed that, while the focus would be on germline mutations, a sidebar in the document would be added that would deal with somatic mutations and other genomic technologies.

Some members wondered whether focusing on issues that HHS can control or on the insurance industry as a whole would have the greatest impact, and on how this should be presented to the Secretary. Another major point of discussion was whether coverage should be considered separately from reimbursement. Some argued strongly that reimbursement should be considered a separate issue regardless of coverage, noting that it is not necessary to decide coverage before considering reimbursement. It also was noted that reimbursement is an issue pertinent to all insurers—Medicare, Medicaid, and the private sector—and that it is a critical issue that represents a significant barrier to access. Recommendations should encompass not only testing but genetic counseling and other types of services that also are inadequately reimbursed. Current Procedural Terminology (CPT) codes and how they apply to genetic testing and reimbursement also need to be addressed.

On coverage, SACGHS needs to address the screening issue, as screening is the one service Medicare does not cover. SACGHS could then suggest that the Secretary recommend to Congress that screening evaluations become a routine part of the Medicare coverage and that CMS be allowed to make those decisions based upon the best data available. Another issue to specifically address is how to determine when the use of a genetic counselor is appropriate.

Discussion then turned to the topics of health disparities and provider education and training and whether they should be included under coverage and reimbursement or stand separately. Some Members argued that these issues should stand on their own. In the area of health disparities, it was noted that it is important to recognize that economic disparities exist and can lead to health disparities and that expanding coverage increases access. Members discussed the implications of establishing criteria for making coverage decisions based on evidence that does not adequately cover certain ethnic populations. Dr. McCabe remarked that an argument already has been made that a different evidence base for different populations may be needed when making recommendations for coverage.

It was suggested that the ways in which the coverage and reimbursement system can create health disparities could be a theme continuing throughout the report. Another suggestion was to remark at the end of the report that overcoming problems involving coverage and reimbursement for genetic technologies does not assure that such technologies will deliver the full promise of genetics to medical practice, as other barriers are present and must be addressed. Lack of access itself is a major barrier to the integration of genetics into health care, and as such it also could serve as a central theme of the report. It was noted that the Committee's recommendations may directly affect access to genetic services.

It was agreed that provider education and training are being addressed in another resolution and that resolution could be cross-referenced in the coverage and reimbursement report. Dr. McCabe also suggested adding to the concluding text some of the data showing that USPSTF guidelines are still not fully utilized. The discussion on the draft report and recommendations for coverage and reimbursement was continued on the second day of the meeting.

Tuesday, June 15, 2004

Public Health Approach to Genetics

Muin J. Khoury, M.D., Ph.D.
Director
Office of Genomics and Disease Prevention, CDC

Dr. Muin Khoury discussed three themes related to public health approach to genomics: the changing landscape of genetics; the role of public health in improving health through scientifically-based approaches; and some of the roadmap activities that the Centers for Disease Control and Prevention (CDC) and other partner organizations have begun to develop.

Dr. Khoury said that we are moving toward conceptualizing genetic information as information that affects all diseases, including the 90 to 95 percent of diseases that normally are not thought of as genetic. He noted that an important paradigm shift in how we view disease is occurring that is illustrated by a continuum that ranges from pure genetic disease to pure environmental disease, with a break somewhere indicating the point at which the genetic services model does not apply. This complex puzzle of gene/environment interaction is where we currently find ourselves in the use of genetic information for the prevention or management of most human diseases.

In the area of public health, a shift can be discerned in the recognition of the importance of genetics in reports issued by the Institute of Medicine (IOM). For example, in 1988 IOM held a meeting that led to a pronouncement called "The Future of Public Health," which did not mention genetics; however, last year IOM, in revisiting the future of the public's health in another report, did discuss genetics and talked about the need for government, academic partners, employers and businesses, the media, communities and those involved in the health care delivery system to work together to help the field of public health develop and translate new science into activities that improve the health of all. The three gaps that need to be filled in this area are determining what it means to have genetic variation; determining the value that genetic information adds to treatment and prevention; and sorting out how to implement change in the system, which can be messy in a health care system that is not prepared for genetics.

