DEPARTMENT OF HEALTH AND HUMAN SERVICES

Eighteenth Meeting of the
SECRETARY'S ADVISORY COMMITTEE ON
GENETICS, HEALTH, AND SOCIETY
March 12-13, 2009

Meeting Summary

Hubert H. Humphrey Building
200 Independence Ave., SW
Washington, DC

Prepared by the Office of Biotechnology Activities
National Institutes of Health

Participants

March 12, 2009

Committee Members Present

Steven Teutsch, M.D., M.P.H., Chair
Mara Aspinall, M.B.A.
Sylvia Mann Au, M.S., CGC
Paul Billings, M.D., Ph.D., FACP, FACMG
David Dale, M.D.
Gwen Darien
Rochelle Dreyfuss, M.S., J.D.
James P. Evans, M.D., Ph.D.
Andrea Ferreira-Gonzalez, Ph.D.
Kevin T. FitzGerald, S.J., Ph.D., Ph.D.
Julio Licinio, M.D.
Barbara Burns McGrath, R.N., Ph.D.
Joseph Telfair, Dr.P.H., M.S.W., M.P.H.
Sheila Walcoff, J.D.
Marc S. Williams, M.D., FAAP, FACMG
Paul Wise, M.D., M.P.H.

Ex Officio Members/Alternates Present

Sharon Alexander, on behalf of Stuart Ishimaru, J.D. (Equal Employment Opportunity Commission)
Michael Amos, Ph.D. (Department of Commerce/National Institute of Standards and Technology)
Sarah Botha, J.D. (Federal Trade Commission)
Michael A. Carome, M.D. (HHS/Office for Human Research Protections and Office of Public Health and Science)
Robinsue Frohboese, J.D., Ph.D. (HHS/Office for Civil Rights)
Phyllis Frosst, Ph.D., on behalf of Allan Guttmacher, M.D. (HHS/NIH/National Human Genome Research Institute)
Denise Geolot, Ph.D., R.N., FAAN (HHS/Health Resources and Services Administration)
Naomi Goldstein, Ph.D. (HHS/Administration for Children and Families)
Alberto Gutierrez, Ph.D. (HHS/Food and Drug Administration)
Alan E. Guttmacher, M.D. (HHS/NIH/National Human Genome Research Institute)
Sherrie Hans, Ph.D., on behalf of Ellen Fox, M.D. (Department of Veterans Affairs)
Peter Kirchner, M.D., on behalf of Daniel Drell, Ph.D. (Department of Energy)
Katherine Kolor, Ph.D., on behalf of Muin Khoury, M.D., Ph.D. (HHS/Centers for Disease Control and Prevention)
Kerry Leibig, J.D., on behalf of Stuart Ishimaru, J.D. (Equal Employment Opportunity Commission)
Sue McAndrews (HHS/Office for Civil Rights)
Douglas Olsen, Ph.D., R.N., on behalf of Ellen Fox, M.D. (Department of Veterans Affairs)
Gurvaneet Randhawa, M.D., M.P.H. (HHS/Agency for Healthcare Research and Quality)
Jeffrey Roche, M.D., on behalf of Barry Straube, M.D. (HHS/Centers for Medicare & Medicaid Services)
Barry M. Straube, M.D. (HHS/Centers for Medicare & Medicaid Services)
Amy Turner, on behalf of Thomas Alexander, J.D. (Department of Labor)
Daniel J. Wattendorf, Lt. Col, on behalf of on behalf of COL. Scott D. McLean, MC, USA (Department of Defense)

SACGHS Staff

Sarah Carr, Executive Secretary, NIH Office of Biotechnology Activities
Kathryn Camp, M.S., R.D.
Andrea Collins, Committee Management Staff, NCI
Tara Hurd Faunteroy
Cathy Fomous, Ph.D.
Darren Greninger, J.D.
Linda Silversmith, Ph.D., Consultant
Abbe Smith, Capital Consulting Corporation

Speakers

Barry M. Straube, M.D. (SACGHS *Ex Officio*, Centers for Medcare & Medicaid Services)
Sylvia Mann Au, M.S., CGC, (SACGHS Member)
William (Greg) Feero, M.D., Ph.D. (National Human Genome Research Institute)
Christy White [via telephone] (Cogent Research, LLC
Larry Thompson, (National Human Genome Research Institute)
Lyla Hernandez, M.P.H. (Institute of Medicine/National Academies)
Amy Miller, Ph.D. (Personalized Medicine Coalition)
Ann Willey, Ph.D., J.D. **(**New York State Department of Health)
Kevin FitzGerald, S.J., Ph.D, Ph.D., (SACGHS Member)
Larry Gostin, J.D., Institute of Medicine Committee on Health Research
and the Privacy of Health Information)
R. Rodney Howell, M.D., Chair, Advisory Committee for Heritable Disorders
in Newborns and Children
Stanley Crosley, Eli Lilly
Thomas Croghan, Ph.D. **(**Mathematica Policy Research)
Andrew Nelson (Health Partners Research Foundation)
Barbara Burns McGrath, R.N., Ph.D. (SACGHS ,Member)
Mara Aspinall, M.B.A. (SACGHS Member)

Public Commenters

Theresa Lee (Advanced Medical Technology Association)

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Michael Amos, Ph.D. (SACGHS *Ex Officio*, National Institute of Standards and Technology)

Mara Aspinall, M.B.A. (SACGHS Task Force on Priority Setting)
Robert Epstein, M.D. (Medco)
Barry Straube, M.D. (SACGHS *Ex Officio*, Centers for Medicare and Medicaid Services)
Bruce Quinn, M.D., Ph.D. (Foley Hoag, LLP)
Samuel Nussbaum, M.D. (WellPoint, Inc.)
Joanne Armstrong, M.D., M.P.H. (Aetna) [via telephone]
Michael Critelli, J.D. (Pitney Bowes)
Richard Luetkemeyer, M.D. (Caterpillar, Inc.)

Public Commenters

Daryl Pritchard, Ph.D. (Biotechnology Industry Organization)

March 12, 2009

Opening Remarks

Dr. Steven Teutsch, Chair of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), welcomed everyone to SACGHS's 18th meeting. After inviting members of the public to sign up to speak during the meeting's public comment sessions, he greeted new Committee members—Gwen Darien, David Dale, Sheila Walcoff, Samuel Nussbaum, and Charmaine Royal—and reported on the resignation of Dr. Paul Miller, due to his appointment as a member of President Obama's transition team.

Goals. The goals of the meeting were:

(1) to examine direct-to-consumer genetic services
(2) to be updated on informed consent issues for sharing genomic data and to consider next steps
(3) to learn about preliminary survey findings from the SACGHS Task Force on Genetics, Education, and Training
(4) to hear from a roundtable of public and private payers related to one of SACGHS's new priorities─genetics and the future of the U.S. health care system
(5) to be updated on federal activities relevant to SACGHS

Conflict of interest reminder. Ms. Sarah Carr, SACGHS Executive Secretary, reminded Committee members that they are special government employees, for whom there is a set of rules of conduct to follow. She especially noted the rules sections on conflicts of interest and lobbying.

Patents draft report. After the SACGHS Task Force on Gene Patents and Licensing Practices completed revisions to the draft report proposed at the December 2008 SACGHS meeting, the draft report became available for public comment on March 9. The deadline for comments is May 15.

Member activities. Dr. Paul Billings was a keynote speaker for the initial meeting of the Center for Translational and Policy Research on Personalized Medicine, which will conduct policy reviews of personalized medicine. The center is located at University of San Francisco and was founded by its director Catherine Phillips.

SACGHS no longer has a formal liaison to the Advisory Committee on Heritable Disorders in Newborns and Children due to a charter change by the latter, but SACGHS staff attended that Committee's February meeting.

Dr. Teutsch participated in an Institute of Medicine meeting of the Roundtable on Translating Genome-Based Research for Health on February 12, which included a workshop on developing systems for evidence generation for clinical utility. Members of the Roundtable also developed a plan to explore three subtopics in greater detail: (1) the effects of genetics and genomics on drug development, (2) the process for translating research discoveries into genetic diagnostics, and (3) the potential value of genetics to medicine and public health.

Staff. Kathryn Camp, senior health policy analyst, recently joined the staff of the NIH Office of Biotechnology Activities. She will be the staff lead for the SACGHS Task Force on Genetics Education and Training.

Update from Centers for Medicare & Medicaid Services (CMS)

SACGHS *Ex Officio*, Dr. Barry Straube, Acting Chief Medical Officer and Acting Director of the CMS Office of Clinical Standards and Quality, noted that CMS is heavily involved with the American Recovery and Reinvestment Act of 2009, the revised Children's Health Insurance Program Reauthorization Act of 2007 (CHIPRA), and the Medicare Improvements for Patients and Providers Act of 2008 (MIPPA), in addition to helping the administration with health care reform. Dr. Straube noted that the Committee's exemplary work over the last number of years provided a base for a broad national discussion on genomics and how it fits into health care reform.

Current Medicare coverage of genetic testing. Medicare beneficiaries are currently covered for genetic tests that are intended to diagnose specific diseases. Cytogenetic testing is covered under a national coverage determination (NCD) for "reasonable and necessary" tests such as genetic disorders of the fetus (e.g., trisomy 21), failure of sexual development, chronic myelogenous leukemia, acute leukemias, and myelodysplasia. Dr. Straube explained that "reasonable and necessary" is defined in the statute and subsequent regulation as a service, treatment, or device that will lead to improved outcomes in the Medicare patient population. He also noted that there has been a tremendous change in genetics since the cytogenetics NCD, and it should be refined.

Local coverage determinations (LCDs). About 85 percent of coverage decisions are made by a contractor medical director at the local level. The LCD system has evolved over 30 to 40 years, and under current law, CMS can guide but not overturn the decisions of local contractors. In the age of electronic communications, Dr. Straube plans to promote centralization and creation of national norms. He recognizes that local decisionmaking can be more flexible and more responsive to local needs, however, local decisionmaking results in a lack of consistency across Medicare administrative contractors and can be limited by local resource constraints and a lack of local subject expertise. The CMS website for local coverage determinations can be found at http://www.cms.hhs.gov/DeterminationProcess/04_LCDs.asp .

Dr. Straube explained that genetic testing is considered a noncovered screening test for patients who do not have a relevant illness, injury, or sign or symptom of a disease. If they are asymptomatic and do not have a past or present history of disease, under the current statute and regulations genetic testing is not covered. For example, cancer genetic testing is permitted only for a patient with a personal history of cancer (i.e., Medicare does not cover genetic tests based on family history alone). In addition, the covered test must be useful for managing a patient and affecting his or her management options. Medicare does not cover a genetic test for a clinically affected individual for purposes of family planning, disease risk assessment of other family members, or when the treatment and surveillance of the beneficiary will not be affected.
Additional specifications of LCDs for genetic testing include that patients receive pre-test genetic counseling by a qualified and appropriately trained practitioner, .patients sign an informed consent form prior to testing, and genetic analysis is provided by a laboratory that meets the requirements of the Clinical Laboratory Improvement Amendments (CLIA).

Council on Technology and Innovation. The Medicare Modernization Act of 2003 established a Council on Technology and Innovation (CTI), which Dr. Straube co-chairs with a political appointee. CTI facilitates the exchange of information about new technologies, particularly questions about coverage and payment policy. It supports a coordinated response to inquiries from the general stakeholder community when there are questions about new technologies and whether they are covered, coded, and/or reimbursed by Medicare. CTI includes a Genomics Working Group that provides support to CTI on genomics and personalized medicine issues.

In 2008, CTI e-published an *Innovators' Guide to Navigating CMS* (available at http://www.cms.hhs.gov/CouncilonTechInnov/Downloads/InnovatorsGuide8_25_08.pdf) to assist stakeholders in understanding the processes used to determine coverage, coding, and payment for new technologies under the Medicare fee-for-service program and facilitate timely introduction of innovative technology for care of beneficiaries. Dr. Straube suggested that CTI could consider recommendations that SACGHS has made to the Secretary of HHS regarding the coverage and reimbursement of genetic tests.

Medicare Evidence Coverage Advisory Committee (MEDCAC). MEDCAC assists CMS in determining the characteristics of evidence to support coverage decisions about medical services, items, and devices for Medicare beneficiaries. The Committee is comprised of 100 members who are selected for their expertise in specific topic areas and broadly represent the health care stakeholder community. MEDCAC met February 25, 2009, to consider diagnostic, prognostic, and pharmacogenomic applications of genetic tests.

The recommendations from the February 2009 MEDCAC meeting included: (1) using a standard framework and methods to evaluate evidence about the diagnostic use of genetic testing; (2) supporting clinical studies that provide patient-focused health outcomes data on the use of genetic results in care management; and (3) encouraging collaboration among federal agencies involved in research and health care policy.

Preventive services benefits. While not part of the original Medicare program, select preventive service benefits were added 10 to 15 years ago for certain diseases such as breast, colon, and prostate cancers, and cardiovascular disease. In addition, Section 101 of MIPPA granted the HHS Secretary the authority to consider other preventive services benefits—such as those with an "A" or "B" rating from US Preventive Services Task Force—through the Medicare NCD process. Dr. Straube noted that MEDCAC will meet again May 6 to discuss genetic screening tests as a preventive service benefit for Medicare beneficiaries. It will advise CMS on ways to use Section 101 of MIPPA to address some of the issues that SACGHS has previously identified.

CMS and the oversight of genetic testing. CMS regulates laboratory testing under CLIA, which establishes quality standards to ensure the accuracy, reliability, and timeliness of patient test results. CLIA standards for laboratories performing moderate- and high-complexity testing also apply to laboratories that provide genetic testing. CMS has initiated development of a proposed rule to update the proficiency testing (PT) regulations for PT programs, which will address genetic tests and better alternate assessment mechanisms. CMS is also working with laboratories offering direct-to-consumer genetic testing to ensure compliance where CLIA is applicable.

Future NCD topics. Consideration of an NCD for genetic testing for responsiveness to the drug Warfarin began in August of 2008, and the proposed decision will be out no later than May 2009. The key question is does the evidence show that the use of genetic testing leads to improved outcomes in patients who are placed on Warfarin. CMS invites public participation in this and future NCDs. Dr. Straube explained that anyone can request an NCD, but the key element is to include preliminary evidence that supports an NCD for a particular test. In December 2008, CMS published 20 potential NCDs for public comment. Dr. Straube noted that two of the areas are ripe for potential NCD topics—gene expression profiles in oncology and pharmacogenetic tests.

Dr. Straube concluded his presentation by stating that SACGHS had raised CMS's awareness of genetic issues and that CMS is responding to the Committee's recommendations.

Discussion. Dr. Marc Williams remarked that he had served on a Wisconsin carrier advisory committee (CAC) and while the quality of CACs can vary, 14 years of experience had shown him that local CACs can be more nimble in making coverage determinations. He wondered if the May MEDCAC meeting would be an appropriate venue for CMS to consider the SACGHS recommendation regarding the evidence-based use of family history. Dr. Straube pointed out that operationalizing family history is problematic due to difficulties in making payment and reimbursement decisions based on someone's recollection or misinformation passed down in the family.

Dr. Andrea Ferreira-Gonzalez commented on the significant differences in local policies around the country regarding genetic testing. For example, some policies restrict the use of a genetic test to once per lifetime even though somatic mutations could make the test relevant more often, an issue she feels needs to be addressed. Dr. Ferreira-Gonzalez also asked whether CTI interacts with local directors to bring them up-to-date on issues discussed at the national level.

In response to Dr. Ferreira-Gonzalez's first comment, Dr. Straube explained that one of the factors that generates an NCD is inconsistency among local carriers. CMS may open an NCD to make sure that coverage is uniform. On the other hand, if there is an NCD but you disagree with the local coverage decision, you can ask local carriers to reopen their coverage decision if you think their evidence is wrong. Regarding Dr. Ferreira-Gonzalez's second question, Dr. Straube responded that there are increased interactions between local carriers and outside experts and stakeholders. However, neither CMS at the national level nor the local carriers have sufficient resources for conducting education and creating an NCD for every topic. Possibly the recent consolidation of local contractors into 15 administrative groups for the whole country will help to address local differences.

Dr. David Dale inquired about the scope of the database for national and local coverage decisions—how could a provider or member of the public know what is paid for from one place to another?. Dr. Straube replied that the database, while bulky and in need of improvements in terms of workability, is searchable and includes all of the Medicare Administrative Contractors (MACs) and their policies. In additions, each MAC maintains a website for information at the local level.

Updates from SACGHS *Ex Officios*

Dr. Teutsch invited SACGHS *ex officios* to describe new programs and activities that respond to the American Recovery and Reinvestment Act (ARRA), particularly those related to SACGHS' mission.

Equal Employment Opportunity Commission **(EEOC).** Kerry Leibig, Senior Attorney Advisor in the Office of Legal Counsel, stated that EEOC is the lead agency responsible for enforcing federal laws that prohibit employment discrimination on the basis of race, color, sex, national origin, religion, age, disability, and in retaliation for protected activity. In 2008, EEOC got a new responsibility when President Bush signed the Genetic Information Nondiscrimination Act, or GINA. She explained that GINA has two titles. Title I addresses the use of genetic information in the health care industry, and it is administered by HHS, Department of Labor, and the Treasury Department. Title II, which will become effective on November 21, 2008, prohibits the use of genetic information in making employment decisions. It prohibits the deliberate acquisition of genetic information about job applicants and employees by employers. It also has strict confidentiality requirements for any genetic information that an employer does obtain..

On March 2, 2009, EEOC published the proposed rule for Title II in the *Federal Register*, with a request for public comments by May 1, 2009. The implementation date for regulations outlined in this rule is May 21, 2009. The EEOC website provides the proposed rule, a question-and-answer document that

reviews the bascis of the rule, and statements made at the February 25, 2009, Commission Meeting (see http://www.eeoc.gov/eeoc/meetings/2-25-09/index.cfm).
The remedy and enforcement provisions for Title II of GINA are modeled on Title VII of the Civil Rights Act.

Agency for Healthcare Research and Quality **(AHRQ).** Dr. Gurvaneet Randhawa from the Center for Outcomes and Evidence explained that AHRQ's mission is to improve the effectiveness, safety, quality, and efficiency of health care. To carry out this mission, an evidence-based evaluation process is used to inform decisionmaking for the development of guidelines and evidence-based practices. The U.S. Preventive Services Task Force (USPSTF), sponsored by AHRQ, and the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group, sponsored by Centers for Disease Control and Prevention (CDC), are examples of this process. USPSTF focuses on clinical prevention while EGAPP is more focused on treatment and management in the clinical context.

