**DEPARTMENT OF HEALTH AND HUMAN SERVICES**

**Twenty-Third Meeting of the**
**SECRETARY'S ADVISORY COMMITTEE ON**
**GENETICS, HEALTH, AND SOCIETY**
**October 5-6, 2010**

# Meeting Summary

National Institute of Health
Building 31, Room 6C6
9000 Rockville Pike
Bethesda, MD 20892

Prepared by the Office of Biotechnology Activities
National Institutes of Health

## Participants

### October 5, 2010

**Committee Member Present**

Steven Teutsch, M.D., M.P.H., Chair
Janice V. Bach, M.S., CGC
Paul Billings, M.D., Ph.D., FACP, FACMG
David C. Dale, M.D.
Gwen Darien
Charis Eng, M.D., Ph.D.
James P. Evans, M.D., Ph.D.
Andrea Ferreira-Gonzalez, Ph.D.
Barbara Burns McGrath, RN, Ph.D.
Samuel Nussbaum, M.D.
Charmaine Royal, Ph.D.
Sheila Walcoff, J.D.
Marc S. Williams, M.D., FAAP, FACMG

***Ex Officio* Members/Alternates Present**

Vence Bonham, J.D. for Eric Green, M.D., Ph.D (National Institute of Health)
Sarah Botha, J.D. (Federal Trade Commission)
Michael A. Carome, M.D. (Office for Human Research Protections)
Denise Geolot, Ph.D., RN (Health Resources and Services Administration)
Eric Green, M.D., Ph.D. (National Institute of Health)
Adam B. Kanis, M.D., Ph.D. (Department of Defense)
Katherine Kolor, Ph.D. for Muin Khoury, M.D., Ph.D. (Centers for Disease Control and Prevention)
Douglas Olsen, Ph.D., RN for Ellen Fox, M.D. (Department of Veterans Affairs)
Gurvaneet Randhawa, M.D., M.P.H. (Agency for Healthcare Research and Quality)
Jeffery Roche, M.D. (Centers for Medicare & Medicaid Services)
Zivana Tezak, Ph.D. and Elizabeth Mansfield, Ph.D., for Alberto Gutierrez, Ph.D. (Food and Drug Administration)
Jennifer Weisman, Ph.D. (Office for Civil Rights)

**SACGHS Staff**

Sarah Carr, Executive Secretary
Symma Finn, Ph.D.
Cathy Fomous, Ph.D.
Allison Lea, M.A.
Andrea Collins

**Speakers**

Paul Billings, M.D., Ph.D., FACP, FACMG (SACGHS Member)
David Dimmock, M.D. (Medical College of Wisconsin)

Charis Eng, M.D., Ph.D. (SACGHS Member)
Elizabeth Mansfield, Ph.D. (Food and Drug Administration)
Barbara Burns McGrath, RN, Ph.D. (SACGHS Member)
Juli Murphy-Bollinger, M.S. (Johns Hopkins University
Marc Williams, M.D. (SACGHS Member)
Karl Voelkerding, M.D. (ARUP Laboratories)

**Public Commenters**

Edward McCabe, M.D., Ph.D. (Linda Crnic Institute for Down Syndrome, University of Colorado)
Mary Steele Williams, M.N.A. (Association for Molecular Pathology)

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Jennifer Weisman, Ph.D. (Office for Civil Rights)

**Speakers**

Laura Arbour, M.D. (University of British Columbia and Vancouver Island Health Authority)
Paul Billings, M.D., Ph.D., FACP, FACMG (SACGHS Member)
Vence L. Bonham, Jr., J.D. (National Human Genome Research Institute, National Institutes of Health)
Charis Eng, M.D., Ph.D. (SACGHS Member)

Morris W. Foster, Ph.D. (University of Oklahoma)
Barbara Burns McGrath, R.N., Ph.D. (SACGHS Member)
Charmaine Royal, Ph.D. (SACGHS Member)
Rebecca Tsosie, J.D. (Arizona State University)
Marc Williams, M.D. (SACGHS Member)