Three major initiatives in the genomics and population health research arena, are the Human Genome Epidemiology Network, the National Health and Examination Survey projects, and genomics and acute public health investigations. Dr. Khoury also discussed initiatives to build public health capacity and practice and develop approaches for population-based monitoring and outcomes research and CDC efforts in ensuring the laboratory quality of genetic testing and practice. For example, in response to a DTC campaign that was conducted in two test cities, Atlanta and Denver, CDC surveyed health departments to determine the campaign's effect. In addition, CDC is working with Myriad Genetics to analyze Myriad's utilization data.

Dr. Khoury noted that some of the material used in CDC's systematic review of genetic tests could be useful to SACGHS's efforts to categorize and provide guidance for coverage and reimbursement and that he would be happy to provide any relevant material requested by SACGHS.

Discussion of the Draft Vision Report
Emily S. Winn-Deen, Ph.D.
Facilitator

Dr. Winn-Deen reminded the Committee that at the March meeting they had decided to prepare a summary of the systematic issue prioritization process instead of a vision report. Since the Committee's charter describes a vision and role for SACGHS, the Committee determined that a summary of its process and issue briefs would be of more use to the Secretary. Dr. Winn-Deen recapped the discussion at the March meeting and asked if there were any comments on the summary, *Toward a Vision of the Integration of Genetics and Genomics in Health and Society*, which will become a public document and one of the Committee's work products. Members discussed adding the term *genomics* to the document as well as definitions of *genetics* and *genomics*. Dr. McCabe suggested that the clinical practice side is referred to as genetics, while genomics refers to the analytical side, and that although the difference may be subtle, to have credibility within the genetics/genomics communities, SACGHS needs to try to use both terms. Members agreed that the report accurately represented the prioritization process and the conclusions of that process. Dr. McCabe suggested adding a brief glossary of key terms. Several changes were suggested including the addition of text regarding diversity in the workforce and cultural competency to the issue brief on genetics education and training.

The Committee discussed some of the public comments received, including a response by America's Health Insurance Plans, and a factual disagreement regarding Employment Retirement Income Security Act. Dr. McCabe said the Committee should check factual disagreements and make the appropriate corrections. He also indicated that discussions at every meeting have addressed whether discrimination by health insurance plans is occurring and that this will be explored at the next SACGHS meeting. It was suggested that a brief summary describing the issue briefs should be added to the beginning of the appendix, that a reminder to readers should be added to the end of each brief noting how that issue was prioritized, and that the briefs should be categorized in priority rather than alphabetical order. It was agreed that the new title of the document would be *The Study Priorities of the Secretary's Advisory Committee on Genetics, Health, and Society: A Roadmap for the Integration of Genetics and Genomics in Health and Society.* After the document is edited with the changes discussed by the Committee are made, the document will be sent to Drs. McCabe and Winn-Deen for final review. After the Secretary receives it, it will be posted to the web site.

Public Comments Session

Joseph McInerney, M.S.
NCHPEG

Mr. McInerney suggested that the Committee address the distinction between genetics and genomics directly in the vision document, because from his experience in working with a broad range of health professionals in the last few years, it has become evident that there is a great deal of confusion about these terms. He said that SACGHS should provide clarification for those who will read this document who have no background in genetics. He emphasized that genomics should not obviate genetics and that the Committee should define the terms up front to provide context to the work that follows. Dr. McCabe remarked that genetics is the study of inherited traits, while genomics is the study of genomes, and that this point will be made clearer in the document.

Michael P. Murphy, M.Sc.
President and CEO ,
Gentris Corporation

Mr. Murphy read a written statement in favor of the resolution concerning DTC marketing of genetic tests. He emphasized that Gentris and other companies have worked diligently to bring to market new tests that have the potential to decrease serious adverse events and allow for a more rational practice of medicine. He said that most in the industry believe that these tests are best conducted in Clinical Laboratory Improvement Amendments (CLIA)-certified laboratories, and he urged the Committee to recommend the necessary legal and regulatory changes that are needed to ensure that progress toward offering these potentially life-saving tests in the best medical setting possible can be continued. As part of his comments, Mr. Murphy noted that it is understandable that patients will seek genetic information that might be used for drug treatment, and he cited the May issue of *Reader's Digest*, which featured an article entitled "Genetic Breakthroughs: Making Medicine Safe." Dr. McCabe asked him to provide his written comments and a copy of the *Reader's Digest* article for the record.