Dr. Randhawa also described the work of AHRQ's Evidence-Based Practice Center (EPC) Program. For example, to inform decisionmakers about emerging technologies, the EPC Program has developed reports for CMS on emerging genetic tests for cancerous and non-cancerous conditions. He also noted that AHRQ has done a fair amount of work on family history and mentioned that a conference sponsored by the National Institutes of Health (NIH) on the state-of-the-science on family history and primary care is scheduled later this year, which will build on work of AHRQ and CDC.

Dr. Randhawa concluded his remarks by noting other AHRQ-sponsored programs that aim to build a better evidence base, including the Developing Evidence to Inform Decisions about Effectiveness (DEcIDE) Network and the Centers for Education, Research, and Therapeutics (CERTs). In addition, AHRQ sponsors activities to disseminate knowledge from its evidence assessments and projects that addresses how to put evidence-based research into practice (e.g., clinical decision support tools for BRCA testing and for gene expression profiling tests).

When asked how ARRA funding will effect programs, Dr. Randhawa replied that AHRQ has built a track record on comparative effectiveness over the past three years based on the following three elements: assessing the evidence base, gathering new evidence, and disseminating information in a usable format. He expects these elements will be incorporated in upcoming ARRA-based programs.

NIH. Dr. Phyllis Frosst, Science Policy Analyst in the Office of Policy, Communications, and Education, at the NIH National Human Genome Research Institute (NHGRI), reported that ARRA funding added $10.4 billion to NIH's $27 billion FY2008 budget. ARRA funds are being allotted as follows: $1 billion for extramural repairs, construction, and alterations and $300 million for shared instrumentation and other capital equipment purchases, which will be administered by the National Center for Research Resources; $500 million for buildings and facilities on the NIH campus; $400 million from AHRQ for comparative effectiveness research; and $8.2 billion for NIH's scientific research priorities, which will be divided among the NIH Institutes and Centers (ICs) in proportion to their budgets and to be spent over the next 18 months. Of the $8.2 billion, the NIH Office of the Director receives $800 million, and the remaing $7.4 billion goes to the NIH ICs and the Common Fund. NIH aims to apply management lessons learned during the years in which the NIH budget was doubled.

The funding of R01 grants will be a first priority, however; these funds can be guaranteed for only two years, with some designated for administrative and competitive supplements to existing grants. Interested individuals can find out about ICs' priorities on the NIH website (www.nih.gov). Challenge grants will also be highlighted to encourage research that can be accomplished within two years. One appropriate area for challenge grants is bioethics; including areas on informed consent and data access, ethical issues in the translation of genetic knowledge to clinical practice, unique ethical issues posed by emerging

technologies, electronic sharing of health information, and recontact issues in genotype and genome-wide association studies. Others topics for challeng grants include biomarker discovery and validation, integrating cost effectiveness, personalized drug response and toxicity, new computational and statistical methods, enhancing clinical trials specifically for rare disease genetic patient registries, and a number of subtopics under genomics.

HHS Office for Civil Rights (OCR). Dr. Robinsue Frohboese reported that OCR has two responsibilities: (1) enforcing Federal laws and regulations that prohibit discrimination on the basis of race, color, national origin, disability, age, sex, and religion in federally funded programs and activities and (2) administrating the Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule.

Dr. Frohboese described a number of activities that OCR has undertaken since the December 2008 SACGHS meeting. OCR has a responsibility under GINA to modify the HIPAA Privacy Rule to ensure that health plans do not use or disclose genetic information that is protected health information for underwriting purposes. It is coordinating its efforts with EEOC, CMS, and the Departments of Labor and Treasury, which are also responsible for implementing GINA regulations. OCR is also involved in health information technology. In December 2008, HHS issued a Privacy and Security Toolkit, which OCR was very involved in developing along with the Office of the National Coordinator (ONC) for Health Information Technology. OCR provided guidance that applies the HIPAA Privacy Rule to various principles of protecting the privacy, confidentiality, and security of electronic health exchanges, with a particular focus on access to electronic health records.

ARRA includes a separate Act entitled the Health Information Technology for Economic and Clinical Healthcare (HITECH) Act, which is devoted to the use of health information technology in health care reform. Subtitle D of the HITECH Act focuses on privacy and security concerns associated with the electronic transmission of health information and requires OCR to engage in rulemaking to strengthen the civil and criminal enforcement of the HIPAA Privacy Rule. The HITECH Act increases the amount of penalties that can be collected from violators of the Rule, and for the first time, it gives enforcement authority outside of HHS. State attorneys general will have the authority to bring actions in federal court. The HITECH Act also applies certain HIPAA privacy and security provisions directly to business associates of HIPAA-covered entities, and it calls for HHS to promulgate regulations requiring health care providers, health plans, and other covered entities to notify individuals when their health information is breached. Public education is another duty for OCR under HITECH. Congress has directed OCR to engage in a nationwide, multifaceted initiative to educate consumers about uses and disclosures of protected health information, as well as their rights under the HIPAA Privacy Rule. OCR has 11 months to develop an education plan.

The HITECH Act provides $20 billion for grants that promote the use of electronic health records and standard-setting in health information technology. The majority of this funding—$18 billion—goes to CMS to promote adoption and use of electronic health records among providers. The remaining $2 billion will be distributed through ONC, and a Departmental working group has been set up to develop proposals and a spending plan.

Dr. Frohboese concluded her remarks by mentioning that as part of the Surgeon General's release of the new Family History Tool, OCR took the opportunity to provide information through frequently asked questions (FAQs) about the HIPAA Privacy Rule and how it impacts ability to collect and use family history information. The FAQs are availabe on the OCR website (http://www.hhs.gov/ocr/privacy/familyhealthhistoryfaqs.pdf) and provide a valuable addendum to the Family History Tool.

Federal Trade Commission (FTC). Attorney Sara Botha, Bureau of Consumer Protection, Division of Advertising Practices, explained that FTC is a national consumer protection agency. Its mission is to prevent unfair and deceptive acts and promises in commerce, whichincludes misleading advertising. A primary area of focus within the Division of Advertising Practices is advertising claims for products that promise health benefits. These products include over-the-counter drug products, dietary supplements, and direct-to-consumer (DTC) genetic tests. Ms. Botha explained that consumers who are misled by advertising claims not only may lose money, but the products may negatively impact health. She added that consumer education is an important mechanism to help prevent consumer deception. In addition, FTC takes law enforcement actions and, where possible, works with industry on self-regulation. In conclusion, Ms. Botha noted that FTC has sent inquiries to some DTC companies and is reviewing their advertising claims and comparing it to the state-of-the-science.

Department of Veterans Affairs (VA). Dr. Douglas Olsen, Nurse Ethicist at the VA National Center for Ethics in Health Care, reported that as part of its clinical services, the VA is in the process of hiring a director of molecular medicine to oversee and coordinate efforts in the clinical genomics and related areas such as proteomics. The molecular medicine program will provide education and clinical guidance to physicians, nurses, laboratory technicians, social workers, and other staff as well as patient education. There are also plans to develop a central clearinghouse for genetics resources, but it may take a couple years to implement these plans.

The VA formed a Genomic Medicine Program Advisory Committee in 2006. Based on a recommendation from this Advisory Committee, focus group surveys were conducted by the Genetics and Public Policy Center to assess veterans' knowledge of genetics and genomics and also their support and expectations for the Genomic Medicine Program. The results of these surveys are in press and will be published in spring 2009.

Two VA programs for information technology infrastructure were recently funded. The Genetic Information System for Integrative Science, or GenISIS, will integrate data from individual genetic and clinical research studies, reanalyze the data and produce new findings. The Veterans Informatics and Information and Computing Infrastructure, or VIICI, will integrate existing and new databases to extract information and meaning while providing data in a secure, high-performance computing environment.

VA is also working with the National Coalition for Health Professional Education in Genetics (NCHPEG) to develop an interactive, web-based, educational program on familial syndromic colorectal cancer. The content will include pathophysiology, risk assessment based on family and medical history, screening, management, testing, and counseling. It is intended for a wide audience of health care professionals and is scheduled to be ready to pilot-test by the end of FY2009.

Dr. Olsen concluded his remarks by describing VA research efforts. In 2008, VA funded a genome-wide association study on amyotrophic lateral sclerosis to examine gene-environment interactions in the development of the sporadic form of this disease. VA also plans system-wide studies in Parkinson's disease, post-traumatic stress disorder (PTSD), mental illness, diabetes, breast cancer, and pharmacogenomics. There are also more than 140 investigator-initiated merit-reviewed projects related to genomics on a wide spectrum of conditions prevalent in veterans, including schizophrenia and bipolar disorder, PTSD, Alzheimer disease, cardiovascular disease, diabetes, substance abuse, stroke, and autoimmune disease.

HHS Administration for Children and Families (ACF). Dr. Naomi Goldstein, Director of the Office of Planning, Research, and Evaluation explained that ARRA funding supports several ACF programs including Early Head Start, childcare subsidies for the Temporary Assistance for Needy Families program, Child Welfare, Child Support Enforcement, and a new initiative to build capacity in nonprofit

organizations. Dr. Goldstein looks forard to exploring with SACGHS how the Committee and ACF can interact.

Department of Energy (DOE). Dr. Peter Kirschner explained that DOE's Office of Science supports research on bioenergy, waste cleanup, and carbon sequestration. It also supports a small program in radiochemistry research and radionuclide imaging instrumentation that in the past created much of the scientific underpinnings for nuclear medicine. This program is being reoriented toward support of the bioenergy and environmental remediation projects In addition, the Office of Science funds a small program devoted to low-dose radiation biology research, which might provide information regarding genetic susceptibility to radiation-induced cancer.

In the past, DOE's ethical, legal, and societal issues program focused narrowly on genetic privacy, education, and intellectual property protections, but it has new aims and is transitioning to bioenergy sustainability issues, synthetic biology, and nanoscience. The Office of Science also supports DOE's Human Subjects Protection Program, which is responsible for human subjects' protection at all DOE sites and any research done with DOE funds. It is this program that intersects with research that is directed at genetic testing, primarily via two large cohorts that have been studied through DOE. Atomic bomb survivors in Japan and their children and the health and safety of a cohort of workers exposed to nuclear and hazardous materials are being monitored. DOE collaborates with universities and other agencies to mine data from these cohorts for additional research.

Discussion

Regarding GINA, Dr. Williams asked Ms. Leibig and Dr. Frohboese how their two groups interact with respect to self-insured employers, who have not only the traditional role of the employer but also insure their workers through self-insurance programs. There are a variety of mechanisms under which that insurance is administered, but the self-insured employers do have rights to certain aspects of protected health information. Ms. Leibig replied that because Congress did not want double liability, there is a firewall between Titles I and II, and no one can make claims about the same situation under both titles at once. Dr. Frohboese commented that the Dr. Williams summarized the situation very well.

Dr. FitzGerald asked Ms. Botha whether FTC talks with other agencies regarding health claims. She replied that because FTC is not a scientific organization, it cooperates with other agencies relative to advertising claims. FTC also has memoranda of understanding with FDA. For example, with drug advertising, FDA regulates prescription drug advertising and FTC regulates over-the-counter drug advertising.

Public Comment Sessions

Dr. Teutsch welcomed public comments and noted that listening to the public is an important function of SACGHS.

March 12, 2009, comments

Advanced Medical Technology Association (AdvaMed). Theresa Lee, Vice President of Payment and Health Care Delivery Policy for AdvaMed, commented that the Association, which represents the medical device and diagnostics products industry, has members around the world in both large and small firms and includes a significant number of in-vitro diagnostics firms. AdvaMed has paid attention to SACGHS's work regarding genetic tests and has commented before at public sessions.

Ms. Lee made the following comments: (1) Because health care providers rely on clinical diagnostic laboratory tests to inform and guide much of the care that they deliver, these tests play a critical role in determining whether we will achieve a more efficient and affordable health care system. She asked SACGHS to work with the administration to develop a health reform plan that builds on the promise offered by diagnostic tests. (2) Ms. Lee pointed out the need for a modernization of the Medicare clinical laboratory fee schedule. She reminded the Committee that it has been nine years since the Institute of Medicine (IOM) completed its assessment of Medicare laboratory payment policies. The IOM report called for a series of fundamental reforms of Medicare's clinical laboratory fee schedule, most of which have gone unaddressed. The report also warned that problems with the outdated payment system could threaten beneficiary access to care and the use of enhanced testing methodologies in the future. (3) Diagnostic tests pose difficult challenges for technology assessors, and AdvaMed believes that current evidentiary standards used to evaluate therapeutic products and procedures may not be appropriate for diagnostics. She hoped SACGHS' attention to this matter will lead to more appropriate standards.

Discussion. Dr. Teutsch asked Ms. Lee to reflect upon what the industry can do to help the government obtain the cost-effectiveness information needed to make a compelling case for the value of diagnostics. Ms. Lee replied that one way to show the value of certain technologies is by cost-effectiveness evaluation, and it may be appropriate to look at this issue in the context of MIPAA. Regarding diagnostics, AdvaMed is working to cosponsor a white paper that will examine the value of screening tests in disease prevention. When completed, AdvaMed hopes to share the white paper with SACGHS members.

March 13, 2009, comments

Biotechnology Industry Organization (BIO). Dr. Daryl Pritchard, Director of Research Programs Advocacy, identified BIO as the largest trade organization serving and representing the biotechnology industry in the United States and around the globe, which includes more than 1,200 biotechnology companies, academic institutions, state biotechnology centers, and related organizations. Regarding the topic of the future of the health care system, Dr. Pritchard said that BIO wishes to suggest and reiterate ways that SACGHS can help ensure that novel molecular diagnostics are used to improve outcomes and efficiency in health care delivery. First, payers need to recognize that innovative diagnostics have value in optimizing patient management and reducing the overall cost of an episode of care, and that the reimbursement of diagnostics needs to reflect this value. Furthermore, reimbursement should recognize the worth of prevention and management of chronic conditions, not just treatment of acute conditions.

Dr. Pritchard advised that SACGHS should submit an action plan for implementing the recommendations in the Committee's February 2006 report, *Coverage and Reimbursement of Genetic Testing Services*, and consider whether any areas in the report need to be updated or reemphasized, particularly related to some of the problems in the CMS rate-setting methodology. Other suggestions that SACGHS could recommend to the Secretary of HHS are to direct CMS to update and reform the antiquated clinical laboratory fee schedule and to consider how to create a transparent and predictable reimbursement system that reflects the value of diagnostic tests. Dr. Pritchard declared that the current CMS rate-setting methodology and clinical laboratory fee schedule are examples of a system that does not adequately reflect the value of molecular diagnostic tests and that inadequate reimbursement slows innovation. Another problem is the inconsistency among CMS contractors. He hopes that SACGHS's efforts will lead to improvements.

Session on Consumer-Initiated Use of Genomic Services

Session Overview and Purpose: Sylvia Au, M.S., CGC

In December 2008, the consumer-initiated use of genomic services was identified as one of SACGHS' priority topics. Ms. Au, who led the discussion of this topic area during SACGHS' priority-setting process, explained that the purpose of the session was to provide an update on government and private sector activities related to DTC genomic services since the Committee's July 2008 session on personal genome services. Following the presentations, the Committee considered how it could contribute to the current debate and discussion in this area.

NIH-CDC Workshop on Personal Genomics: William (Greg) Feero, M.D., Ph.D.

Dr. Gregory Feero, Chief of the Genomic Healthcare Branch in the Office of Policy, Communication, and Education at NHGRI, reported on the outcomes a December 2008 NIH-CDC workshop initiated by Dr. Muin Khoury, the CDC *ex officio* member of SACGHS. The workshop focused on the clinical validity and clinical utility of analyzing hundreds of thousands of single nucleotide polymorphisms (SNPs) across the genome, and education needs of providers and consumers. Approximately 100 participants contributed government, academic, and industry perspectives. The workshop was set in the context of (1) increasing accessibility of personal genome-wide testing because of decreasing costs and (2) rapid movement of research discoveries from genome-wide association studies of common diseases to the marketplace and the vigorous debate about how and when to translate discoveries into health care applications.

Dr. Feero noted that a consensus paper is under development that outlines the following recommendations from the workshop:

- Develop and implement industry-wide scientific standards for personal genomics services.
- Develop and implement a multidisciplinary research agenda, in conjunction with private-public partnerships.
- Enhance credible knowledge synthesis and dissemination of unbiased information to providers and consumers; this effort could build on of AHRQ, EGAPP, and others.
- Link scientific research on validity and utility to evidence-based recommendations for the use of personal genomic tests.
- Consider the value of personal utility, personal genomic information may have value to individuals beyond reducing morbidity and mortality.

Additional information about the workshop is available at http://cancercontrol.cancer.gov/od/phg/workshop.html.

Discussion. Dr. James Evans voiced concern about personal genomics being derailed by lack of objective standards. Dr. Feero replied that the solution might be to come up with methods and metrics for evaluating personal utility. Dr. Billings commented that personal utility, an evolving concept, is not a new issue in genetics and can depend on whether your perspective is that of a provider looking for immediately useful steps or of the individual with the genetic condition trying to plan for his or her future.

Dr. Marc Williams inquired about the assurance of analytic validity, and Dr. Feero replied that it must be taken into account but also noted that the workshop focused on clinical, not analytic, validity. Agreeing with Dr. Williams, Dr. Andrea Ferreira-Gonzalez agreed with the importance of considering analytic validity. Dr. Feero made the point that a genomic test could provide the correct genotype 99.9 percent of

the time, but only 15 percent of the time is that genotype actually reflective of actual risk. The major problem is not with analytic validity but with clinical validity.

Genomic Attitudes and Trends: Christy White (by teleconference)

Christy White, a principal at Cogent Research LLC, presented data from a three-year (2005-2008) longitudinal study about Americans' attitudes toward genomics over time that showed an increase in the awareness of genomics. By 2008, close to 80 percent of 1,000 consumers answering a 120-question, web-based survey said they had heard or read about using individual genetic information to understand or optimize health. However, only about 12 percent had heard of DTC genomic testing. Interest in using genomics for the purpose of understanding and optimizing one's health hovers at about 50 percent. However, when provided with a list of 40 specific conditions for which they could have a genetic test, 91 percent of respondents identified at least one test they would be interested in having.