**Public Commenters**

Joann Boughman, Ph.D. (American Society of Human Genetics)

**SACGHS Staff**

Sarah Carr, Executive Secretary
Symma Finn, Ph.D.
Cathy Fomous, Ph.D.
Allison Lea, M.A.
Kimberly Taylor

## Tuesday, October 5, 2010

### Opening Remarks

Dr. Steven Teutsch, Chair of the Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) welcomed everyone to SACGHS' 23rd and final meeting. He explained why the Committee was sunsetting and that the SACGHS charter would expire February 28, 2011. He highlighted the past accomplishments of SACGHS and proposed sending a letter to Secretary Sebelius summing up the Committee's prior work and concluding thoughts on affordable genome sequencing, genomic data sharing, and comparative effectiveness research as well as offering guiding principles for the continued integration of genetics and genomics in clinical medicine and public health. The Committee collectively agreed to draft a letter at the meeting.

Dr. Teutsch reviewed the meeting agenda and mentioned the following updates:

- Several Committee members volunteered at the last SACGHS meeting to serve on the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) Working Group for Carrier Screening. Although SACGHS is coming to closure, SACHDNC has invited SACGHS members to continue serving on this working group.

- The SACHDNC draft report on the retention and use of residual dried blood spot specimens after newborn screening has been revised based on comments from SACGHS and others, and the final report was to be sent to the Secretary later that week.

Executive Secretary, Sarah Carr, reminded the Committee of the standards of ethical conduct for employees of the executive branch.

### Updates from the Food and Drug Administration (FDA)

Dr. Elizabeth Mansfield, from the Office of In Vitro Diagnostic Device Evaluation and Safety, Center for Devices and Radiological Health, provided the Committee with an update of recent FDA activities related to the oversight of laboratory-developed tests (LDTs). The FDA held a public meeting in July 2010 to explore this topic and had an additional period for the submission of public comment. The comments would be used in the development of a document to provide the regulatory framework of oversight for LDT. As a part of the update she discussed the history and definition of LDT and clarified the FDA's intent to regulate devices, not laboratories. She pointed out the loopholes that developed as a part of FDA's initial approach and laid out a possible framework for LDT oversight. Elements of the framework could include risk-based oversight (addressing the highest risk tests first), registration and listing to understand which genetic tests are available and by whom, and classification panels to classify tests with no predicates or existing regulations (FDA hopes to avoid numerous *de novo* actions). Regarding direct-to-consumer (DTC) tests, Dr. Mansfield remarked that the DTC model is not appropriate for enforcement discretion because consumers receive test results without the involvement of a health care provider. FDA has, however, been involved with DTC testing companies as a separate activity.

Discussion. A Committee member asked if FDA had a timeline for LDT oversight, to which Dr. Mansfield reported that it does not.

## Public Comments

Edward McCabe, M.D., Ph.D., Executive Director of the Linda Crnic Institute for Down Syndrome at the University of Colorado and former SACGHS Chair, spoke to the Committee to invoke awareness of the current discriminatory practices against children with Down Syndrome involving denial of medical coverage for certain therapies by Medicaid in Mississippi and Aetna in Colorado. He noted that the Patient Protection and Affordable Care Act identifies speech, physical and occupational therapy as an integral part of the developmental growth of children with Down Syndrome and have shown to increase quality of life, cognitive ability, and life expectancy over the last 50 years. Denying coverage for such services could prove to be quite detrimental for these individuals.

Mary Steele Williams, M.N.A., Chief Operating Officer and Director of Scientific Programs, Association for Molecular Pathology (AMP), spoke to the committee on behalf of AMP to commend SACGHS on their attention to whole-genome sequencing and for their work over the past decade. She relayed AMPs concerns of the clinical applications related to whole-genome sequencing and AMP's recommendation to create a central repository for all sequencing data and corresponding phenotypic information. Additionally, AMP recommended relevant advisory committees, stakeholders, and agencies to consider the challenges involved with whole-genome sequencing.