Dr. Winn-Deen asked Mr. Murphy what his recommendation would be for the CLIA-certified laboratories that also are marketing directly to consumers and whether the American Association for Clinical Chemistry (AACC) committee is working on some recommendations in this area. Mr. Murphy responded that the group is not working on any recommendations but emphasized that what is important is that the marketing information be honest and fair to the consumer and that the consumer receive tests through a health care provider so that the benefits to be derived are honestly and fairly described. He commented that well-established medical utility tests, such as some that are currently coming out for predicting adverse drug reactions, are being mixed into discussions about other less well-established tests.

Kelly Ormond, M.S.
National Society of Genetic Counselors

Kelly Ormond, incoming president of the National Society of Genetic Counselors (NSGC), said that NSGC believes that the vision report and the issue briefs accurately reflect its understanding of the issues and encourages SACGHS to continue to address them as proposed and discussed today. Ms. Ormond addressed three areas: the draft resolution on DTC advertising; the draft report on coverage and reimbursement of genetic services; and NSGC's concern regarding the draft resolution on the genetics education and training of health care providers. With regard to DTC marketing, NSGC supports an individual's right to full disclosure of all appropriate medical information regarding genetic testing and believes that genetic counseling services by a board-certified or board-eligible genetics professional should be an essential component of any genetic testing program that is marketed directly to consumers. NSGC agrees with SACGHS's statements in the draft coverage and reimbursement report that billing by genetics counselors is limited by the current lack of CPT codes and by the lack of inclusion of genetic counselors as nonphysician Medicare providers. NSGC encourages SACGHS and the Secretary's office to consider ways to address these two issues. It also asks that SACGHS promote the development of federal funding to support evidence-based studies of both genetic technologies and clinical genetic services.

In addressing the draft resolution and issue brief on genetics education and training of health care providers, NSGC applauds SACGHS' efforts to actively consider the issues that impact the genetics health care workforce and to recognize the educational efforts that already are occurring. However, although NSGC strongly believes that the provision of quality genetic medicine requires the involvement

of health care providers of all specialties, it is concerned that this draft resolution does not address the need for additional training of genetic specialists and the need to expand the current number of certified genetics providers.

In addition, in order to address the issues in health disparities raised in Healthy People 2010, SACGHS must consider the limited cultural and ethnic diversity of current genetics professionals and the fact that most genetic specialists currently work at academic medical centers that often are limited in their ability to provide outreach to underserved regions or populations. She commented that there continue to be multiple impediments to increasing the training pipeline for both medical geneticists and genetic counselors and that an infusion of federal funding would increase the number of quality genetic training programs in a short period of time.

NSGC urges SACGHS to actively address the education and training needs for both specialists and nonspecialists to ensure the availability of a competent genetics workforce in the future and is willing to work with SACGHS to develop an issue brief and draft resolution reflecting this approach. Ms. Ormond noted that genetic counselor training programs are willing to consider expansion if funding is available for support. Asked to provide her thoughts on the tension between increasing the number of specialists versus integrating genomics knowledge into the practice of daily medicine, Ms. Ormond said that NSGC is incorporating into its strategic plan a consideration of how genetic specialists may become integrated into the various areas of genomic medicine.