The survey also revealed that Americans are not in agreement regarding what genomic tests can tell them about their risk for particular diseases. About a third said the test would identify the chance of getting a specific disease; 29 percent responded that the test would determine the level of risk (i.e., greater or lower than most people) but not the exact level of risk; and 17 percent thought that the test would indicate whether their genetic variants were similar to those associated with the disease but not whether they had any increased risk of developing the disease. About 7 percent of respondents said genomic testing would determine whether they definitely will or will not get a specific disease, and 4 percent believed that testing would determine whether they already had a specific disease.

The Cogent survey also probed the willingness to share genetic test results with others, such as family members and health care providers. Compared to 2006, Americans are less interested in sharing their results. For example, in 2006, 61 percent of respondents indicated they would share test results with their doctor versus 52 percent in 2008. In 2006, 34 percent of respondents would share results with their children, versus 25 percent in 2008. The willingness to share information with their doctors or families varied with education and age. College graduates and Americans with no children were more likely to say that nobody should have access to their results. Americans 55 or older were more likely to indicate that their doctor and children should have access to their genetic test results. However, if a genetic test indicated a risk for a specific disease, 88 percent of survey participants indicated they would reach out to their doctor, while only 30 percent would tell their family members. About half of the respondents said they would make lifestyle changes (e.g., diet, exercise) based on the test results.

At a previous SACGHS meeting, Cogent reported on perceived barriers to genetic testing, such as concerns about discrimination. Ms. Christy reported that awareness of the GINA legislation was quite low, and there was no change in consumer confidence that genetic information would not be used in a discriminatory fashion.

Discussion: Dr. David Dale asked whether the survey queried if people wanted their samples saved for future discoveries, or in some way asked about the concept of a biobank or storage of specimens. Ms. White replied that 46 percent of respondents indicated that DNA samples should be retained for future tests of their choosing. When asked who they would want to keep the DNA samples, two-thirds said they wanted the sample retained by the company that conducted the test. Less than 10 percent indicated that a government agency should have that information.

NIH Website for Consumer-Level Information about DTC Genomic Services: Larry Thompson, M.F.A.

Larry Thompson, Chief of NHGRI's Communications and Public Liaison Branch, spoke of the need for NIH to provide an information resource on genomic testing. The decision to create such a resource stems from concerns within the NIH leadership that complicated tests with nuanced interpretations of results were being offered directly to consumers, outside of the medical model. Dr. Zerhouni—NIH Director at the time—charged IC directors to develop a communication plan that would provide authoritative information for consumers interested in genomic testing. A trans-NIH committee was created to develop this plan.

As part of the efforts of the trans-NIH committee, the National Cancer Institute (NCI) funded 10 focus groups (in Chicago, New York, and Washington, DC area) involving 84 consumers and interviews with nine primay care physicians. Of the consumers, 61 percent were women; 39 percent were men; and 77 percent were white. All participants had high school diplomas, and many had been to college. Half had children and were worried about diseases that run in families. The consumers were stratified into three groups: people who were not considering genetic testing, people who were thinking about genetic testing, and people who had been tested. The recruiters were asked to try to find people who had purchased DTC tests from companies such as 23andMe or Navigenics, but they did not find any.

NCI's focus group findings were consistent with Cogent's work. Most consumers were broadly aware of genetic testing, which is probably why they agreed to participate in a focus group. However, most did not have a deep knowledge of genetic testing. Many did not want to know their risk of developing a diseases if there was no treatment or cure. Some said they wanted to know their risks, especially if they had a family history of a certain disease. Most consumers were very concerned about privacy and confidentiality, particularly regarding insurance companies and employers. Most participants also recognized that their abilities were insufficient to interpret test results and thought that a trained health professional should be involved in the testing process. All those who had taken a genetic test had done so because of a family history of a particular disease. In general, consumers wanted the government to provide reliable, unbiased information.

Among the 10 physicians, six were in small private practice, two in large private practice, and one in a hospital practice. Two of the phsicians had practiced one to 10 years, two had practiced 11 to 20 years, and five had practiced 21 or more years. The physicians reported few queries from their patients about genetic testing. However, they felt that their patients did not understand probability and did not know how to interpret the results of a genetic test. Most physicians indicated that NIH should provide information about genetic testing, but some thought NIH should not be involved because it is an issue between doctors and their patients.

Based on the data gathered by NCI, the trans-NIH committee learned that an NIH resource on genetic and genomic testing should provide information that is (1) basic, practical, and clearly explained and (2) developed for different audiences (e.g., consumers, health professionals).

Mr. Thompson recognizes that most consumers are not interested in genetic testing until and unless a problem arises in their own family. His goal for the new NIH website is that it be an authoritative, reliable, unbiased source of information when the time comes for a consumer to need it. In addition, the site needs to be engaging by using video blogs and video interviews and providing mechanisms for dialogs with site users.

Among the challenges is the lack of a budget for this project; everyone working on it is a volunteer. Even so, there needs to be a mechanism to ensure that the website (not yet named) will be kept up to date.

Discussion: Dr. Williams commented favorably on the video idea. He suggested including interviews of individuals who tell how they made their choices about whether to be tested and whether to tell their physicians the results.

Plans for the National Academies DTC Workshop— Lyla Hernandez, M.P.H.

Ms. Hernandez, a senior program officer at the National Academies' Institute of Medicine, reported that several units of the National Academies decided to work together for an academy-wide look at DTC genetic testing. The groups working together through a workshop planning committee include the Committee on Science, Technology, and Law; the Board on Life Sciences; the Roundtable on Translating Genomics-based Research on Health (which Ms. Hernandez directs); the Forum on Drug Discovery, Development, and Translation; and the National Cancer Policy Forum.

The goal of the workshop is to bring together scientific, medical, legal, and policy communities along with members of the public to examine issues, opportunities, and challenges, addressing the following areas of DTC genetic testing:

1. Current state of knowledge and future research trajectory in this area: What do we know about the analytical validity, clinical validity, and clinical utility of DTC tests? What types of genetic testing will become available over the next five to ten years?
2. Shared genes and emerging issues in privacy: How do we balance consumer desire for self-awareness that is driving this market against the need to protect privacy? What are the risks and benefits for family members of users of these tests? For public figures? For the legal system? Who owns an individual's genomic data? What are the implications of online social networks based on DTC genetic testing results?
3. Regulatory framework: What are the oversight roles of various agencies at state and federal levels? Are the claims verifiable? What are the codes of professional conduct for informed consent, analysis, and disclosure?
4. Education of the public and medical community: Are providers well enough informed about genetic testing? How do we ensure proper interpretation of the tests? What is the minimum knowledge required to make informed decisions based on DTC testing?

The workshop planning committee is considering dates in late summer or fall for scheduling the workshop. Ms. Hernandez can be contacted at lhernandez@nas.edu for further information.

Standards for Analytical Validity and Clinical Validity of Genomic Scans: Amy Miller, Ph.D.

Dr. Miller, Public Policy Director for the Personalized Medicine Coalition (PMC), explained that in July 2008, three companies that provide DTC genomic testing—23andMe, Navigenics, and deCODE—agreed to work with PMC on a set of standards for the scientific validity of gene scans. As a result of this effort, they adjusted their algorithms so that tests results are more consistent between companies. In addition, they have made their methods for calculating disease risk more transparent by making them available on their websites and the following NIH website: http://www.cancercontrol.cancer.gov/od/phg/docs/pmcscivalid.pdf. These companies also see a role for the government in building consensus on how to calculate disease risk and which SNPs to include in testing.

PMC also developed an educational guide to help consumers make informed decisions about DTC genetic testing. To obtain consumer input and with sponsorship from DTC genetic testing companies, PMC arranged for a roundtable discussion in February 2009. Feedback provided during this discussion was

incorporated into the consumers' guide. Copies of the are available at http://www.personalizedmedicinecoalition.org/sciencepolicy/public-policy_consumer-genomics.php . PMC also is planning to prepare a small educational brochure.

New York State Laboratory Requirements Relevant to Genomic Services Companies: Anne Willey, Ph.D., J.D.

Dr. Willey, Director of the Office of Laboratory Policy for the New York State Health Department, stated that she would focus on New York State's oversight of clinical laboratories. Oversight is the same whether these laboratories are engaged in genetic testing or other kinds of laboratory tests. All laboratories testing any specimen derived from the human body collected within the geographic jurisdiction of New York must have a permit from the New York State Department of Health, regardless of any other permits or accreditations. As part of this process, the State conducts onsite laboratory inspections whether the laboratory is in New York, another state, or out of the country.

Dr. Willey reviewed the requirements for (1) a qualified laboratory director (including a doctoral degree, four years postdoctoral experience in a relevant field, including a certain amount of recent experience), (2) the permit application and fee (ownership and financial interests, facility and equipment, personnel, test menu, initial and annual fees. And onsite inspection), and (3) assay validation (assay description, consent process, analytic and clinical validity, and reporting format that must be suitable for a nongeneticist).

As New York State is not a direct-access state, individuals cannot order their own laboratory tests, with some specific exceptions. Consequently, genetic tests are performed only at the request of a person authorized by law to make use of those test results—usually a clinician, generally a physician, or possibly a lawyer. In New York State, genetic counselors are not licensed health care practitioners and cannot order laboratory tests.

Laboratories must report the results only to the person who ordered the test. The laboratories may also communicate those results to the patient or person tested but only with written authorization of the ordering person; this report must be an exact copy of what was reported to the authorized person.

Billing must be direct to the person who was tested, which eliminates extra charges by unnecessary intermediaries. There are certain exceptions, such as if a laboratory is unable to perform a test and has to send the specimen to a different laboratory; in this situation, the first laboratory can bill the second one. If a company, such as DNA Direct, has arranged for the test that company bills the consumer for its services without getting involved in the laboratory billing the consumer or the consumer paying the laboratory.

New York State also has anti-kickback statutes so the laboratories cannot provide fiscal or other incentives to the ordering practitioner. The laboratory can provide genetic counseling education and general education on the types of tests available to the practitioner, but it cannot directly provide genetic counseling to the person tested. In New York, genetic counseling is considered an aspect of practicing medicine—which laboratories cannot do.

New York State also requires written informed consent, and the statute specifies eight elements. Four of those elements can only be described by the laboratory and include what test is being performed, the predictive value of the test, and what the laboratory will do with the specimen. The laboratory has to provide this information to the physician for the consent. The execution of the consent occurs between the ordering physician and the patient.

The State monitors the Internet to let entities know that they cannot offer tests to New York State residents without first obtaining a permit, and the State has been following up, as appropriate, with about 40 entities. Some respond by posting on their websites that they do not operate in New York. Others have obtained or are applying for State permits, or have been determined not to be a laboratory.

Committee Discussion

Dr. Evans asked about the entity that is not a laboratory, and Dr. Willey explained that entity is DNA Direct, which facilitates medical genetic activities but does not do any testing itself. Dr. Licinio described a hypothetical situation in which a nonresident of New York has a specimen collected in New York but it is sent out of state for analysis. Dr. Willey made clear that if the specimen is collected within the geographic boundaries of New York State, then the laboratory that performs the test is subject to the jurisdiction of the State of New York. She added that no laboratories are approved in New York State to offer whole genome scans.

Dr. Evans then inquired of all the speakers about their views on clinical utility as it was not discussed much in their presentations. Dr. Willey made clear that only the medical practitioner, not the testing laboratory, could talk with the patient about what to do about a positive test result under New York State law. Dr. Feero said the December 2008 NIH-CDC workshop dealt with clinical utility, but it is difficult to define. Dr. Miller agreed, adding that she believes that consumers are beginning to learn about probability through the discussions of their results.

Dr. Billings asked Dr. Willey for clarification on what genetic counselors employed by testing laboratories could do in New York State. She said that they can provide education to the ordering medical practitioners or guide them concerning the result; then only with written permission of the practitioners can they speak directly to patients. Dr. Billings then wondered what is working well and what needs to be changed in the N.Y. system. Dr. Willey stated that New York is satisfied with how the laboratories are applying for permits. She also noted that the State does make exceptions sometimes when a clinician says that a patient needs a particular test from a laboratory without a permit.

Dr. Willey added that the N.Y. program costs $20 million to run, has issued permits to 1,600 laboratories, has standards stricter than CLIA, and regulates more than 75 percent of U.S. genetic testing laboratories. By law, specimens must be destroyed after 60 days unless their owners allow further retention. Specimens can be retained "de-identified" for unspecified research. If it is retained in an identified format or used for genetics research, it must have an explicit genetics research consent. Gene scans can be retained if future mining of the data has been validated.

Dr. FitzGerald asked about public utility—data placed in a public database. Dr. Feero commented that one would then need some kind of measure of utility, which would vary depending on your needs as a payer, regulator, provider, or consumer.
Dr. Amos suggested that it would be timely to ask the panelists their advice concerning possible next steps and actions on DTC testing. Dr. Miller suggested that SACGHS be forward-looking as SNP technology will be completely different in five years.

Dr. Evans noted that claims must be reconciled with utility. If laboratories are going to make medical claims, then they have to be held to traditional models of clinical utility. carefully.

Proposal for Short-Term Action: Ms. Sylvia Au

Ms. Au proposed formation of a short-term Task Force to develop a brief document that reviews concerns about DTC genetic testing—such as limited data on clinical validity and utility of tests, consumer and

provider understanding of test results, privacy protection, companies that skirt oversight regulations, and false and misleading claims—and highlights prior SACGHS recommendations that address these concerns.. The paper would also identify issues that would benefit from further evaluation by SACGHS. Discussion. Dr. Ferreira-Gonzales declared that DTC issues need to be addressed in a separate report, as suggested by Ms. Au. Dr. Teutsch commented on the need to monitor DTC enterprises to identify trends in consumer usage and cost of DTC testing. . Dr. Amos remarked that the Committee should be forward-looking and consider how technology will change in five years, such as whole-genome sequencing becoming affordable.

Dr. Licinio predicted that genetic testing would become cheaper, which argued for the timeliness of a paper on DTC genetic testing. Dr. Frosst said, and Dr. McGrath agreed, that while the volume of tests is currently small, the broader issue is that people are buying or getting information for which the validity and utility are unknown and rapidly changing, which is why it is important for SACGHS to look at the issue.

Dr. Williams observed that physicians want a government repository of testing information, which fits with the prior SACGHS proposal for a centralized registry for genetic testing. Dr. Dale proposed pushing the research agenda to define where medical applications are most appropriate. Dr. Frohboese said that, regarding the privacy issue that has been cited, there is a difference between a true lack of privacy protections and an unawareness that such protections exist. Dr. Aspinall added that the Task Force should consider the efforts of other groups or organizations in this area.

Dr. Teutsch indicated that a brief document could be ready for the June 2009 SACGHS meeting. He proposed that a small Task Force work on the DTC issues and report in June. The following individuals volunteered to serve on the Task Force: Sylvia Au, Sarah Botha, David Dale, Jim Evans, Andrea Ferreira-Gonzalez, and Julio Licinio. Syliva Au will serve as Task Force Chair.

Session on Informed Consent, Privacy, and Discrimination Issues Related to Genomic Data Sharing

Session Purpose and Overview: Kevin FitzGerald, SJ, Ph.D., Ph.D.

Dr. FitzGerald noted how earlier discussions at this SACGHS meeting have tied in with the topic for this session. For example, Dr. Feero brought up ethical, legal, and social issues; Dr. Frohboese mentioned privacy; and Ms. White mentioned lack of public awareness of GINA. The bioethics area of the challenge grants that Dr. Frosst referred to includes many relevant topics, such as informed consent and data access policies, unique ethical issues posed by emerging technologies, health disparities and access to participation in research, electronic sharing of health information, the translation of genetic knowledge to clinical practice, the blurring between treatment and research, and recontact issues in studies with genome-wide associations. Dr. FitzGerald explained that the session will include presentations from two organizations that work in parallel to SACGHS—the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) and the National Academies Institute of Medicine (IOM)—followed by a discussion of the Committee's next steps in this priority topic area.

Informed Consent Issues of Concern to ACHDNC: R. Rodney Howell, M.D.

Dr. Howell, ACHDNC Chair, indicated that much of his Committee's work has focused on newborn screening. In the United States, 4.1 million babies are tested each year. At the current time, 30 tests, on average, are included in newborn screening panels, so about 120 million tests are performed using genetic technology. Although ACHDNC recommends national standards and national policies for newborn

screening, in actuality these tests are conducted under the auspices of State health departments. The variability among the states was a matter of some concern at the time of the creation of this Committee.

ACHDNC was authorized under the Children's Health Act of 2000 and began its work in 2004. At that time, 28 states in the country were screening for less than 20 of the ACHDNC core panel of 29 conditions. By December 2008, 49 states were testing for more than 21 of the 29 core conditions. The growing acceptance of the core set of conditions by so many states was due in part to ACHDNC developing national standards and in part to parental work at the state level.

The Newborn Screening Saves Lives Act of 2008, passed unanimously by both houses of Congress, reauthorized the Committee. This act also requires the Secretary of HHS to ensure the quality of laboratories involved in newborn screening and to develop a national contingency plan for newborn screening. The latter is in response to Hurricane Katrina, which destroyed the State newborn screening laboratory in Louisiana. The Act also authorized additional NIH research on new screening technologies and relevant disease management strategies, naming the NIH program the Hunter Kelly Newborn Screening Research Program.

ACHDNC has developed a nomination process for adding conditions to the core newborn screening panel. Among the requirements for this process are broad access so anyone can nominate a condition, transparency, consistent criteria, and a structured evidence review group. Previously, there was no formalized method for reviewing rare conditions because traditional approaches did not work well. The Committee has contracted with Dr. James Perrin of Harvard to conduct systematic evidence reviews for nominated conditions. The application to nominate a condition requests information in the following three areas: (1) the condition (e.g., incidence, timing of onset, severity), (2) the laboratory test (e.g., modality, clinical validity and performance data, method to confirm a positive screen), and (3) treatment (e.g., modality, urgency, efficacy, risk, availability).

Applications for nominated conditions first go through an administrative review at the Health Resources and Services Administration (HRSA) to ensure that they are complete. An internal ACHDNC workgroup then assesses the nomination package to determine whether there is likely to be sufficient information for each of the three components to support a full evidence review. Based on this assessment, the full Committee decides whether the nominated condition merits a systematic evidence review, which is a time consuming and expensive process.

The following areas are considered as part of the evidence review:

- Natural history of the condition, including variations in phenotype;
- Prevalence of genotypes and phenotypes;
- Impact and severity of the disorder;
- Methods of screening and data such as sensitivity, specificity, predictive values;
- Methods to diagnosis infants with a positive screen;
- Feasibility and acceptability of screening;
- Harms or risks of screening, diagnosis, and/or treatment;
- Benefits of treatment; and
- Costs of screening, diagnosis, treatment, late treatment; failure to diagnose in newborn period.