## Genetics Education and Training

Dr. Barbara Burns McGrath, Chair of the SACGHS Genetics Education and Training Task Force presented the draft report on genetics education and training and asked that the Committee come to consensus and approve the final recommendations. She began by recounting the history and scope of the report and briefly discussed the public comments received earlier in the year as well as the Task Force's analysis and integration of these comments into the report. She then reviewed each of the six recommendations and the proposed points for the cover letter to the Secretary.

Discussion. The Committee offered suggestions for the recommendations and the cover letter to the Secretary. The revised recommendations were presented the following day for the Committee's final vote.

## Affordable Whole Genome Sequencing

Dr. Charis Eng, Committee member and Co-Chair of the Task Force on Implications of Affordable Whole-Genome Sequencing (WGS), introduced the topic and recapped the Committee's previous work on WGS. She noted that the two goals of the session were to (1) learn about the practical implications of affordable WGS from the laboratory and clinic perspectives and (2) identify the issues and needs in this topic area that should be brought to the Secretary's attention and come to consensus on any guidance or recommendations that would address these needs.

WGS from the Laboratory Perspective

Karl Voelkerding, M.D., Medical Director for Advanced Technologies and Bioinformatics at ARUP Laboratories, discussed the progression of next-generation sequencing and the use of this technology for multigene panels, whole-exome sequencing, and WGS. He explained that genetic testing is transitioning to a bioinformatic world because algorithms and alignment and assembly methods are needed to analyze and interpret sequence data. Considerable computational power and storage capacity are required to perform these analyses. Dr. Voelkerding noted that storing large databases offsite from the institution that generated the data raises issues related to patient privacy and compliance with the Health Insurance Portability and Accountability Act (HIPAA). He remarked that sequencing technology is getting easier,

but the cognition required for the interpretation of these data is escalating. In conclusion, Dr. Voelkerding said that it is important to keep an historical perspective in moving forward—next-generation sequencing is a new technology as were polymerase chain reaction (PCR) and array-based testing. Scientists, physicians, and others will work through the issues. In addition, it is important to expand education and training, develop technical standards and guidelines by leveraging existing infrastructure within professional organizations, develop a genome-wide genotype-phenotype database, promote appropriate medical use of sequencing technologies, and address genomic information access and portability.

WGS from the Clinical Perspective

David Dimmock, M.D., Assistant Professor of Pediatrics and Genetics at Medical College of Wisconsin, and Children's Hospital of Wisconsin, presented a case of a young male patient at the Children's Hospital in which whole-exome sequencing was used to assist diagnosis and inform clinical decisionmaking. He also reviewed some of the ethical concerns associated with whole-exome and whole-genome sequencing. Dr. Dimmock noted that WGS reveals variants that are not relevant to the clinical question at hand. Although other technologies, such as array comparative genomic hybridization, also produce variants outside the clinical question, the scale of this event is much larger with WGS. The ethical question concerns what information should be returned to the patient. At Dr. Dimmock's institution—Children's Hospital of Wisconsin—the return of any or all genomic information was considered morally permissible and the decision of what should be returned remained at the discretion of the informed parent. As part of the consent process, parents are asked what data they would like returned. He also explained the key principles that guide the selection of cases for WGS—equity of access, standard clinical testing has not provided an answer, and WGS has a reasonably high likelihood to advance clinical decisionmaking. Dr. Dimmock noted that his institution is currently using WGS only for rare disorders, not common conditions. He also pointed out that WGS data in and of itself is not clinically or medically actionable until variant data are confirmed by independent tests.