Gail Javitt, J.D., M.P.H.
Genetics and Public Policy Center

Ms. Javitt said that the analysis of DTC marketing of genetic testing must clearly distinguish between the advertising of genetic tests and their commercial availability, as each of these activities is subject to distinct systems of regulatory oversight and is amenable to different possible policy solutions. She commented on the Federal Trade Commission (FTC) role and its statutory mandate to protect consumers, but noted that concerns about the impact of DTC advertisements on consumers that are unrelated to their veracity would not likely provide a sufficient basis for FTC intervention and that the government is significantly constrained by the First Amendment in regulating commercial speech. In addition, FTC must choose its enforcement actions carefully based on the nature and magnitude of the harm caused by the advertising in question. She said that the Genetics and Public Policy Center (GPPC) recommends that the Committee consider ways to foster the gathering of data on the possible harms and benefits of DTC advertising to consumers, which could be provided to FTC to be used as a basis for its involvement. She said that overall she is trying to draw a distinction between information provided to consumers and products or tests, for which foreseeable harms could indeed be a basis for intervening before concrete harms are demonstrated.

She said that it is unclear what entity would have the authority to implement the draft resolution recommendation that genetic tests should not be sold directly to consumers without the informed guidance of appropriately trained health care professionals, because no federal or state entity regulates when or under what circumstances genetic testing services may be commercially offered to consumers or health care providers. GPPC also questions FDA's willingness to enhance oversight of genetic tests without a clear mandate, particularly in the absence of more concrete evidence of consumer harm. She noted that the draft resolution does not mention CLIA, even though more rigorous oversight of genetic testing laboratories under CLIA could enhance public health protection. She suggested that SACGHS could have an important role in identifying the benefits and drawbacks to the public health of a more rigorous system of oversight by the federal government and that the Committee should foster data

collection concerning consumer impact of DTC genetic testing. The Committee also should consider the merits and drawbacks of a federal oversight entity that would set standards that genetic tests must meet before they are made commercially available.

Mr. Daynard clarified FTC's legal authority, remarking that all advertisements would be subject to FTC jurisdiction, as FTC law requires only that an advertisement be likely to mislead consumers in terms of purchase or use decisions. He also said there is no per se First Amendment protection for deceptive commercial speech and that there is a much higher First Amendment burden on FDA than on FTC.

Discussion of Draft DTC Resolution
Christopher Hook, M.D.
Chair, DTC Task Force

Before the Committee considered the draft DTC resolution, Dr. Guttmacher provided an update about a National Human Genome Research Institute (NHGRI)-organized workshop on the status and future of DTC advertising of genetic technologies held on March 23, 2004. The workshop participants generally agreed that it would make sense to facilitate the development of a stakeholder consensus document or white paper outlining best practices for DTC advertising in the realm of genetic tests and perhaps in genetic services as well. There was some discussion about the distinction between testing and services, and. It was agreed that a formal petition to FTC outlining the concerns with current DTC advertising practices for genetic tests should be developed. Finally, participants suggested the need for more research, involvement in specific collaborations with the private sector, and the development of a research agenda to inform future advertising practices and policy development. Dr. Guttmacher said he would provide the Committee with a list of attendees, including industry and provider representatives. Dr. McCabe called Members' attention to the written public comments regarding DTC marketing in response to the resolution and reminded them to note the public comments heard this morning.

Dr. Hook noted the work of the Task Force and staff in developing the draft DTC resolution. DTC marketing was ranked fourth on the list of top-priority issues requiring in-depth study; however, the Committee thought that the topic warranted an immediate response in order to encourage and facilitate FTC's efforts in this area. Dr. Hook reviewed the process of creating the resolution and read the document to the Committee. The Committee discussed a number of topics including:

- Should the resolution emphasize DTC advertising or direct access or both, and should DTC marketing and direct access be treated separately?

- Data are needed on the types of marketing that is reaching consumers and whether harm is occurring. This information could inform future discussions and the development of recommendations. It was suggested that public health agencies such as CDC could begin collecting the necessary data.

- It is important to continue to discuss and educate the public about the importance of having genetic testing done in the context of a relationship with trained health care providers. It is also important to distinguish medically useful tests from unsubstantiated tests.

- Variation in state laws must be taken into account when considering the ability to market medical services directly to consumers.