The review process involves a literature review of English-only publications (case studies are not included), analyses of any available raw data from unpublished studies, focus groups with experts familiar with the nominated condition (e.g., investigators, family members, clinicians), and data synthesis.

Evidence reviews summarize key findings and indicate the level of certainty of the evidence, what evidence is lacking, and if lacking, what particular evidence is needed for the Committee to reconsider the nomination. ACHDNC considers the findings of the evidence review and makes one of the following recommendations:

- Add the nominated condition to the core newborn screening panel.
- Do not add the nominated condition to the core newborn screening panel but conduct additional studies to supplement data that are promising.
- Do not add the nominated condition to the core newborn screening panel because there is insufficient evidence (e.g., studies are inadequate or inappropriate).
- Do not add the nominated condition to the core newborn screening panel because the data do not justify adding it to the panel.

The Committee's decisions are available on the ACHDNC website and are published in a journal. To date, no nominated condition has become part of the core panel.

Dr. Howell also reported that ACHDNC discussed two topics at its February 2009 meeting that are pertinent to the SACGHS session on data sharing. One topic was about translational research policy and included an overview of the regulation and oversight of research with children, regulatory options for multicenter research, alternative models of Institutional Review Boards (IRBs), and proposals to hold IRBs directly accountable for compliance with human subjects research regulations. As part of the discussion, the experiences of California and Massachusetts in obtaining informed consent from parents of infants screened for similar projects were compared. When California introduced tandem mass spectroscopy for newborn screening, it was deemed an experimental technology. A large pilot project was designed, which required written informed consent from parents invited to participate in the project. Only 25 percent of parents agreed to participate. On the other hand, Massachusetts had a similar type of project when the State decided to expand the newborn screening panel, and virtually all the parents consented. The expanded screening was optional and parental permission was required, but written consent was waived and verbal consent was documented. These differing experiences are of particular interest to ACHDNC as the Committee is aware of plans for multicenter trials related to newborn screening. The Committee plans to draft a white paper on IRB issues.

The second ACHDNC activity related to genomic data sharing is consideration of the storage, retention, and use of residual dried blood spot (DBS) specimens. Dr. Howell noted wide variation in state policies for the retention of residual DBS specimens. Some states discard them shortly after screening, and other states save them for months, years, or indefinitely. Some reasons for retaining DBSs are for quality assurance of existing tests, to help establish new screening tests, to confirm/rule out conditions in infants with positive screens, and retrospective analysis of biomarkers in people diagnosed with conditions after the newborn period. Dr. Howell noted that Denmark has more than 25 years of experience in the storage and use of residual DBSs, and these specimens have great research value. After thorough discussion, ACHDNC plans to make recommendations to the Secretary of HHS regarding policies for retaining residual blood spots and informed consent for stored samples.

IOM Report—Beyond the HIPAA Privacy Rule: Larry Gostin, J.D.

Mr. Gostin, Chair of the IOM Committee on Health Research and the Privacy of Health and one of the editors of the IOM report *Beyond the HIPAA Privacy Rule: Enhancing Privacy, Improving Health Through Research*, explained that the IOM Committee was charged with assessing whether the HIPAA Privacy Rule undermines or interferes with health research. He noted that the report is particularly timely as ARRA includes money for health information technology and also strengthens some of the provisions in the HIPAA Privacy Rule. He introduced three other IOM Committee members—Mr. Stanley Crosley,

Dr. Tom Croghan, and Mr. Andrew Nelson—who would join him after his presentation to help answer questions:

The IOM Committee concluded that the HIPAA Privacy Rule undermines important and valuable health research. In formulating its recommendations, the Committee considered equally compelling societal values—privacy and security of personal information and the value of research. Thus while patients have strong expectations that their personal information will be kept in a private and secure way, the public is less safe and less healthy without good quality research.

In its research, the Committee found that the HIPAA Privacy Rule increased the cost and time of research projects, complicated recruitment and increased selection bias, confused participants regarding their rights and protections, created barriers to the use of patient specimens, and failed to create an effective way to conduct studies with de-identified data. It also found that the Privacy Rule is inconsistent with other regulations such as the Common Rule, impedes research that is exclusively information-based, is inconsistently interpreted and implemented by covered entities, and creates challenges for multi-institutional research.

The IOM Committee therefore made recommendations that it believes will improve privacy and facilitate important and valuable research. Its "first and foremost" recommendation is that HHS should develop a new framework for protecting privacy in health research. This recommendation calls for Congress to authorize a new approach that enhances privacy protections through improved security, transparency, and accountability and ensures that privacy applies uniformly to all health research. The new framework would apply to any person, institution, or organization conducting health research in the United States, regardless of the source of data or funding; distinguish interventional research and research that is exclusively information based; and facilitate greater use of de-identified data in health research, and include legal sanctions for unauthorized reidentification. In addition, HHS should exempt health research from the HIPAA Privacy Rule.

Recognizing that not everyone would agree with its innovative strategy, the IOM Committee provide an alternative recommendation—HHS should revise the Privacy Rule and associated guidance.. These revisions include (1) reducing interpretive variability through revised and expanded guidance and harmonization (e.g., expand use and usability of data with direct identifiers removed to enhancing privacy in research), (2) developing guidance materials to facilitate more effective use of existing data and materials for research (e.g., clarify the circumstances under which DNA samples or sequences are considered protected health information), and (3) revising some provisions of the HIPAA Privacy Rule that currently hinder research but that do not provide meaningful privacy protections (e.g., reform requirements to account for disclosures of protected health information for research).

The Committee also recommended changes, independent of the Privacy Rule, that are necessary for either recommendation. These changes include: (1) safeguarding personal health information, (2) protecting members of IRBs and Privacy Boards who serve in good faith, (3) disseminating research results to study participants and the public, and (4) educating the public about how research is done and its value.

The aim of these recommendations is threefold: (1) to have strong privacy safeguards to ensure that institutions that hold data for research purposes are certified and are trustworthy, (2) for institutions to achieve consistent privacy practices that strictly govern who is authorized to receive information, and (3) to have detailed and careful security provisions.

Mr. Gostin noted that a paper will appear in *Journal of the American Medical Association* in April 2009 that summarizes the Committee's conclusions and adds additional observations. (See Gostin LO and

Nass S. (2009). Reforming the HIPAA privacy rule: safeguarding privacy and promoting research. *JAMA.* 301(13):1373-1375.)

Discussion

Ms. Au provided some insight regarding the newborn screening pilot project in California. Separate IRBs were supposed to be established at each site of the pilot project. However, there was not enough time or manpower to set up IRBs at every medical facility, so only 25 percent of the newborns could participate; the other 75 percent were born in institutions that they did not have an IRB. Dr. Howell commented on the difficulty of dealing with so many IRBs and also observed that in Massachusetts, it is really an informed dissent program.

Dr. Teutsch asked if ACHDNC had any plans to go back and look at conditions already recommended for the core panel in newborn screening, Dr. Howell said this idea had been discussed, but no decision had been made.

Dr. FitzGerald noted that there are several extensive databases within the Department of Veterans Affairs, Department of Defense, and Indian Health Service. Dr. Croghan spoke of the usefulness of linking the databases for health service researchers and said that such a process should be undertaken by an organization certified as having met various privacy and security standards. Certified organization could then make the database information available in a de-identified form or as a limited dataset, as appropriate.

Dr. FitzGerald then wondered about a standard for de-identification when it is now possible to identify an individual from as few as 70 SNPs. Mr. Crosley indicated that the approach being recommended by the IOM committee is to set up a model to prevent harm rather than trying to pursue an elusive concept of continually updating the de-identification criteria. He also pointed out that there are European data protection approaches as well as those in the United States. The European Union has an organizing body around data protection called the Article 29 Working Group.

Dr. Carome inquired whether the IOM committee had identified any particular aspects of the Common Rule as being problematic. Mr. Nelson replied that it is more a matter of trying to harmonize the Common Rule, which is an HHS-wide rule, with the HIPAA Privacy Rule. Trying to harmonize the Rules sometimes confuses IRBs, which can lead to conservative decisions such as less willingness to approve multisite studies. However, if one looks only at the Common Rule, it is not specific enough and does not go far enough in its privacy protections. Mr. Nelson also commented that the IOM report has very specific security recommendations and there may be overreliance on the Common Rule for how IRBs should advise on whether the consent form is sufficient to apply to some secondary research uses. Also, the Common Rule only covers research funded by the Federal Government.

Ms. McAndrews, from OCR, thanked the IOM Committee for its efforts to balance the need for data and the need for privacy and confidentiality when individuals share their data in the context of treatment. She stated that OCR does not want fear of secondary research uses to interfere with patients' ability to get care in the first place. Ms. McAndrews referred to substantial realignments that took place in 2002 to the accounting for disclosures provisions and to simplify waiving the authorization requirements for access to information. She asked to what extent the IOM report and recommendations in those two areas took into account the steps that were made in 2002 and on the practices and problems that may have continued to reside in those two areas.

Dr. Croghan replied that it is the opinion of the IOM Committee that privacy and health research are both private and public goods, and that neither one occurs adequately without the other one. The IOM

committee had heard from OCR and was aware of the 2002 changes. It had also heard from the research community that, despite the 2002 realignments, some barriers still existed. For example, physicians who did not understand the Rules simply would not release data that had been requested by a research project.

Dr. FitzGerald remarked that much of the data we have today, with the possible exception of newborn screening data, is not representative of our population. Groups that have been marginalized up to this point may have good reasons for suspicion of benefits coming from any major research projects and overrepresentation in research programs will be needed to obtain the right balance. He asked how this challenge might be addressed. Dr. Croghan reported findings from public polls showing that some groups with particular health issues, for example, individuals with AIDS or mental health problems, were actually the most willing to endorse releasing data and participating in research. These individuals are also the most likely to gain from participating in research.

Noting the capacity for multisite studies, Mr. Nelson observed that some supportive guidance from HHS is needed to help locally based organizations understand and give permission to contribute to the societal good. Dr. Croghan added that scientists also need to make their research more understandable to the public. This effort includes providing education about the research process and research results.

Dr. Randhawa asked to what extent the IOM Committee considered different models of data aggregation—centralized, de-identified aggregate databases or small federated databases where data are identified and controlled locally but allow for queries specific to a research question or project. Mr. Nelson commented that there is an increasing capacity for small, federated databases to be able to transfer aggregated, de-identified results to outside researchers. This capability is encouraging in terms of protection and safety issues; however, not all organizations have the capacity to map and configure data in an aggregated, de-identified way.

Mr. Crosley added that the IOM report also considers having a certification agent, modeled on the Ontario privacy law that has qualified entities who can hold reidentification keys. It would certainly be possible to have that encryption key exist at the data level. It also would be possible to have a federated query authority as a trusted agent or an authentication agent that could then do the same thing.

Dr. Telfair remarked that he had some contingency questions. How would one go back and obtain reconsent in a longitudinal study from someone who was picked up through newborn screening, keeping mind that the public and vulnerable populations are involved. Dr. Croghan responded first with a simpler case in which an adult needs to be reconsented. In contrast to the HIPAA Privacy Rule, under the Common Rule, subjects may consent to future research. The IOM committee did not address the more complicated case of getting the consent of children and/or guardians. This issue would be an important one for SACGHS and others to discuss.

Dr. Howell spoke about recent NIH funding for a major newborn screening translational research network. This network would identify children with rare disorders, follow them in a systematic way, and provide prospective data on these conditions. The data may be retained locally but would require the help of an infrastructure like NCI's cancer biomedical informatics grid (caBIG®). Noting that parents consent for children and adolescents, Dr. Telfair asked about those who consent for vulnerable adults. The person who signed consent on behalf of a vulnerable adult may not be available 20 years later to reconsent.

Dr. Croghan said that the IOM committee drew a distinction between interventional research that would always require consent and informational research that would not necessarily require consent if certain controls are in place, for example, IRB oversight or other appropriate security measures.

Dr. Dale noted that the HIPAA Privacy Rule is a national rule, but IRBs are locally controlled. He asked whether the IOM Committee took a position on national IRBs, particularly related to rare diseases. Mr. Nelson responded that while the IOM committee did not present anything on national, multisite IRBs, it did stress the need for better harmonization of rules to help the local IRBs. Mr. Crosley added that the Committee also recommended that IRBs be given some layer of indemnification protection and liability protection. If HHS will sanction some best practices, these can also provide guidance to the local IRBs.

Dr. FitzGerald then asked the Committee members and panel if there are additional charges that SACGHS should consider. Noting that genetics is not mentioned in the HIPAA Privacy Rule, Dr. Croghan commented on the need to consider integration of genetic information, how those data are maintained, and how they are integrated with other protective health information and made available to the research community. Dr. Howell proposed looking at mechanisms of informed consent and having one central IRB for multisite studies.

Mr. Nelson said that it will be important to involve the public to a much greater extent, doing so through transparent, clear communication, and engagement, thereby ensuring a community-supported approach to privacy. Mr. Crosley remarked on the great value of having privacy advocates, patient advocates, people with chronic illness, and public and private researchers on the IOM committee.

Committee Discussion of Issues and Next Steps for Issues Related to Informed Consent on Genomic Data Sharing

Dr. FitzGerald invited everyone to contribute ideas for both short- and long-term next steps. He noted that SACGHS has addressed public engagement in its reports on large population studies, pharmacogenomics, and the oversight of genetic testing. He suggested that the question of how informed consent will be reconceptualized, redescribed, and redefined might be an area to explore. He asked if there is a need to obtain more information on a specific area and whether the Committee should form a task group to focus on specific issues. Dr. FitzGerald also mentioned that with his term on SACGHS ending, Dr. Charmaine Royal of Duke University will take his place as the lead in this topic area.

Dr. Dale asked if the full IOM report is available, and Dr. FitzGerald replied that it is. (See http://www.iom.edu/CMS/3740/43729/61796.aspx.)

Dr. Billings suggested that it would be helpful to know what IOM is planning for its next steps, and Dr. Teutsch wondered what other federal entities are doing. Ms. McAndrews reported that IOM presented the same report to the Secretary's Advisory Committee on Human Research Protections (SACHRP) last week. SACHRP has made recommendations in the past on privacy and the intersection of the HIPAA Privacy Rule and research. Ms. McAndrews anticipates that SACHRP will look again at its prior recommendations in light of this new report and will develop additional recommendations to the Secretary.

OCR's regulatory agenda and requirements stemming from the HITECH Act and GINA are likely to keep that Office busy for the next 12 to 18 months. Nothing in the HITECH Act addresses research; however, a study of de-identification of data is a mandated area. Broadening disclosures is a more likely outcome than the IOM proposal to remove disclosures from accounting. OCR may also be able to look at authorizations.

Dr. Carome spoke further about the March 2009 SACHRP meeting. He reported that SACHRP made a series of recommendations about the Privacy Rule several years ago that are still undergoing deliberation and consideration by HHS. Those recommendations align well with the general framework of the recommendations made by IOM. They tend to reinforce one another in terms of the concerns and issues

that have been raised. Dr. Carome also said that all of SACHRP's recommendations to date are directed at the Privacy Rule and, with input and consultation with others at HHS, would require action by OCR.

Regarding the concerns of the IOM committee about the Common Rule, Dr. Carome stated that it is still unclear to him what problems the Common Rule would pose to research if there was no Privacy Rule and the Common Rule covered all research. He believes that the current regulations offer a lot of flexibility. When a research activity is not covered by the regulations, it is either because it does not involve human subjects or it is exempt. For research that is not exempt and is covered under the Common Rule, there are procedures that have always existed for waiving informed consent.

Regarding the provisions on privacy, Dr. Carome stated that when an IRB reviews and approves research, it must ensure that there are appropriate provisions to protect the privacy of the data. That simple provision gives IRBs and investigators sufficient discretion to design appropriate privacy protections. Stronger protection, controls, and restrictions for data release, along the lines that the IOM Committee has discussed, can be accommodated within the framework of current regulations.

Dr. Randhawa noted that SACGHS has not heard about from the health information technology community, and it may be helpful to engage with the successor of the American Health Information Community (AHIC).

Dr. Teutsch suggested that SACGHS might want to consider specific issues such as the use of genetic information and privacy concerns, as well as some data-sharing issues with the electronic medical records. He asked whether SACGHS should continue to monitor broad issues or form a small workgroup to consider specific concerns.

Dr. Telfair suggested examining whether there is discrimination in working with vulnerable populations. Dr. Billings proposed looking at what can be done regarding newborn screening residual DBS specimens. He pointed out that it might be possible to sequence the whole genome from the blood sample. If so, what would opt-in/opt-out rules look like? How would DBS specimens, which represent a nonbiased population, be used? SACGHS may have a useful role in addressing these issues.

Dr. FitzGerald suggested checking into whether some of these questions might be addressed through the NIH grant for translational work in newborn screening. He added that depending how on vulnerability is defined or delineated, it might be an important issue to examine. Dr. Williams mentioned that with the recent establishment of a Secretary's Advisory Committee on Health Information Technology, there are now at least four HHS Secretary's committees that have relevant charges. He suggested establishing a liaison group to coordinate and prevent duplicative efforts.

Dr. FitzGerald agreed that coordination is key. Dr. Teutsch said that vulnerable populations fit under a population health component and that privacy, research, and consent for those populations would be a discrete subset. Dr. Billings pointed out that the Common Rule has provisions for vulnerable populations as well. Dr. Williams reminded everyone that engaging the public is still an important SACGHS topic. Dr. Teutsch then invited Dr. FitzGerald to work with Dr. Royal and a few others to develop concrete proposals to bring to the June 2009 SACGHS meeting.

Noting first that he is an active researcher, Dr. Dale commented that the IOM Committee report is worth looking at and discussing more thoroughly. Dr. Teutsch proposed that everyone take time outside the meetings to read the report and consider how it fits with the SACGHS' work.

Session on the Genetics Education and Training Task Force

Update on Education and Training Task Force Data Gathering: Barbara Burns McGrath, R.N., Ph.D.

Dr. McGrath stated that goal of the session was to provide an update on the activities of the Genetics Education and Training Task Force, including preliminary results from the data-gathering efforts of the Task Force's three workgroups. Following the update, she hoped Committee members would provide input about the direction of the Task Force, which Dr. McGrath estimated was halfway through its work.

Dr. McGrath provided background for new SACGHS members, noting that the topic of genetics education and training has been a high priority since the inception of the Committee. In 2004, SACGHS held a roundtable on this topic and subsequently developed a resolution and series of recommendations for the Secretary of HHS. The current Task Force was formed in 2007 following a second roundtable highlighting that genetics education and training had not kept pace with emerging genetic technologies.