- It is important to determine which agencies have jurisdiction in this area. Members discussed the scope of FDA jurisdiction. Dr. Mansfield said that the FDA enabling statute, as interpreted by General Counsel, says that in-house developed tests are not medical devices, but services. The Secretary could ask the FDA General Counsel to explore what authority the agency may already have in this area, and if it is limited, the Committee could ask the Secretary to seek further legislative authority if necessary. Any communications must be clear that the Committee is not moving into the area of oversight of laboratory-developed tests, which are regulated under CLIA. The Committee must be clear that it is asking the Secretary to clarify which agency has the authority to regulate false and misleading advertising and the delivery of services to the public.

Dr. McCabe reminded Members that the Committee undertook the process of drafting this resolution in large part because of a concern that misleading marketing was occurring and that the Committee would like to move forward quickly before conducting an in-depth study in order to give FTC and FDA the opportunity based on this resolution to look into any misleading DTC marketing that may be occurring.

After discussion, the Committee decided that the document will be sent to the Secretary in the form of a letter rather than as a resolution and that it will inform the Secretary that information: has been presented to the Committee regarding false and misleading DTC advertising; the Committee is concerned that it is not in the public's benefit and could perhaps be a detriment to public health; the Committee will be gathering more information; the Committee would like to have the Secretary identify ways in which the agencies under his jurisdiction can begin to work to identify the impact in this area; and the Secretary should have the relevant agencies work with FTC to deal with the issue of false and misleading advertising.

Discussion of Draft Resolution on Genetics Education and Training of Health Professionals (continued)
Joan Y. Reede, M.D., M.P.H., M.S.
Chair, Education Task Force

Dr. Reede read the resolution that had been revised to reflect the previous day's discussion. Discussion points included:

- The consensus from the previous day's discussion was that an example of an organization engaged in genetics education and training and with which HHS might collaborate was needed and that NCHPEG would be an appropriate organization to mention.

- Some of the items lack specific recommendations and are relatively vague and philosophical. Dr. McCabe said there is a role for both implementable recommendations and for presenting issues philosophically at times.

- The paragraph involving promoting and actively incorporating genetic information should discuss genetics as part of the general spectrum of health information rather than as exceptional.

The Committee began to discuss the recommendations and how they should be prioritized. It was decided that a hard copy of the document would be distributed to participants and that discussion would continue later in the afternoon.

Discussion of Draft Coverage and Reimbursement Report (continued)
Cynthia E. Berry, J.D.
Chair, Coverage and Reimbursement Task Force

The discussion on coverage and reimbursement continued from the previous day. Members agreed it was more appropriate for the report to focus on genetic tests and services rather than genetic technologies and that these terms need clarification. Also, it was stated that the public comments received during the meeting need to be incorporated into the report's next iteration.

Members discussed the merits of IOM or an HHS agency such as AHRQ conducting a study of the state of genetic testing. However, Dr. Khoury remarked that while there may be a need for such a study, the current need is to develop a rigorous methodology to assess the validity and utility of each genetic test by intended use. The Committee decided that, because the lack of an evidence base hampers coverage decisions, what is needed is a process to be developed for reviewing the evidence base for genetic tests. He explained that CDC, through a process it is calling ACCE, is developing a methodology that evaluates genetic tests' analytic validity, clinical validity, clinical utility, and ethical, legal and social issues. With the help of the Foundation for Blood Research, CDC has applied this framework to five genetic tests.

The discussion then turned to the need for a process for assessing the evidence base to help CMS and other payers make appropriate decisions about what tests to cover. Dr. Khoury, in providing more details about ACCE and its status, said that after the initial phase of trying to put all the technology assessment pieces together, the plan is to establish a working group that will arrive at a consensus for test methodology review and then review the entire spectrum of tests—old and new—and make some pronouncement of what is known and what is not known, probably using the AHRQ EPC reviews. This information then would be posted on the web to try to influence interim policy. Because many gaps would come to light during this process, work would proceed with NIH and others to fund the various research efforts that are needed to fill those gaps.

Dr. Khoury said this process should proceed in a way that engages all stakeholders. Dr. Khoury also described ACCE-plus—that is, ACCE plus the other technologies that have been evaluated by USPSTF, the Cochrane Collaboration, and the Canada Technology Assessment.

Dr. Khoury noted that at this time it would be sufficient for the Committee to communicate with the Secretary about the importance of such a process and to provide information to him about the need for the different kinds of activities to be implemented and coordinated by the various agencies that are under his jurisdiction. No single agency alone will be able to move this process forward. He also remarked that the process does not focus on oversight or regulation but rather represents a way in which an evidence base can be developed for the evaluation of new tests and the integration of those tests into practice by working together with academia and professional organizations.

In addition, Dr. Khoury explained that this process is not necessarily geared toward driving coverage. Rather, it would summarize what is known and what is not and where the gaps are so that further research can be done to fill them. In the interim, the information that is available can be used for some policy or guideline development, and could leverage a discussion of the coverage issues. It was noted that the request for coverage in the absence of sufficient evidence will become increasingly common in the world of genetics and genomics, and it is important that some entity summarize the status of information so that both consumers and health care providers are armed with the right evidence at any given point in time. Dr. Khoury said he would provide more information about the process at the October meeting.

Dr. McCabe suggested that the recommendation generally addresses the need to develop a well-thought-out methodology or process for evaluating the evidence and looking at all the factors that insurers or federal health programs need to review in order to assess whether a particular technology or service is covered. The Committee can then refer to this effort as a potential model for SACGHS to monitor, without coming to a firm conclusion about what the precise criteria should be, because it sounds like that process already is under way through the ACCE effort.

Comments included a concern about the possibly of ultimately establishing a higher standard for coverage and reimbursement for genetic tests and services than for other medical interventions and treatments. Also, it was not clear whether payers have been asked about what kind of information would be most useful to them as they make decisions.

Coverage Conclusion

A mechanism is needed for assessing the sufficiency of the evidence base for establishing clinical utility of genetic tests, test by test. The gaps that are found in the evidence base and in various applications will then identify areas that require further research. In those areas, Request for Applications could be offered that would attempt to identify and prioritize tests that could impact heavily on the public's health; the hemochromatosis model was discussed as a pertinent example. It was concluded that this process would examine the evidence using specific quantifiable methodologies, identify the gaps for a particular test, and loop back to the research phase where more knowledge is accumulated, while at the same time communicate in a transparent way what is known and not known so that the right coverage decisions can be made and the public and health care providers can be better informed. The Committee would not recommend that any particular federal entity or structure conduct this process, but rather would examine the approach already under way at CDC and evaluate it as it progresses with a view toward determining whether it is a model that can be used across all federal health programs and in the private sector.

It was noted that the Committee could talk about the need to remove the screening exclusion, which would require a legislative change and is not something the Secretary could do unilaterally. Although the Committee cannot lobby Congress to make this change, it can reference in its report that the Secretary could include it in the administration's budgetary submission to Congress.

Reimbursement Conclusion

To move forward on reimbursement, the CPT code system needs to be addressed. The Committee considered choices for an individual who could address the Committee on this issue included someone from the American Medical Association or Mark Synovec from the College of American Pathologists. The Committee reviewed the reimbursement portion of the document and discussed changes in the language.

Health Disparities and Provider Education and Training Conclusion

The issues of health disparities and provider education and training were removed from the recommendations as separate subjects and will be referenced and discussed in the document as they relate to coverage and reimbursement.

Discussion of Draft Resolution on Genetics Education and Training of Health Professionals (continued)
Joan Y. Reede, M.D., M.P.H., M.S.
Chair, Education Task Force

Discussion points:

- The text regarding assuring equitable access will be changed to say that the education of health care and public health care professions is a *component* of ensuring equitable access.

- It was agreed to use both the terms *genetics* and *genomics* throughout the document and to use the phrase *health care and public health professionals* where appropriate. Also instead of using the term *genetics/genomics revolution*, *genetics/genomics knowledge* will be used.