In determining the scope of the Task Force and considering who might benefit from greater genetics education and training, SACGHS considered K-12 education and groups such as laboratorians, hospital administrators, and speech pathologists who also have genetics education needs that not been adequately addressed. Guided by the principle of point-of-care, the Task Force decided to focus on health care professionals, public health providers, and patients and consumers, including the general public. The Task Force then established three workgroups with each concentrating on one of these areas. Aiming for a report with measurable and actionable recommendations to the Secretary of HHS, the Task Force plans to examine existing education tools and current education needs, as well as anticipating future needs. Each workgroup is reviewing the literature to help inform the genetics education gaps and needs and also developed survey instruments to gather data.

Health Care Professionals. The health care professional workgroup, led by Dr. Feero, is using two surveys to assess the training needs of health professionals. One survey targeted health professional organizations, and the other queried federal agencies about activities related to genetics education and training. The outcomes of these surveys can be compared to similar surveys conducted in 2004. The survey of health professional organizations was sent to 57 professional organizations, including 28 general professional organizations (e.g., American Medical Association and American Academy of Family Physicians), eight genetics-specific organizations, 18 organizations devoted to professional education, and three federal advisory committees. Currently, the overall return rate is 58 percent. The genetics-specific organization response rate was 100 percent, general professional organizations had a fair response, while the response rate for those organizations devoted to professional educational was low.

Preliminary data indicate that half of the organizations have an established committee, workgroup, or staff that deals with genetics or genomics topics. Barriers reported to providing genetics education include competing priorities, genetics is not emphasized on certifying exams, genetics is not applicable to the organization's mission, lack of accessible educational resources, lack of leadership knowledge, and lack of evidence to support clinical effectiveness of care based on genetic/genomic information. One genetic-specific organization reported no barriers. Dr. McGrath speculated that if increased clinical utility were demonstrated for genetics and genetics testing, then it would rise as a priority.

The workgroup sent surveys to 18 SACGHS *ex officio* agencies in its survey of federal activities, and 15 surveys have been returned. Of the 15, seven agencies reported no genetics education and training activities. Among the eight agencies reporting such activities, five can be compared to 2004 survey results. Data analyses are underway. The workgroup will also examine other federal reports on genetics education and training and articulate personalized medicine initiatives.

Public Health Providers. The public health provider workgroup, led by Dr. Telfair, has similar goals but an approach that begins with development of competencies. The workgroup gathered genetics and genomics competencies from various public health organizations and synthesized them into a concrete set of 12 core competencies. Examples of these competencies are maintaining up-to-date scientific knowledge of genetics and genomics; identifying opportunities to integrate genetics and genomics into public health; identifying political, ethical, legal, and societal issues associated with integrating genetics and genomics into public health; and implementing research findings into policies or practices.

Based on the 12 core competencies, a survey will be developed, administered, and analyzed to determine whether appropriate personnel are achieving the competencies, where they receive genetic/genomic education, and the gaps and needs in genetic and genomic education and training.

Consumers/patients. The consumer and patient workgroup, led by Mr. Vince Bonham is addressing the genetic education needs of consumers and patients. The workgroup's approach is to conduct five paired semi-structured qualitative interviews with professionals in the following areas: health communications, advocacy and outreach, clinical genetics, industry, and education. The workgroup will then develop a survey targeted at consumer advocacy and community-based organizations that will use the themes identified from these interviews.

While the interview data are just beginning to be analyzed, one can note that consumers get education from health care professionals and the media and expect the government to provide guidance. The professionals interviewed see consumers as needing a greater understanding of multiple risk factors and how genetics and the environment interact with these risk factors. Other needs include discerning the level of expertise among health care providers regarding genetics and genomics and a need for tools to help consumers evaluate the veracity of genetic information.

Several barriers to genetic literacy among consumers were identified by the interviewed experts and advocacy groups including poor health literacy in general, a notion of genetic determinism or fatalism (e.g., why learn about genetics when there is nothing one can do about it), and fear of discrimination continuing past the GINA era. Dr. McGrath reported that the Task Force is struggling with how to reach members of the general public, individuals not specifically interested in genetics and genomics, to assess their attitudes.

Dr. McGrath concluded her remarks by stating that she expects to present preliminary draft recommendations for discussion at the June SACGHS meeting and to complete a draft report for discussion at the October meeting.

Committee Discussion

Committee members complimented the Genetics Education and Training Task Force on its progress.

Closing Remarks

Dr. Teutsch briefly reviewed the accomplishments of the day—several updates on federal activities, formation of a task force to develop a paper on DTC genetic testing, a discussion of the challenges of data privacy and informed consent, and the progress of the Genetics Education and Training Task Force—and he previewed the following day's activities that include additional federal agency updates and discussion of the implications of genetics and health reform from the payers' perspective.

March 13, 2009

Opening Remarks

Dr. Teutsch thanked departing members Drs. FitzGerald and Telfair for their valuable contributions as SACGHS members, presented each with a certificate, and expressed hope that they could be called on the future to provide ad hoc assistance. He also noted that the SACGHS progress report will be delivered to the new HHS Secretary when he or she is appointed and confirmed.

Updates from SACGHS *Ex Officios*

FDA. Dr. Alberto Gutierrez, Deputy Director of the Office of In Vitro Diagnostic Devices, Evaluation, and Safety (OIVD) in the Center for Devices and Radiological Health, first reviewed FDA's two-fold mission, which is to protect and promote public health while ensuring that food, drugs (icluding veterinary drugs), devices, and biologics,on the market continue to be safe and effective.

Recently, FDA has moved to strengthen the role of genomics within the Agency. Dr. Frank Torti, the Acting Commissioner, recently created a new position of Senior Genomics Adviser in the Office of the Chief Scientist. Currently, Dr. Elizabeth Mansfield, who is attending today's meeting, has been detailed from OIVD to fill this position.

FDA's genomic programs fall into three areas: (1) the National Center for Toxicological Research, which is responsible for standardizing microarray data analysis; (2) the Office of Clinical Pharmacology (OCP), which has a biomarker qualification program and helps drug companies use genomic data in drug development; and (3) OIVD, which has a staff for personalizd medicine that will coordinate both outreach and internal issues related to personalized medicine and genomics.

Dr. Gutierrez noted that the In Vitro Diagnostic Multivariate Index Assays (IVDMIA) guidance has not yet been finalized; FDA is waiting for the new administration to continue the clearance process. He also mentioned that two panels were convened in December 2008 to consider topics relevant to genetic testing. The Immunology Devices Panel considered the human epididymis protein 4 (HE4) test when used in combination with the CA125 test and the mathematical algorithm called the Risk of Ovarian Malignancy Algorithm (ROMA^{TM}) to estimate the risk of epithelial ovarian cancer in pre- and postmenopausal women with an adnexal mass. The Oncologic Drugs Advisory Committee discussed the type and amount of data needed to support product labeling using predictive biomarkers such as KRAS mutations that can inform response to panitumumab therapy for colon cancer.

Other OIVD activities include taking enforcement action when needed. For example, in September 2008, FDA sent a warning letter to LabCorp stating that the OvaSure test was not within the scope of laboratory-developed tests as OvaSure was designed, developed, and validated by investigators at Yale University but not at LabCorp.. In addition, OIVD—in collaboration with NCI and CDC—continues to work on critical path programs related to genetics.

OIVD is also involved with the Cancer Biomarker Consortium that addresses issues related to biorepositories, bioinformatics, bioassay validation, and data sharing. It is also part of an interagency task force with NCI that focuses on oncology, particularly molecular diagnostics, biospecimens, and pharmacogenomics

In concluding his remarks, Dr. Gutierrez noted that in response to a SACGHS recommendation and a petition filed by Genentech that requested FDA to regulate all laboratory tests, OIVD is putting together background and options for the new administration to consider.

HRSA. Dr. Denise Geolot explained that HRSA aims to provide quality health care to people who are underserved, uninsured, isolated, and medically vulnerable. HRSA funding supports 7,000 clinics that provide primary preventive health care services in every single state and almost every community in this country, serving more than 16 million low-income people, Through its Ryan White Program, HRSA also provides care and medications for 530,000 individuals with HIV/AIDS.

HRSA administers a range of programs for women and infants in need and children with special health care requirements. The Maternal and Child Health Bureau includes a specific focus on genetics, which includes the Heritable Disorders Program. This Program supports regional genetics and newborn screening service collaboratives. The regional centers have as their primary goal ensuring that children with heritable disorders and their families have access to quality care and appropriate genetic expertise and information. A national coordinating center was established to work with regional centers and other partners to identify and address issues regarding access to and the utilization of genetic services at the national, state, and community levels. In addition, there are two new genetics-related projects: Screening for Heritable Disorders in Children: The Efficacy from a Consumer Perspective, and Ensuring Access to Quality Information and Education in Genetics. HRSA has also funded a family history project, which provides a downloadable, customizable brochure that communities and specific genetic disease groups can use.

Another HRSA activity is to support clinician recruitment and services for underserved areas. The Agency strives to ensure a health care work force that is diverse, well trained, and adequately distributed about the nation. Through the National Health Service Corps, scholarships and student loans have supported more than 28,000 clinicians who have served over the past 35 years in some of the most economically deprived and geographically isolated communities in America.

HRSA also supports health professional workforce development and safeguards the foundation of the U.S. health care system by targeting grants to academic institutions to support post-graduate faculty retention, administering scholarships to increase staff in critical specialties such as nursing, and funding leadership development programs. About 10,000 clinicians benefit from these programs annually. Two reports of interest to SACGHS are *Genetic Counselor Workforce Training Program: Professional Practice: The Issues Affecting Supply and Demand* by Judith Cooksey and *Clinical Laboratory Workforce: The Changing Picture of Supply and Demand, Education, and Practice*. In addition, HRSA has a rural health officer program to provide increased accessibility.

Dr. Geolot mentioned that HRSA has a Rural Health Office, which makes health care accessible for more than 60 million residents of rural America. She concluded her remarks by noting that Dr. Mary Wakefield, who was appointed as HRSA Administrator, is an expert on rural health, and also has expertise in patient safety, Medicare payment policy, workforce issues, and public policy.

CDC. Dr. Kathryn Kolor, Policy Officer in the Office of Public Health Genomics (OPHG), described the CDC mission as a collaborative effort to create the expertise, information, and tools that people and communities need to protect their health through health promotion; prevention of disease, injury, and disability; and preparedness for new health threats. Genomics is a cross-cutting discipline at CDC, focused on the effective and responsible application of genomics knowledge and tools to promote population health, with applications that span chronic disease, environmental health, occupational health, infectious disease, and other areas.

Of particular relevance to SACGHS are activities at OPHG and the Division of Laboratory Systems. OPHG works to accelerate and streamline the effective integration of validated genomic knowledge and tools into the practice of medicine and public health. For example, the EGAPP Working Group published three new evidence-based recommendation statements that assess the validity and utility of three cancer genetic-testing applications. In regard to genomics translation research, CDC recently awarded more than $1.5 million per year for three years to fund five projects to conduct genomics translation research, education surveillance, and policy interventions to help move evidence-based genomics applications into practice. Additionally, CDC and NIH are working together to launch a network of research programs called the Genomics Applications in Practice and Prevention Network (GAPPNet). GAPPNet's inaugural meeting is planned for October 2009.

Other relevant activities include a CDC-funded clinical trial that examined whether family history risk assessment and personal prevention messages influence health behavior and the use of medical services. The first report based on this trial assessed risk beliefs across chronic diseases based on family history information and was published in February 2009 in *Preventive Medicine*. In November 2008, based on the National Health and Nutrition Examination Survey, CDC published the first prevalence estimates of 90 genetic variants for a nationally representative sample of the U.S. population that includes major racial and ethnic groups. These prevalence estimates form the foundation for CDC's new Comprehensive Databank of Human Genetic Variation in the United States. The databank will assist investigations of the roles genes play in population-level risk for a disease and how genetic variants might contribute to health disparities. In addition to co-sponsoring the December 2008 NIH-CDC workshop on personal genomics that Dr. Feero mentioned yesterday, CDC has conducted consumer and health care provider surveys on awareness and use of personal genome scans, and analyses are underway.

The Division of Laboratory Systems developed a report on good laboratory practices for molecular genetic testing for heritable diseases and conditions, which will be published in the *Morbidity and Mortality Weekly* (MMWR) *Recommendations and Reports* this spring. This report will serve as a guide to improve the quality of health care outcomes of molecular genetic testing for heritable diseases and conditions and to enhance the oversight and quality assurance practices for molecular genetic testing under the CLIA regulatory framework. A second project is underway that focuses on good laboratory practices for biochemical genetic testing. In addition, CDC is funding and working with the Rand Corporation to improve the framework for reporting molecular genetic test results from laboratories to clinical settings to promote the understanding of relevant genetics and the appropriate use of genetic tests for patient care. The Division of Laboratory Systems also sponsors and supports the Genetic Testing Reference Materials Coordination Program. This Program fosters coordination among the broader laboratory community to facilitate the development and characterization of publicly available genomic DNA samples and cell lines that can be used by the research and clinical laboratory community for test development, validation, proficiency testing, and quality assurance.

Regarding ARRA, CDC is collaborating with other HHS agencies to address the provisions of the Act, in particular the areas of prevention and wellness, health information technology, and comparative effectiveness.

Office for Human Research Protections (OHRP). Dr. Michael Carome, Associate Director for Regulatory Affairs in OHRP, also represents the Office of Public Health and Science (OPHS). Both Offices are within the within the Office of the Secretary, and the current Acting Assistant Secretary for Health is Dr. Stephen Gossen, who is also the Acting Surgeon General. Dr. Gossen's priorities for the Office of the Surgeon General include disease prevention, eliminating health disparities, increasing public health preparedness, and improving health literacy. (See http://www.surgeongeneral.gov/priorities/index.html for details of these public health priorities.)

OPHS activities that may be of interest to the Committee include the January release of an updated Surgeon General's Internet-based Family Health History Tool. The newer version of this took can be used in electronic health records and personal health records, and it can be easily shared with relatives and physicians because of those mechanisms. The uploaded information, for those who have privacy concerns, is not retained by the government. OPHS is also in the process of developing HHS's Healthy People 2020 objectives (See http://www.healthypeople.gov/hp2020/ for additional information.)

Turning to OHRP activities, Dr. Carome explained that OHRP has a lead role in promoting the protection, safety, and welfare of human subjects who participate in research conducted and supported by HHS. For example, IRBs that review and approve HHS-funded research have to register with OHRP. This Office also has education and training programs, a compliance oversight program, and staff who develop policy and provide guidance to the human research community. Additionally, OHRP developed a guidance document on GINA that describes important implications for IRBs and investigators to consider when doing genetic research and the types of protections that are provided by that Act. The guidance is going through final HHS clearance.

In concluding his remarks, Dr. Carome mentioned that OHRP published and sought public comment on a draft document, *Guidance on Important Considerations for When Participation of Human Subjects in Research is Discontinued*. A companion document was developed by the FDA regarding issues related to data retention when subjects withdraw from research and what investigators can continue to do with data they have collected up to that point. The OHRP document talks about parallel issues, in particular, issues related to what investigators can do with tissue samples that have been obtained but a subject chooses to withdraw his/her consent for that research. Public comments to this document are being reviewed, and the final guidance is expected in a few months.

Department of Defense (**DOD).** Lt. Col. Daniel Wattendorf, of the Office of the Air Force Surgeon General, noted that he was representing the Office of the Assistant Secretary of Defense for Health Affairs. DOD has a dual health care mission—to ensure combat readiness and to provide health care benefits for about 9 million individuals through a large and complex health care system that utilizes the CMS structure for reimbursement. Preventive care is of particular importance for the mission of ensuring combat readiness; however, the funding stream depends on treatment codes as in the civilian arena, which do not always align with preventive care.

DOD is actively engaged in changes to personalized health care and electronic health records. DOD has representatives on the Healthcare Information Technology Standards Panel (HITSP) and HL-7 and closely follows ONC activities, particularly in areas where it is examining aggregating research in federated systems.

DOD also supports research related to genetics. The largest amount of breast cancer research money in the world is handled by Medical Research and Materiel Command, which is a Congressionally directed research program. It is a peer-reviewed, NIH-style of research and includes other research areas such as prostate cancer, neurofibromatosis, autism, tuberous sclerosis, and the genetics of food allergies.

Other DOD programs relevant to SACGHS are the Armed Forces Institute of Regenerative Medicine and the Clinical and Rehabilitative Medicine Research Program, both of which started in the past two years. They are engaged in stem cell research and regenerative medicine using genetic reprogramming of cells.

Newborn genetic screening is of particular interest since 1 in 80 births in the United States comes under DOD health care auspices. The Department is actively involved with the ACHDNC and is looking into aggregating the HHS newborn screening data with a national registry being developed for DOD

beneficiaries. DOD is also developing a comprehensive newborn screening program so all infants born within the DOD health care system, worldwide, will receive the same newborn screen.

Department of Commerce (DOC). Dr. Michael Amos, Scientific Advisor, Chemical Science and Technology Laboratory, National Institute of Standards and Technology (NIST) in the Department of Commerce, informed attendees that ARRA provided DOC with $7.9 billion, including $610 million to for NIST. The NIST stimulus funds are budgeted for the following programs: $360 million for construction; $180 million to provide competitive construction grants for science facilities around the United States; and $10 million for an interoperable smart grid. In addition, NIST received $20 million transferred from HHS for developing test beds for health information technology infrastructure and $220 million for grants, fellowships, equipment, and supplies. The spending plan is pending until the DOC Secretary is confirmed.

In 2008, NIST had about $15 million in diagnostic spending and about $21 million for health care. Of that, only about $5 million was ever appropriated by Congress specifically for health care-related activities. The rest of it has been reprogrammed from other NIST activities based on decisions by laboratory directors. Under the 2009 omnibus appropriations bill, NIST will receive an additional $3 million this year to work on current-generation diagnostic measurements. This funding will focus on laboratory medicine and medical imaging, in order to improve the information that goes into the electronic health record. By 2011 Dr. Amos hopes that NIST will be ready to tackle some of the challenges that he mentioned at the SACGHS meeting in December 2008. The immediate laboratory medicine focus will be nucleic acids and proteins, and the focus in medical imaging will be on MRI, PET, CT, and medical optical imaging in the short term, with expansion into molecular imaging.

Roundtable on Genetics and the Future of the Health Care System

Dr. Teutsch commented that this roundtable, as well as another one at the next meeting, would focus on one of the Committee's new priority topics—genetics and the future of the health care system. Through these roundtables, the Committee would provide a comprehensive assessment of the role that genetics can play in a new approach to health care. This work fits in with the new administration's interest in reforming health care.

Roundtable Purpose and Overview: Mara Aspinall, M.B.A.

Ms. Aspinall, who led the session, elaborated that the practical objective of the session was to identify what needs to be done today to prepare for a future where genetics and genomics play a larger role. As background for the session, Ms. Aspinall imagined potential innovations in the future and their implications for clinical care. She suggested that $1,000 full genome sequencing would soon be available, and its advent would require better education of both physicians and patients to understand genomics and genetic testing. Ms. Aspinall also speculated on having a laboratory-on-a-chip that could be on or embedded in one's arm, or maybe just carried in a wallet.

Ms. Aspinall then said that the next speaker would provide an overview.

Overview of Key Issues in Health Care Reform: Robert Epstein, M.D.

Dr. Robert Epstein, an epidemiologist and Senior Vice President and Chief Medical Officer at Medco Health Systems, commented that in the Medco systems covering 60 million people there are now tens of thousands who have obtained some genotype information and want to be able to apply it to their decisionmaking about drug treatments. He also noted that it can take 17 years for basic research

discoveries to be translated into bedside practices. One reason for this delay is that health care providers become overwhelmed by the enormous amount of new information available and simply overlook many journal articles. Another reason for the delay in clinical uptake of new practices is that the process for gathering evidence for a practice—a prerequisite often to its adoption—is slow-moving. Although randomized trials are seen as the gold standard for establishing evidence supporting a practice, they take a long time. Furthermore, observational studies are often more than adequate—as demonstrated by observational studies that linked lung cancer and smoking. Dr. Epstein wondered if it is really best to use randomized control trials to assess the clinical utility of genetic tests.

Dr. Epstein observed that going forward the payer community will need a framework for providing access to genetics and genomics technologies. In thinking of how that framework for making coverage decisions should operate, one naturally must consider the controversial issue of how return on investment should be evaluated by payers. How does one assess the value of a technology or procedure? Does one rely on cost-benefit analysis? Or is the more appropriate system one that looks at cost per life-years saved? Dr. Epstein noted that these are difficult questions that need to be resolved for the field of genetics to move ahead faster.

Dr. Epstein added that another challenge in the current health care system is that information exists in various silos and that any reformed system must provide for the integration of this information. Integration of this data will require standardization and appropriate privacy safeguards. Once the information is integrated, there will still be a need for software that can analyze it and provide alerts and guidance to health care providers.

Dr. Epstein concluded by observing that the pharmacy world has been electronically linked since 1990—now it is time to begin to integrate genomic information into this established electronic infrastructure.

Questions and Answers

Dr. Wise commented that the cost to implement something is different under a capitated system versus fee-for-service system. In the integrated system in which he works when costs are tracked, it turns out that sometimes the hospital is "taking the hit" when a new test is introduced. Each stakeholder can have a different amount at stake when change is taking place. Dr. Epstein agreed that there is a patchwork of decisionmakers and stressed that transparency could help by showing how each stakeholder is involved.

Dr. Evans next commented that he was glad Dr. Epstein had brought up the question of how one determines when something is ready for clinical practice. He agreed that a randomized clinical trial cannot always be afforded but would not want to be misled by inferior evidence, as happened with hormone replacement therapy. Good outcome data are essential. In response, Dr. Epstein reiterated that Bradford-Hill criteria provide an example of how one can do an observational study in a rigorous way that generates confidence in its results. Dr. Evans then suggested that provisional coverage would be appropriate for a practice shown to be effective through something such as the Bradford-Hill criteria. Dr. Epstein agreed.

Dr. Billings returned to the topic of return on investment, noting that in the present economic climate, there is pressure for near-term returns on investments, while in biotechnology, good returns are usually 5 to 10 years away. Consequently, are there any genetic technologies that could generate near-term cost-reductions? Dr. Epstein prefaced his response by observing that different payers and employers have different expectations for when they must see a return on their investment—some must predict a return within a year to pay for a technology while others, such as those with long-term employees, are willing to pay for services that will not yield benefits until some years later. In genetics, payers generally are willing to pay for pharmacogenomic testing because models predict it will lead to a favorable return on

investment in 1 to 3 years. On the other hand, payers generally are not willing to pay for disease-predisposition testing.

Ms. Aspinall asked about Dr. Epstein's remark that pharmacies are getting only 25 percent of physicians to change a drug after a particular drug-alert message. She wanted to know why compliance was not 100 percent given that pharmacies are capable of effectively disseminating this information. Dr. Epstein clarified that there are often valid reasons for why the physician will choose to overrule this drug alert—the provider may know, for example, that the patient has done well on the current drug. Because the physician will always know relevant patient information that is absent from the database, 100 percent compliance should not be expected.

Dr. Licinio wondered what the biggest barriers are to bringing genetic testing into practice. He noted that most people who take drugs that are metabolized by a particular gene have not had testing for the presence of variants in that gene. Dr. Epstein reported briefly on a study soon to be published in which Medco and the American Medical Association collaborated to do a national survey of physicians to find out their attitudes and awareness to pharmacogenomics information.
While the survey showed that nearly 90 percent of physicians believe that genes do provide information about drug response, nearly 90 percent also said that they did not remember having genetic training in medical school, did not feel comfortable ordering the test, and did not understand how to interpret it.

Ms. Darien then asked Dr. Epstein how he reconciled a push for generalizable guidelines with the recognition that generalizations cannot be made about patients. Dr. Epstein suggested that more personalized guidelines are needed —guidelines tailored to particular subsets.

Public Health Payer Perspective: Barry Straube, M.D. and Bruce Quinn, M.D., Ph.D.

Dr. Straube. Dr. Straube, Chief Medical Officer for CMS, stated that he and Dr. Quinn were asked to address two principal questions: (1) how can the value of emerging genetic and genomic technologies best be evaluated in a timely manner for coverage determinations and (2) what changes in coverage and reimbursement determination will be necessary to address the increasing trend in prevention-based medicine?

Overview of coverage process. CMS may have preliminary meetings with technology developers, people in industry, and advocacy groups for patients or other entities to discuss what needs to go into a proposal for an NCD at CMS. These meetings can include a discussion of the type of data points that CMS needs for coverage determinations in the research protocols that are being developed. If a benefit category request is made to the Agency, staff make a decision as to whether CMS, in this case for Medicare, can cover the potential device Some situations are specifically excluded by statute.

If a formal request is made to CMS, the requester must have sufficient data from testing and literature searches to warrant decisionmaking. Routine NCDs undergo six months of review (including determinations of what is reasonable and necessary), leading to the posting of a decision memorandum. It may be necessary to seek outside technological assessment. After 30 days for public comment on the memorandum, CMS has another 60 days to finalize its decision and provide implementation instructions.

Recently CMS added the option of approving coverage with certain conditions. For example, perhaps only special centers can handle a particular technology. The Agency can also grant approval on the condition that a small amount of additional evidence is obtained and provided.

Making Timely Coverage Decisions on Genomic Technologies. One way to make decisions more timely would be through coordination of research, including providing earlier guidance to technology developers

on what their research must demonstrate for the technology to win coverage. Saving time by shortening the NCD process does not appear to be a viable option. There is not much opportunity to cut the six-month staff review period, and no one wants to cut the 30-day comment period for the public.

Another possibility to shorten the review process is to use LCDs instead of national ones. However, as was discussed yesterday, there are many who prefer one NCD to differing local ones.

As comparative effectiveness and cost effectiveness increasingly become factors in the review process, looking at these earlier would also help. Another possibility is lowering the standards for evidence. CMS could also look at how to align public and private sector coverage decisionmaking processes so that technology developers have a clearer idea of what evidence is needed.

Changes in coverage that will be necessary to address the increasing trend in prevention-based medicine. Dr. Straube proposed that the first topic to look at in considering changes in coverage is how to define a screening test versus a diagnostic test—often the distinction is unclear and in some instances a diagnostic test can be a screening test. To cover screening tests, CMS might need a statutory change as currently Medicare excludes them (with a few exceptions). Some flexibility is possible through Section 101 of MIPPA (as Dr. Straube mentioned yesterday), which allows CMS to make preventive services coverage decisions, including using cost-effectiveness analysis in those determinations.

Alternatively, using coverage with evidence development (CED) more liberally opens up the possibility of covering some genetic testing or genetic interventions sooner—but with the requisite that additional information be developed.

Dr. Straube also noted that CMS can review, update, and revise as necessary the National Coverage Decision Manual, the definitions, and the guidance documents. Given yesterday's example of the antiquated language on cytogenetics, the revision of materials is certainly needed. Genomics could be a pilot case for achieving better local-national coordination, and CMS could aim for earlier collaboration too with FDA on parallel reviews.

Dr. Quinn. Dr. Quinn, a physician, researcher, and policy expert who is the Senior Health Policy Specialist for Foley Hoag and the former contract medical director for the California Medicare Part B Program, observed that from the perspective of a local medical director, covering diagnostic tests but not screening tests can be murky and illogical in practice. An example is that an African American can easily get a screening test for glaucoma although the risk of this condition is only a few percent, but individuals of other ethnicities, even with a family history of glaucoma, cannot get the test unless they already show signs or symptoms of glaucoma.

Definitions. Dr. Quinn also touched on how the definition of safe and effective for FDA is easier to specify than the definition of reasonable and necessary that Congress gave to Medicare. Furthermore, determining reasonable and necessary may be additive among several pieces of evidence, none of which is sufficient by itself.

Reimbursement. Besides looking at ways to make the process faster, one should consider reimbursement as well. For example, the low prices for molecular tests were not set by the power of the market but by arbitrary decisions mostly made in 1984 that have no relation to value. Certainly this approach does not promote innovation (as economist John Kenneth Galbraith observed earlier about price fixing). A few tests are reimbursed adequately on this price schedule, but not many.

Savings. Dr. Quinn described ways Medicare carriers can ease the way for doctors even though unable to raise the fees—for example, carriers can cut down on lost checks and erroneous claims that cost doctors money.

Unfortunate rule change. Dr. Quinn noted that until 2008, laboratories were responsible for billing and recoupment. Now, however, Medicare has a 14-day rule that makes the hospital that originally draws the blood responsible for blood samples into the future, even though they may be sent to different laboratories around the country. It is the hospital that now has to pay for those tests and attempt to get reimbursement. Dr. Quinn views the prior system where the laboratory was responsible as a better arrangement logistically and better for Medicare financially as well.

NCDs. Dr. Quinn also mentioned that he agrees with Dr. Straube that there should be more NCDs. In fact, he had referred some for California to be considered for NCDs, but their reviews have not yet taken place.

Questions and Answers

Dr. Evans wondered if the United States can take advantage of work done in other countries. Dr. Straube said that international data and decisions are considered and can influence CMS's decisionmaking. In addition, he believes that additional steps should be taken to look for models of coverage decisionmaking internationally that may be worth implementing here.

Dr. FitzGerald remarked that he would like to ask about the concept of "benefit." He elaborated with several examples: Should a person who genetically is not susceptible to lung cancer be allowed to smoke (as long as second-hand smoke is not an issue) without any health insurance or other societal penalties? Also, if a person turns out to be susceptible to diabetes, is that person then responsible for avoiding weight gain? Should his health insurance be affected by whether he takes responsibility for avoiding weight gain? What if a hospital is transmitting a pathogen: should that hospital be reimbursed at a lower level if they do not address this problem? Dr. FitzGerald wondered if we are thinking far enough ahead about where personalized medicine can lead us.

In replying, Dr. Straube first noted that Medicare has been predicted to run out of money by 2016 unless either premiums are raised or expenses are cut; consequently there are no funds for gathering discretionary information even though it might help personalized medicine. Dr. Straube also observed that it is unclear whether payers will embrace the personalized medicine principle that exceptions should be made for individuals based on differing health factors.

Dr. Teutsch wondered about balancing public needs and public good when new technologies usually tend to add to health care costs. Dr. Straube pointed out that ARRA provides funding for adoption and use of health information technology and for a series of prevention and wellness issues, some related to personalized medicine. In addition, the Act provides for NIH to lead comparative effectiveness research, with involvement by all of HHS. Dr. Straube expressed the belief that comparative effectiveness research cannot be done without incorporating an evaluation of cost effectiveness. A relevant example is the recent conclusion by CMS from literature searches that CT colonography has no advantages over existing technology in preventing colon cancer. (An official final determination has not yet been issued.) It also might not be cost effective for the administrators of CT colonography when they would be reimbursed at the same rate as for other colon tests.

Dr. Randhawa inquired whether GINA somehow forbids the use of genetic information in coverage decisions. Dr. Straube replied that his understanding is that medical researchers can still gather genetic information through clinical trials and such but must be sure while doing so to respect the privacy

components of GINA. Dr. Randhawa then asked Dr. Quinn whether he was distinguishing cost from value in his presentation. Dr. Quinn explained that he had been talking mainly about cost and how limits on reimbursement can prevent less costly innovations from being developed. For example, if a $500 procedure could be replaced by a $100 test, yet the fixed fee schedule for the test is still $10, then nobody will develop the $100 test to save on the $500 cost. He added that "cost effectiveness can have different value depending on where you draw the circles around the service and the assumptions you use, but there are some things that are cost effective under nearly any assumptions."

Perspective of Private Health Insurance Companies: Sam Nussbaum, M.D. and Joanne Armstrong, M.D., M.P.H.

Dr. Armstrong. Dr. Armstrong, Senior Medical Director and head of the Women's Health and Genetics Unit at Aetna, presented by teleconference and noted that the cost of health care has outpaced our ability to pay for it—much less for new technologies. Furthermore, the needed care is often not delivered or is of poor quality and, when delivered, may be ineffective. It is in solving these problems that personalized medicine has promise. Dr. Armstrong remarked that at least 11 of 15 major causes of death have already been shown to involve genetic factors.

Rates of testing. New types of genetic testing are increasing at a rate of 10 percent per year, and the use of genetics-based testing is increasing at a 20 percent annual rate. While the total spending in genetic diagnostics is still very small—less than 0.5 percent of total medical spending—the trends are significant. For example, Aetna data show that the cost trends have been about 20 percent per year for the past four to five years.

Pricing. Dr. Armstrong observed that some new genetic diagnostic tests have breathtaking prices (e.g., $3,000 and $5,000 each), even in situations where some equivalent tests are still on the market at $20 and $100 per test. She also commented that there is scant literature that actually links such prices to value.

Diagnostic testing is increasingly being linked to therapies, mostly biological ones. The rate of increase in prices for biological therapies has consistently been 17 percent over the past several years, which is about twice the increase in pricing of other treatments. The prescription costs of biological therapies can be as high as $50,000 to $200,000.

Challenges. Dr. Armstrong presented four "high-level" challenges in trying to integrate personalized medicine and genetic-based medicine into medical care, noting that there are also many other issues. The first challenge is to ensure that the evidence base for these technologies is strong, both to support the coverage and reimbursement for the technologies and to prioritize which are introduced into clinical practice and then used well.

Second is the need for clinical and economic outcome data that demonstrate the value of personalized medicine strategies compared to the status quo. Comparative effectiveness is one way to do this demonstration. Third is the need for decision support tools for clinicians, consumers, health plans, and everybody else who uses these technologies—to make sure that they are used in an effective manner. The fourth challenge is to look again at the current procedural terminology (CPT) system for laboratory testing. As cited earlier today, the current system hinders our ability to use genetic tests and to examine the data and understand the activity that is taking place.

Dr. Armstrong further noted that better information (resulting from better coding) could help in planning medical management strategies from a health plan perspective and work in the area of reimbursement strategies as well—including value-based reimbursement or coverage with evidence collection. She also emphasized how the current lack of specificity of the coding to identify the testing that is being done and

the clinical conditions that it is being used for is a big hindrance to the potential utility of the data for various types of research activities.

Coverage principles. In presenting Aetna's coverage policy principles for genetic technology, Dr. Armstrong pointed out that Aetna uses the same principles for other technologies as well. The following are among the plan's principles: technologies that are covered, promoted, and used must have a strong evidence base; and the health services (requested/provided) need to relate to the prevention, the diagnosis, and the treatment of an illness. Thus, while some genetic services may have personal information utility for financial planning needs and such, they are currently not related to prevention, diagnosis, and treatment of illness and so are not covered in a reimbursement environment.

Also, the information needs to affect the course of treatment of the member, the care and/or treatment needs to be likely to improve health outcomes, the improvement should be attainable outside of investigational settings, and the services need to be consistent with plan design. Plan designs are rarely used in genetics. The evidence standards that are required for the coverage of genetic or personalized medicine technologies are the same as for other technologies, and the consensus appears to be that that is appropriate. In addition, the covered services need to demonstrate improved net health outcome and be as beneficial as an established alternative.

Approval. Generally in the health care field having final approval from the appropriate governmental regulatory bodies is required. However, in the area of diagnostics, most of the testing on the market has not required government approval. Consequently, as a necessity, health plans do much more technology assessment on diagnostics than any of the medical professional bodies or the governmental health agencies.

Value of technologies. Dr. Armstrong explained that it is not clear what value is—whether it is cost-effectiveness or something else. Even if the cost-effectiveness of a therapy is its value, very few therapies have had their cost effectiveness assessed. In addition, health plans have studied only a few services for cost-effectiveness because data for these studies are often lacking.

Aggregating data. Dr. Armstrong said that health plans are already using aggregated data to help consumers and health care practitioners make decisions. A personal electronic record is a tool that makes data more readily available for use in this way. The software applies best practice rules to the aggregated data to identify gaps in care and potentially dangerous drug-drug interactions.

Dr. Nussbaum. Dr. Nussbaum, a new SACGHS member and Executive Vice President and Chief Medical Officer of WellPoint, a health benefits company, stated that in many ways the key driver of health care costs is advancing medical technologies applied to an aging population with chronic illness. Better care coordination and better use of evidence-based medicine would provide the best opportunity to control cost immediately. Only by controlling cost will there be opportunity for more innovation. The IOM work and the new legislation on comparative effectiveness should help in identifying what really works.

Cautioning. There are about 600 specialty drugs in development besides the large number already available. As mentioned earlier today, some biological therapies now cost several hundred thousand dollars. It is important to get solid information on utility, unlike what happened with bone marrow transplants for breast cancer (when millions were spent but survival was no better than for those who did not have transplants). Herceptin is a different kind of example as it is a treatment that works only for women with breast cancer tumors with the right receptors.