- It was suggested that two bulleted items, one involving training for the implementation of models and the other involving promoting communication to enhance dissemination of these models, should be combined and the language clarified. Instead of using the term *pipeline programs*, the phrase *inter-educational K-12 and undergraduate pipeline programs* will be used.

- Members discussed whether a recommendation should be made about genetics training programs for specialists. One remark was that this document was written with the goal of looking across the full spectrum of health professionals, with the idea that the Committee later would review the issues involving specific disciplines. It was decided that a reference to specialized genetics training would not be included and that the focus of the document will be on improving genetics education and training across disciplines and provider types.

- It should be noted that many other subspecialists need training on the genetic anomalies within their particular organ system subgroups.

- It was suggested that the issue of not having sufficient numbers of genetics professionals is more pertinent to the coverage and reimbursement report, as the real driver of the number of genetics professionals is who gets reimbursed and to what degree.

Report on the NHGRI Working Group on Large Population Studies
C. Christopher Hook, M.D.
SACGHS Liaison to NHGRI AGES Work Group
Alan Guttmacher, M.D.
Deputy Director, NHGRI

Dr. Hook, the SACGHS liaison to an NHGRI working group organized to consider large population studies, said that the working group has had two meetings and one phone conference and has discussed issues such as the ability to utilize existing population cohort studies in a new collaborative or consolidated fashion versus starting a new project. The group also discussed some of the complex issues involved, such as statistical power, recruitment, single versus rolling informed consent, how to follow a population over the span of a lifetime, and how to account for technological changes that will occur during that period. He said that at least one more meeting is planned for August, as well as other phone conversations, before the group issues a report. He noted Dr. Francis Collins's May 27 article in *Nature* and a modification to the Request for Information (RFI) for public comment.

Dr. Guttmacher noted that the RFI requests input on 14 specific points or on other issues, such as how to utilize existing cohorts and how a new cohort might provide additional benefit. Many comments have been received and have been helpful to the working group, and it is hoped that by the end of the summer or early fall, the group will have achieved its purpose of determining what the science and to a smaller degree the logistics of such a study might look like. At that point, officials at NIH and other HHS agencies and the administration will need to determine whether to move forward. Dr. McCabe asked Members to review the 14 points and to respond to them if they wish.

Plans for Future Meetings

Genetic Discrimination

Individuals with experiences with or problems related to genetic discrimination will address the Committee during the October meeting. To identify these individuals, the Committee will speak with the Genetic Alliance and other groups and to Mr. Paul Miller to help identify those who have come through the Equal Employment Opportunity Commission.

Coverage and Reimbursement

The Committee discussed the possibility of hearing from individuals who have been refused coverage or have not been reimbursed for genetic services and its impact on their health care. Dr. Khoury will provide an update on the CDC working group effort for ACCE-plus, and a speaker to be identified will address the Committee on the process of establishing CPT code reimbursement, both of which will inform the discussion of the draft report at the next meeting. It also was suggested that someone should speak who can explain how coverage decisions are made. Committee Members will review the revised coverage and reimbursement report before it goes out for public comment, and in addition to Ms. Berry, participation on the task force will be increased for this effort to include Dr. Winn-Deen, Dr. Leonard, Dr. Khoury, and Dr. Tuckson.

Patents and Access

A brief presentation at the next meeting will be requested on the National Academy of Sciences' committee on patents and licensure.

Pharmacogenomics

A Task Force was appointed to begin addressing the topic of pharmacogenomics before the October meeting. Members include: Dr. Winn-Deen, Dr. Hunt Willard, Dr. Leonard, Dr. Hook, Dr. Feetham, and Dr. Phurrough.

Large Population Studies

A Task Force was appointed to begin addressing the issue of large population studies prior to the October meeting. Members include Dr. Hook, Dr. Willard, Dr. Guttmacher, and Dr. Khoury.

The meeting was adjourned.

We certify that, to the best of our knowledge, the foregoing meeting minutes of the Secretary's Advisory Committee on Genetics, Health, and Society are accurate and correct.

Edward R.B. McCabe, M.D., Ph.D.
SACGHS Chair

Sarah Carr
SACGHS Executive Secretary