WellPoint reviews and policies. In describing how WellPoint sets policies—that affect 34 million people—Dr. Nussbaum said that input is gathered from a Medical Policy and Technology Assessment Group and other sources such as medical specialty societies, literature, Hayes' Technology Compendium, and the U.K. National Institute for Health and Clinical Excellence (NICE). WellPoint staff work with many academic medical centers and medical specialty societies and also survey changing practice patterns and FDA decisions. Along the way, the company has subcommittees of leading hematologists, oncologists, or behavioral health experts also contributing to WellPoint's decisions on coverage and whether treatments are medically necessary.

The company has a policy of transparency and puts a compendium of all the information gathered on a treatment on its website [http://www.wellpoint.com/]. Anyone wishing to comment is then welcome to contact the company.

WellPoint considers the clinical validity as well as the analytical and diagnostic validity of a test and, if the test has clinical validity, then its clinical utility becomes important as well. The test has to have an incremental health benefit compared to current care.

Covered. Genetic tests covered by WellPoint include all genetic testing for cancer susceptibility (e.g., BRCA testing), pre-implantation genetic diagnostic testing, Oncotype DX® gene expression profiling (but not yet Mammaprint, which is a different profile), and KRAS testing to predict response to anti- epithelial growth factor receptor (EGFR) therapy.

Not covered. Tests not covered by WellPoint include biochemical markers or testing for the diagnosis of Alzheimer disease because these tests are not yet proven to confirm a diagnosis reliably or screen asymptomatic patients with or without family history. Other examples are EGFR testing in patients with small cell lung cancer, cardiac ion channel testing for long QT syndrome, and genetic polymorphisms to determine "metabolizer" status for several compounds.

Best quality medicine and evidence-based medicine are higher priority concerns than cost in the decisions not to cover tests. Another factor is false negatives, which could result in individuals not seeking care that they actually need.

Research. WellPoint has a company Health Corps that does health outcomes research. It also partners with health plans, forms strong collaborative research relationships with academic medical centers, and is building a research network. As noted by others, information needs to disseminate more rapidly that the 17-year average that was quoted.

The company is also interested in an integrated electronic health record, which should of course include any known genetic information and generate clinical alerts when there are gaps in care.

By building partnerships, situations like the adverse effects of Vioxx could have been identified within just a few months of its FDA release. Observational studies done well can make a major difference—for example, 40,000 people who had had myocardial infarctions took Avandia without any increased risk. Continued evaluation will show whether genetic technologies work and improve outcomes.

Questions and Answers

Dr. Amos commented that to prove clinical utility, one needs to have good assay systems and appropriate measurement technologies. Too many false positives and false negatives can make a test meaningless. Dr. Nussbaum agreed.

Dr. Billings asked Dr. Armstrong how to assure that the people doing the prior authorization understand the technologies (such as molecular diagnostics) that require prior authorization for payment—given that some of these are fairly complicated and difficult for the experts to understand. Dr. Armstrong replied that, at Aetna, some of these technologies are on a precertification list and there is a small and extensively trained staff to handle the rest.

Dr. Billings wondered why Aetna was an early adopter of BRCA1 testing, and Dr. Armstrong responded that from an evidence point of view, it met the standards of coverage. However, about 5 percent of requests did not meet the medical appropriateness criteria during about 10 years of experience with this test. She also observed that currently about 20 to 25 percent of BRCA1 tests are inappropriate, and she stated that she believes it relates to DTC campaigning.

Perspective of Employer-Based Health Insurance Plans: Michael Critelli, J.D. and Richard Luetkemeyer, M.D.

Mr. Critelli. Mr. Critelli, recently retired Chairman and CEO of Pitney Bowes and current Chair of the CEO Health Transformation Community, first observed that when he took over responsibility for health care in 1990, Pitney Bowes had 14 percent increases in health costs per year and poor employee satisfaction and health. Before explaining the reforms he instituted at Pitney Bowes, he expressed his general support for employer health care systems, noting that employers' strong interest in having healthy, productive employees motivates them to design effective plans. He also observed that there are four payment and coverage systems in this country: the public system, the private insurance system, each state's system of mandates, and the employer system. One of the advantages of the employer system is that it can draw upon the work of the other systems.

Strategies. Pitney Bowes has four strategies for designing a good employer system. The foremost strategy is primary prevention, including nutrition, exercise, lifestyle changes, immunization, and infectious disease prevention and containment. The company provides food in many of its facilities, with encouragements to choose healthier foods through pricing, presentation, information, and merchandising. The company also offers onsite health care facilities (including clinics in the building), an onsite pharmacy, and a smoke-free environment. In addition, the company practices infectious disease control and has redesigned its work spaces.

Mr. Critelli spoke about the benefits of having onsite health care facilities. The convenient access to care that they offer enables continuity of care and increased adherence to treatment plans. Indeed, a study through MedState shows that employees who use Pitney Bowes' clinics are more likely to stay on chronic disease medication programs. Describing the system as value-based health care, Mr. Critelli said that the company works on both patient and provider behaviors to drive the right behaviors, drawing on the best evidence to change plans.

Besides effectiveness, the company looks at behavioral responses. After finding that increasing employee copays cost more over time because people ended up in emergency rooms and hospitals, Pitney Bowes decided to charge nothing for preventive disease or chronic disease medications to drive adherence.

Some of the results of these changes have been saving a net $2.30 for every $1 spent in the clinics, reduced disability and sick days, an average cost-of-care decrease for diabetes and asthma, and reduced hospitalizations. Total overall savings were about $40 million when you looked across all programs: medical, disability, and workers' compensation. Presenteeism (present but not working efficiently) and absenteeism savings are probably significant but are not easy to measure.

Another strategy looks at ways to improve implementation of health information technology. The company is a founding member, and Mr. Critelli chairs an initiative called DOSSIA, which is a personal, patient-controlled, portable, lifelong electronic health record (EHR). This EHR should be ready within a year. DOSSIA is different from an electronic health record in that a lot of the focus is on patient self-management. Thus, DOSSIA includes personal data sources. The system is meant to supplement existing EHRs. The company also aggregates the population-level data from all sources through MedStat to get insights on its self-insured health plan.

In addition to participating with nine companies in a consortium, Pitney Bowes also is a member of the Continua Alliance, which tries to determine better ways to obtain interoperability between medical devices that capture data and the personal health record.

Genetics and genomics. Mr. Critelli said that the company's approach is to use tools to determine what to cover or offer for what populations at what reimbursement rates. There is a possibility of having a process like the behavioral health process to prescreen potential patients.

Behavior health process. For behavioral health, Pitney Bowes uses its employee assistance program (EAP) providers in a sense as incentives or screens. Eight free visits, at company option, are offered for someone wanting to enter the behavioral health system. Employees can make the choice to go out of network into a behavioral health system, with a 70 percent reimbursement rate. However, if they go through the eight-free-visit model before entering a behavioral health system, the reimbursement rate becomes 90 percent.

The benefits of this approach include that the company's behavioral health costs are growing only at the low single-digit rates, more people are being identified with conditions like clinical depression, and, by identifying comorbidities with conditions like diabetes and cardiovascular disease, those conditions can be better managed.

Mr. Critelli would like to see the positive steps that Pitney Bowes has taken be applied to a larger health system.

Dr. Luetkemeyer. Dr. Luetkemeyer, Assistant Medical Director at Caterpillar, Inc., and formerly an internal medicine practitioner and professor at the University of Illinois Medical School, addressed how Caterpillar, being self-insured, makes decisions about coverage and noncoverage. First, Dr. Luetkemeyer offered background information on Caterpillar, Inc., noting that it is a Fortune 500 company employing 110,000 workers and covering 150,000 people. About 50 percent of its sales and 50 percent of its employees are outside the United States. Its workers belong to the same union as autoworkers, and the company has annual health expenditures of $650 million a year. Employees' average age is 41, and the company's turnover rate is between 5 and 10 percent. Since employees often have life-long careers at Caterpillar, it benefits the company to keep them as healthy as possible.

Strategies. In 1930, only staff in the executive office got wellness exams, but now every employee is offered a wellness exam on a regular basis. In 1992, in an effort to reduce health costs, Caterpillar set up a network of preferred hospitals and preferred physicians. In 1995, as part of the demand strategy, the executive office approved a health promotion program. The aim was to get 90 percent of employees, spouses, and retirees to participate twice a year in health risk assessments. The participation rate does run about 90 percent, achieved with an incentive of premium reduction.

In 1999, the Caterpillar-preferred hospitals, the University of Illinois in Peoria, and the Caterpillar Benefits Plan Design in Corporate Medical set up a collaboration. That same year phenotypic hemochromatosis screening was added to the wellness exam because the target population is basically of

northern European descent. In 2000 or 2001, the company decided to find a way for employees to obtain investigational procedures (at that time high-dose chemotherapy and bone marrow transplants specifically for breast cancer) by establishing a special benefit program outside the regular one called Group Insurance Plan A. Employees and dependents could then get these treatments if they entered the NCI studies, which would speed a determination as to whether these treatments really benefited patients (it was determined they that did not).

Additions to the health risk assessment program in the early 2000s were free addiction counseling and the smoking cessation program involving nicotine replacement and the drug Bupropione. For the smoking cessation program, the company only accepted people judged as ready in the health risk assessment program. At the start, the company smoking rates were 25 percent for production workers, 15 percent for salaried employees, and 10 percent for management. Interested individuals were evaluated to determine what stage they were in based on Prochaska's model of change, with those deemed to be in the preparation stage approved for the cessation program. The company evaluated the effectiveness of the program by checking on program participants five years after they had been in the program. After five years, 38 percent of them had quit, while only 5 percent of those who had been in the preparation stage but did not participate in the program had quit.

In 2007, the company eliminated any charges for medicines deemed essential for chronic care of diabetes—including antidiabetic medications, antihypertensives, and antilipidemics. In 2008, Caterpillar started worksite health coaching programs to work with employees on lifestyle changes, using motivational interviewing as a tool. The goal of the program was not just to make employees aware of what they needed to do but to motivate them to change.

Where genetics could help. Dr. Luetkemeyer presented a diagram showing the company's continuing care model for colon cancer that includes preventive strategies and supportive care as well as treatments. The goal of the company's approach is to reduce the incidence of colon cancer, and genetics could help. The company does ask in the initial health risk assessment about whether there is a family history of colon cancer, and if the employee reveals that a first-degree relative has/had colon cancer, then the second health risk assessment delves further into family history information.

Caterpillar provides 100 percent coverage for colon cancer screening at age 50 and has advised 1,200 people under the age of 50 who have at least one first-degree relative with colon cancer to start colonoscopy screening at age 40 but does not cover that cost. Dr. Luetkemeyer noted that it is hard to pay for coverage of a screening colonoscopy without a CPT code. It is also relevant that Caterpillar now uses United Healthcare instead of remaining self-insured.

Using the Duke Evidence-Based Practice Center, the company has developed eight criteria for measuring a program's quality of colonoscopy and obtained agreements from plan colonoscopists to use those criteria.

Drug costs. In 2006, to deal with drug costs at a time when cholesterol-lowering statins were a major cost, Caterpillar established a policy of no co-pay charges whatsoever for the use of generic versions of the statins. When this did not produce enough voluntary changes to the lower-cost drugs, a step therapy plan was put into effect, with physicians and employees being informed of the change. The response produced an 80 percent switch to generics. Adherence also went up from about 70 percent to 82 percent.

Dr. Luetkemeyer explained that they have tracked the impact of their step therapy plan, and health indicators for employees showed improvement.

Dr. Luetkemeyer also mentioned that Caterpillar had the goal that the growth of their health care costs would not exceed growth of the consumer prince index (CPI). The company's actual costs in recent years have been below CPI growth.

Questions and Answers

Dr. Williams asked how GINA will impact the potential movement of personalized medicine into Caterpillar's disease management and other health programs. Dr. Luetkemeyer replied that if genetics could help the company to target people at higher risk of particular diseases (e.g., of colon cancer), there could be cost-savings in screening programs. Herceptin-positive testing is already used by the company's pharmacies. Dr. Critelli added that he sees the personal health record as a critical tool, especially once data sets can be readily aggregated and data examined at the population level. He also noted that the House version of the stimulus package would have crippled aggregation of population-level data but fortunately the Senate version prevailed.

Dr. Wise wondered if there are conditions in GINA that would "firewall" some genetic and genomic data, making them unavailable for use. Dr. Luetkemeyer described the following problematic example: With hemochromatosis screening, that question came up with the testing for variants in the HFE gene in people who had high transferrin and ferritin levels, that is, phenotypic iron overload. Caterpillar's lawyers would not permit any genetic testing, so the company instead developed a letter to the employees saying you ought to go talk to your doctor about this test. In that letter the company did educate the physicians on the importance of testing for the patients in question. Many of them had ferritin levels above 1,000 ng/mL, and they were all asymptomatic. Thus, without some protection, the lawyers will not allow genetic testing of an employee because of fears that it will get out into the public through the human resources departments.

Dr. Critelli added that Pitney Bowes is looking to see to what degree outsourced providers have more freedom of action. Four company clinics are operated by company employees, and four are operated by outsourcers. Because outsourced clinics have more freedom to treat dependents and retirees than the in-house people do, the company is likely to increase outsourcing. It appears to be a workable model because there are other benefits to the outsourcing model in the states in which the company has clinics.

Dr. FitzGerald inquired about any alternatives for the women with breast cancer who did not want to participate in the Special Group Insurance Plan A and enter an NCI clinical trial. Dr. Luetkemeyer's reply was that they would then have no coverage for high-dose chemotherapy or stem cell and bone marrow transplants; they would still have access to standard approved treatments.

Regarding (last-chance) experimental breast cancer treatments, Dr. Critelli noted that the Pitney Bowes ethics committee looked at a different situation, bariatric surgery. The company's approach was to require a preprocess before the bariatric surgery would be approved. Patients who go through the other process first get a much higher rate of reimbursement when going on to bariatric surgery. If anything is on the margin, the company aims to have a predecision process and to encourage participation in the predecision process by rewarding participants with higher rates of reimbursement.

Committee Discussion with Roundtable Participants

Introduction. Ms. Aspinall introduced a discussion with the roundtable participants by asking them to talk about needs for the future of health care and what areas SACGHS could address. She proposed looking at several categories: drugs, physicians, employers, laboratories, payers, and patients.

Regarding drugs and pharmaceutical companies, the trend is to more targeted drugs with smaller targeted markets and more effective drugs with fewer side effects. Will that increase the cost per drug? Will it increase compliance? Looking at oncology, the case is rather compelling. Ten percent of drugs were targeted in 2001 and maybe 60 percent will be targeted in 2010, with probably 80 percent of those targeted on a genetic basis (not necessarily an inherited basis).

Physicians need more tools and more education on genetics and genomics. Currently, 17 percent of medical schools have no formal education on genetics and genomics in their four-year education. Physicians also need more treatment guidelines. Will there be increased liability as we look to the future?

Regarding employers, they seem to be taking a long-term view of employees' health. Will the trend towards self-insurance plans continue? Certainly there is an increasingly aggressive use of wellness plans.

Laboratories have intense data acquisition and storage requirements. Personalized medicine and genomics is all about data and not just about the wet laboratory. There are, as spoken about today, reimbursement challenges with new technologies. What can be expected for the future, and what actions are needed along the way?

Payers are demanding evidence-based medicine, so one issue is how to provide it. Payment may be contingent on drug effectiveness. One possibility is to pay only if the drug works. BlueCross has talked for many years about funding its own database on patient outcomes rather than relying on pharmaceutical or diagnostic companies. Besides outcomes data, payers want tests that prove the procedure's relevance to the patient and physician.

Lastly, regarding patients, what is the impact of consumer-directed health plans? Patients are more educated but more stressed. They are increasingly affected by decisionmaking, such as whether or not copayments will be charged. Ms. Aspinall thinks that patients need to get more involved than they were in the past. Improved compliance is a result as personalized treatments grow. Implementing predispositional testing increases the number of individuals who are living with the potential of a disease, not the disease itself.

The pattern in health care currently is to spend relatively little early and much more as patients get older and sicker. Is there potential for genetics, genomics, and personalized medicine to change this trend? And will investing in diagnostics and prevention genomics achieve a benefit in quality of life and financial savings? If this is the future, (A) Is this the future we want? and (B) If so, how do we get there?

It may be the time to take proactive action as there seems to be tremendous openness around the country to health care reform.

Discussion

Dr. Licinio remarked that with clinical studies data are collected in very artificial conditions because of the restrictions imposed by the inclusion criteria. For example, in his protocols, he has generally accepted only 10 percent of candidates. His question is, then, how to apply the information gleaned in such studies to real conditions in which individuals may have several different diagnoses all at once.

Dr. Nussbaum agreed that finding out what works in a real-world setting is a major issue. He suggested that the more entities can work together to increase the size of a database, the more useful the data will be. If such combined databases are created, new ones will not be needed to achieve useful studies of safety, effectiveness, and outcomes.

Dr. Epstein commented that it is advisable to consider together the data from randomized clinical trials (RCTs) and real-world observations. One example is the importance of being aware that in RCTs of lipid-lowering compounds, 92 percent of study participants stayed on the test medications for 4.5 years; in contrast, in real life, 50 percent of 60 million patients stop taking the medication after one year. Thus, real-world effectiveness studies are needed.

Dr. Luetkemeyer added that in the real world where one individual can have multiple diagnoses, the delivery care system needs to transform into processes of team care. Currently, there are no processes for creating this kind of transformation.

Ms. Aspinall wondered if the current discussion is raising evidence standards for diagnostics higher than those required for drugs. Dr. Evans responded that evidence for drugs has become higher to meet prior safety concerns, and he thinks genetics is approaching the same level but not exceeding it.

Commenting that having to make decisions based only on claims data is limiting, Dr. Williams stressed that SACGHS should endorse the integration of databases (with rules to protect individuals) as such databases will be critical for learning. He also noted that it is difficult for some of this important real-world information on effectiveness to get into the scientific literature. As an example, Dr. Williams's institution has done some interesting work around Warfarin management that has not been accepted for publication because it is not a randomized control trial.

Dr. Nussbaum agreed with Dr. Williams that much of the data that exist in observational studies often do not meet the rigorous criteria for publication in academic journals nor do such data manage to intrigue many of their academic colleagues. Perhaps the clinical and translational science institutes will create a new breed of scientist that will work with different databases, including partnering with such entities as Aetna and Medco.

Dr. Nussbaum also remarked that he was excited to hear of the various company databases. He suggested with ARRA-funded comparative effectiveness research and $1 billion dollars to CDC, perhaps some of that money can be earmarked for new methods of analyzing these large databases. Dr. Quinn noted that AHRQ came out a year or two ago with a 200-page book called *Using Registries for Outcomes Analysis* (available from the AHRQ clearinghouse).

Mr. Critelli remarked that patients are seeking health information through websites like WebMD or gleaning anecdotal information from family and friends without necessarily getting an accurate picture. He also thinks that if the medical community insists on perfect evidence a vacuum will be created that is filled by inaccurate speculation. For example, while the medical community waited for a perfect answer regarding the effectiveness of bone marrow transplants for treating breast cancer, advocacy groups got legislation mandating coverage of this procedure. RCTs later established that the procedure did not work.

Mr. Critelli also expressed the wish that the Committee, in exploring the development of evidence-based medicine, will propose a mechanism to "revisit" state legislative mandates requiring coverage of certain procedures; Mr. Critelli observed that these mandates are often based on bad or nonexistent science.

Ms. Aspinall asked Mr. Critelli about the reaction from employees at Pitney-Bowes as the company implemented the changes described during his presentation. She also inquired how long it took for the company to begin to get a return on its investment.

Mr. Critelli said that a favorable return on investment can be seen after only a few months for immunizations because the company saves money from having fewer of its employees needing to see

outside doctors. On the other hand, receiving a favorable return on the investment in a plan design change usually takes two to four years; during that time, avoided hospitalizations result in cost savings that eventually accumulate to exceed the costs of the plan change. Mr. Critelli added that raising the copayment can result in an immediate favorable return on investment. When Pitney-Bowes raised the co-pay on MRIs, the company observed reduced usage of MRIs the next year. Conversely, it will take longer to receive a favorable return on investment from the company's decision to charge nothing for chronic disease medications. After such a policy change, cost-savings slowly accumulate over time from avoidance of emergency room visits or hospitalizations.

Mr. Critelli also noted that one advantage companies have over the government in designing health benefit plans is that companies are not required to balance their budget each year, so companies can look ahead more than one year at a time when budgeting.

Dr. Luetkemeyer also answered Ms. Aspinall's question. He stated that for colon cancer he anticipates that it will take three to four years for the company to receive a favorable return on its investment.

Dr. Teutsch observed that the Committee's discussion was overlooking the fact that 16 or 17 percent of the population have no health insurance—a situation that creates major equity issues. Also, only about 3 percent of health care dollars go to preventive care at the present time. He asked everyone to think about how SACGHS can optimize social benefit. Dr. Nussbaum responded that a basic benefit package is being debated as a component of health care reform, and his company and others who offer insurance believe that preventive services should be "first dollar covered."

Dr. Nussbaum also said that at WellPoint consumer-directed health plans are the fastest-growing health plans. In these plans, consumers have their own savings accounts and have shared accountability for spending. After a certain amount, it becomes a coinsurance model. A critical factor is the emphasis on preventive services, with the company health plan paying the first dollars for these.

Referring to work by Rand several years ago and more recent work in children, Dr. Nussbaum stated that preventive services ought to be delivered 100 percent of the time and should be part of pay for value in any of the government or private sector reimbursements. He then posed a question as to what genetic services should be part of that preventive care and described a cost graph with an early increase to pay for preventive services, then a flat interval where, hopefully, potential patients are exercising, eating nutritiously, and living a better lifestyle, then another peak in costs for treatments that comparative effectiveness studies show can be prescribed later, such as generic statins. The sum of overall health costs should be less with this approach.

Dr. Luetkemeyer referred to USPSTF as a very useful external source of unbiased recommendations for specific preventive services. Caterpillar Inc. now covers completely all of the Task Force's Grade A recommendations. It would certainly be helpful to have a similar source of recommendations for genomics.

Mr. Critelli agreed with the concept of 100 percent coverage as an incentive for specific treatments and behaviors. He said that the recommendation he would add is to provide health services at a convenient site, such as near or at the workplace. In the case of prenatal care, besides delivering prenatal counseling onsite, Pitney Bowes gives the gift of a portable baby carrier after birth. The rate of premature, low weight births has dropped significantly. Subsidies for services with exceptional medical benefits also help.

Dr. Teutsch commented that he worked for many years at USPSTF and is aware that the standard for its recommendations is set high. A general question then is what kind of information is necessary to justify a recommendation.

Dr. Epstein responded that if one looks at the criteria that USPSTF considers for making Grade A recommendations, one will discover they are largely looking for RCTs. Their Grade B and C recommendations are based on observational studies. Dr. Epstein suggested that the Committee could work on coming up with a different system—perhaps one in which the level of certainty required would change based on the severity of the disease.

Dr. Armstrong added that the American Health Information Community (AHIC), which has changed its name to the National eHealth Collaborative, and others are creating an evidence-based medicine matrix. She explained that the X-axis of the matrix would be the medical benefit that accrues. On the far left side would be negative medical benefit and, all the way to the right, substantial medical benefit. Along the Y-axis one would plot the level of certainty about the effectiveness from the published literature. Thus, at the high end of the Y-axis is high certainty and near the bottom is low certainty.

Then in mapping the various types of studies, the traditional USPSTF A level would be in the far upper right-hand range. Below that might be studies of moderate certainty of effectiveness but substantial net medical benefit. Lower yet would be studies of no known potentially significant benefit and so-so certainty of the science; these would be studies that require additional evidence for approval of coverage. It would be important to be able to get all the various entities that use these evidence-based grids and matrices to agree on what is sufficient for a coverage position.

Dr. Nussbaum commented that a lot of science is unproven except within a framework of certain medical professions can lead to legacy issues in health care with debatable effectiveness (e.g., arthroscopic knee surgery or back surgery). It may be necessary with new science, new biology, and a new set of diagnostic and clinical tests to build a different framework. This effort could lead to reversing legacy issues and emphasizing genetic science and clinical science. The emphasis would then be on comparative effectiveness, effectiveness, and outcomes research. How to fund this research could be a challenge as small companies tend to be involved rather than big pharmaceutical companies. Dr. Nussbaum added that he would not like to see the health care field at the same stage in five years, where 40 percent of medicine does not have proven benefit.

Dr. Williams observed that Dr. Armstrong's matrix still assumes *a priori* that RCTs provide the best kinds of evidence. However, two presenters today have said that sometimes real world evidence is more important. Yet, that will not be acceptable as long as this type of matrix is used.

Noting that he had worked on the matrix, Dr. Teutsch responded that it is based on the level of certainty and more than just RCTs are accepted as providing a high degree of certainty. Ms. Aspinall wondered if some guidelines are then needed for applying the certainty criteria.

Returning to breast cancer and bone marrow transplants, Ms. Darien mentioned that she had written some articles on this subject, and that while it is an example of advocacy gone amok, there was one organization that always opposed the treatment because of insufficient evidence—the National Breast Cancer Coalition. Besides advocacy groups, some oncologists genuinely believed that the treatment worked, and one medical researcher in South Africa, Dr. Werner Bezwoda, published falsified data favoring the treatment. The media too had a role by demonizing insurance companies that refused to pay for the treatment. Indeed, Caterpillar, Inc., was courageous at the time for restricting its coverage to those employees who had the procedure within the context of a clinical trial. Ms. Darien added that her aunt was one of the patients who died from the bone marrow transplant instead of from her breast cancer.

Dr. Evans cautioned that one should not go too far in the other direction after saying that RCTs are not the only sources of solid evidence. He agrees with Dr. Armstrong's statements that one should use the "same coverage policy principles for genetic technologies as for all other technologies."

Dr. Quinn commented that with therapeutic studies one may have to wait 10 years to be sure of the results, but, once a therapy is shown to be effective and a diagnostic test has been linked to it, it would be pointless to wait another 10 years to confirm the effectiveness of the diagnostic.

Dr. FitzGerald agreed with Drs. Evans and Quinn and said that what counts as evidence depends on what the goals are, who decides what the goals are, and which goals are being pursued. So then one has to consider who gets to decide which goals are going to have preeminence. This, in turn, leads to the issue of public and stakeholder engagement. Different people have different goals, obviously, and should.

Dr. FitzGerald added that he suspects that all members of the committee are in favor of evidence-based medicine and observed that there will be political pressures and other interests in health care reform that may be in opposition to evidence-based medicine. Therefore, it will be important to have clear standards for evidence-based medicine when dealing with these pressures.

Ms. Aspinall asked if, after this meeting's discussions of study design and integration of databases, participants have recommendations about what SACGHS can do to make a smoother future. She cautioned participants to consider this question in light of limits of time and resources and the need to prioritize.

Dr. Epstein responded by saying that perhaps the Committee could weigh in on principles that would facilitate the population having access to their own genetic information. Two possible principles mentioned today are (1) treat genetics the same as other new technologies and (2) determine the type of evidence or evidentiary standards that are needed. SACGHS could add discussion under each principle.

Dr. Nussbaum agreed that starting with principles is a good idea. He added one—that the evidentiary bases should be comparable. He wondered if SACGHS ought to look at where there are similarities and variances in tests among public and private payers.

Dr. Nussbaum also proposed designing a path forward by organizing a national registry, observational study, or database on selected unproven but promising processes. Dr. Billings inquired if this would mean that everyone would have access to those tests that had passed through the evidentiary process. Dr. Nussbaum replied that if the evidence is overwhelming, he would expect CMS and health plans to accept it. When evidence is clear, there is commonality; it is when evidence is ambiguous and uncertain that coverage among payers is uneven.

Dr. Billings observed that, in his experience, what constitutes adequate evidence has varied among different entities. Dr. Williams added that some health plans may choose to eliminate a whole category, such as genetic testing.

Dr. Luetkemeyer pointed out that the most likely area for cost-savings in a health plan is the drugs (e.g., switching to generics). DTC advertising by genetics companies scares him. He declared that the Committee ought to take a stand against DTC advertising by genetic companies until they have outcome data that everybody will support. Ms. Aspinall assured Dr. Luetkemeyer that that issue is on the SACGHS agenda.

Dr. Armstrong stated that she agreed with Dr. Nussbaum regarding the need to get uniformity across the public and private sectors. She also said that she wondered if the right questions are being asked to support coverage decisions. She would like to see a consensus as to what constitutes ideal evidence and what constitutes sufficient evidence.

When Ms. Aspinall inquired whether Drs. Nussbaum and Armstrong are referring to scientific or economic evidence or both, they both replied "scientific first." Dr. Luetkemeyer said that he would say scientific first and then outcome because effectiveness must be measured.

Dr. Epstein added that the system should be geared to promote and reward cost-saving technologies. Currently, the system does not do that. One should not have to add to cost to add to quality (especially in a one trillion dollar system). He referred to the health care writings of Rick Carlson who, besides being a public policy expert on genomics, insisted that the system is upside down. Some states have half the health care costs of other states but the same longevity. We need to learn why.

Dr. Williams wondered if SACGHS should therefore advise the HHS Secretary not to invest anything in genetics and genomics until the current situation is fixed; otherwise there will be additional costs. By one perspective, this is an issue on the luxury fringe. Dr. Quinn indicated that there are genetic/genomic tests that are cost-saving (e.g., showing whether a particular treatment will be effective); these should be encouraged. Dr. Evans said that he agrees with Dr. Williams that genetics should not be oversold. There could even be a backlash.

Lt. Col. Wattendorf observed that it can be difficult for the large military health system to depend on USPSTF because it can assess only a few diseases at a time. Indeed, the Task Force has questions about how well the PSA test works that obscure the fact that, for African Americans with family histories of prostate cancer, the PSA test can indicate low or high risk.

Lt. Col. Wattendorf also said that there is a need for evidence from small cohorts with modest gene variants. He wondered if it would be possible to match DOD cohorts with those from other health plans and whether the Federal Government could enable this effort.

Dr. Amos proposed that the Committee advise the HHS Secretary on how genetics can be used in health care. While nucleic acid testing can provide great value in medicine, it is unclear whether that will save money in the long run. This issue may have to be examined on a case-by-case basis. Maybe genetic testing should be defined as understanding the environment's influence on the genome.

Dr. Amos also agreed with Dr. Nussbaum that the biggest cost in health care now is medical care for chronic diseases. He asked about the value proposition of limiting the discussion to nucleic acid testing. The discussion should be expanded to other ways to look at genetics.

Ms. Walcoff noted that while it did not make it into the final budget, initially the fiscal year 2008 budget had $15 million in seed money to explore how to create a merged database of phenotypic and genomic information that could be searched by investigators but that had privacy protections in place. HHS staff is also thinking about this possibility. Dr. Walcoff suggested getting some information on what NIH is doing regarding genomics and the environment—to be reported on at the next SACGHS meeting.

Dr. Feero recommended paying more attention to family history as a low-cost tool. He said that the literature base shows that little is known about the utility of family history as a screening tool for preventive services and that this viewpoint is likely to be borne out in an upcoming State-of-the-Science conference. Dr. Feero's also noted that EGAPP and its processes provide a laboratory to study evidentiary requirements. He suggested that the Committee recommend EGAPP look at the effects of

setting different evidentiary thresholds than USPSTF. Dr. Feero added that he does not expect USPSTF to take up genetic applications at this point in time, considering the state of the literature base.

Dr. Williams suggested that the assembled group is an appropriate one to project into the future and think about how their various sectors, as well as the work of SACGHS, will be impacted by health care reform. Ms. Aspinall summarized the question as "where do you think health care reform is going and where would you like it to go?" (in general and vis-à-vis genetics).

Dr. Nussbaum offered the following ideas: (1) The U.S. should still be a beacon for science, scientific achievement, and advancement for science. (2) The U.S. health care system is too expensive, partly because of using technologies before they are proven. (Dr. Nussbaum also pointed out that the U.S. system is no where near the top 10 in quality.) (3) The U.S. should use services that have an impact, and this point is relevant to genetics, genomics, proteomics, and everything else that is biologically based. (4) The science should drive consensus viewpoints on coverage. A good place to start with science-based coverage review would be a review of some of the companion diagnostics for $100,000 biological therapies for cancer.

Dr. Nussbaum then observed that as we get to know our genome to predict illness and manage public health over time, we can all theorize and we can have hypotheses that can be tested, but those are not going to get proven for decades. He suggested selecting both the best opportunities to prove science and the best opportunities for economic review.

Dr. Quinn suggested that the best value would come from looking at companion diagnostics for the most expensive drugs. Dr. Luetkemeyer added that companion diagnostics help not only the person who responds to the drug but also the person who would suffer complications from the wrong drug. Dr. Evans noted they were talking about companion diagnostics that are not genetic and that companion genetic diagnostics have not yet proven their worth.

Summation of Key Roundtable Issues and Next Steps: Ms. Aspinall

Ms. Aspinall emphasized three issues related to priorities and perspectives from the roundtable and earlier discussion: (1) the role of DTC information, whether advertising or testing (a topic that SACGHS has already been addressing); (2) the need for evidence-based medicine, with the Committee calling for a clear and transparent roadmap for diagnostics, procedures, and drugs; and (3) encouraging the use of systems, products, or services that currently exist to improve the health of Americans or to lower the cost of health care. The third issue is the low-hanging fruit; the group definitely has said to use the resources that we have today as a society more effectively.

Dr. Williams proposed adding a fourth item—the database sharing issue. It will be important for SACGHS to weigh in on this issue. He suggested that the Committee should specifically promote the idea that genetic data needs to be coded.

Ms. Aspinall mentioned genetic exceptionalism as an aspect of the four topics already listed. She suggested that the Committee present the principles suggested by Dr. Epstein and discussed by the roundtable separately instead of including them under the evidence-based issue.

Dr. McGrath inquired if or how EGAPP's approach differs from other evidence-based assessments. Dr. Roche said that CMS anticipates a convergence, with the EGAPP methods and others forming the basis for getting to the evidence that will be used for coverage determinations in the future for screening and diagnostic uses. Ms. Aspinall added that she believes that EGAPP is working on literal standards (e.g., the number of people for a clinical trial).

Dr. Teutsch indicated that EGAPP has specifically examined how to review evidence for the use of genomic tests—everything from prevention on through prediction, prognosis, and pharmacogenomics—with the idea of making recommendations for clinical evidence that is of net benefit. However, there are lots of different decisionmakers using somewhat different standards and having different informational needs. The AHIC work that Dr. Armstrong referred to (based on the USPSTF approach) aimed to build a roadmap that would use EGAPP information for decisionmaking. It was also designed to provide benchmarks for innovators. The EGAPP bar may be too high for some.

Ms. Aspinall then thanked everyone for helping to develop the lists of issues and principles.

Concluding Remarks

Dr. Teutsch expressed appreciation for the fascinating and rich discussions and summarized the meeting highlights.

Federal updates. The Agency presentations provided valuable insights on the many federal activities relevant to the work of SACGHS. It is likely that the Committee will want to revisit the topic of the implementation of GINA at future meetings.

DTC genomic service. Regarding consumer-initiated genomic services, SACGHS decided to form a task force to examine prior SACGHS recommendations that are germane to concerns about DTC genetic testing. The task force will prepare a draft paper for the June meeting.

Genomic data sharing. In the stimulating discussion about informed consent, privacy, and discrimination, the Committee considered new paradigms for research. SACGHS staff will explore the opportunities for collaboration with other federal advisory committees interested in genomic data sharing issues.

Genetics education and training. Findings from the Genetics and Education Task Force data-gathering efforts will be presented at the next SACGHS meeting, and a draft report is anticipated in the fall.

Future of the health system. The Committee discussed possible next steps such as discussing principles that are needed for access to genetic technologies, including an evidence-based roadmap and evidentiary standards. In addition, the Committee can begin to explore whether some technologies can lower the cost of health care, and incentives or barriers to developing and implementing them. A process for data sharing is another priority.

June meeting topics. Dr. Teutsch proposed the following as topics for the June 2009 SACGHS meeting: implementation of GINA, continued discussion of the future of the health care system (including patient, provider, and industry perspectives), a preview of public comments on the patents report, and the draft paper from the Direct-to-Consumer Genetic Testing Task Force.

Ms. Aspinall suggested including someone from industry, someone from HHS, and someone who could provide an update on health care reform in the planned discussion on the future of the health care system. Dr. Teutsch commented that having the new HHS Secretary in place by the June meeting could be helpful, too.

Dr. Teutsch thanked everyone for their active participation in a productive meeting.

ADJOURNMENT

The meeting was adjourned at 2:31 p.m.

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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.

Steven Teutsch, M.D., Ph.D.
SACGHS Chair

Sarah Carr
SACGHS Executive Secretary