Discussion. Dr. Paul Billings, Co-Chair of the Task Force, concluded the session by proposing the following WGS-related issues that require further action and should be brought to Secretary's attention: challenges in evaluating the clinical validity and clinical utility of WGS data, challenges in communicating WGS data to patients, a coverage and reimbursement paradigm that does not meet the needs of WGS testing, timely and appropriate reassessment of WGS data as research reveals new findings, and disparities and barriers to the equitable access to WGS technologies. He asked the Committee for revisions or additions to these concerns and to come to consensus on any guidance or recommendations that would address these concerns. The Committee discussed these proposed issues and suggested additional topics, such as informed consent, storage capacity for WGS data, analytical validity, testing children, comparative effectiveness. Because SACGHS had just begun to explore implications of WGS, the Committee decided a general recommendation would be more appropriate than specific recommendations in the letter to the Secretary. Revisions were made for discussion the following day.

## Public Awareness of the Genetic Information Nondiscrimination Act (GINA)

Juli Murphy-Bollinger, M.S., from the Genetics and Public Policy Center (GPPC) at Johns Hopkins University, shared findings about public awareness of GINA and attitudes towards genetic privacy, which were part of two GPPC studies that examined public opinion about a proposed U.S. prospective cohort study that would enroll 500,000 participants and collect DNA and other specimens. The first GPPC study began in 2006 and was completed in 2008, prior to GINA's enactment. The findings revealed that privacy of personal information was a general concern for participants (n=4,659), but only 34 percent of participants thought some information in medical records needed extra privacy protections. When asked what types of information needed extra protection, genetic testing results were not a top concern—92 percent of participants (n=1574) indicated that social security numbers needed extra protection compared

to 44 percent for genetic test results. In addition, fewer participants were concerned that data collected in the cohort study could be used against them than were concerned about privacy (37 percent vs. 91 percent). The second study, built upon the results of the first, began in 2009 and will continue until 2011. Data from 10 focus groups suggested that people think privacy no longer exists. Concerns related to privacy included insurance or employment discrimination, identity fraud, stigmatization, being cloned, and receiving unsolicited marketing materials. Most participants had never heard of GINA, and once they had, were not reassured by the protections it offers. Participants also believed it would be very difficult to control access to data.

Discussion. Dr. Teutsch asked the Committee if they believed issues surrounding GINA should be included in the letter to the Secretary. The Committee agreed not to include this topic.

## Clinical Utility and Comparative Effectiveness

SACGHS member, Dr. Marc Williams presented an update of the Committee's Clinical Utility (CU) and Comparative Effectiveness Research (CER) Task Force and reviewed the inventory of CER projects funded by the Department of Health and Human Services (HHS). He explained his search methods and then reviewed his findings, which included about 30 NIH-funded CER projects related to genomics and three projects funded by other HHS agencies. One project is to build a genome-enabled electronic medical record, which SACGHS has recognized as a critically important infrastructure need. Dr. Williams reviewed specific needs related to CU/CER that could be addressed in the letter to the Secretary and proposed the following six recommendations:

- Support adoption of recommendations from the American Health Information Community Personalized Medicine Workgroup
- Encourage incorporation of family history, and genetic and genomic information in CER studies for all 14 priority health conditions as appropriate
- Provide ongoing funding to support and expand development of systematic evidence-based recommendations by HHS-funded centers
- Increase visibility of family history, genetics, and genomics in the inventory and evaluation of CER studies t
- Direct available funds from the Agency for Healthcare Research and Quality (AHRQ) Unfunded Meritorious Applications program to expand development of systematic evidence-based recommendations by HHS-funded centers
- Encourage the Government Accountability Office to solicit a member with specific expertise in genomics for the Governing Board of the Patient-Centered Outcomes Research Institute (PCORI) and assure appointment of individuals with expertise in evidence-based genomics to the PCORI methodology committee

Discussion. The Committee revised the scope of the recommendations and narrowed them to five. Final revisions were made the following day.

## Closing Remarks

Dr. Teutsch discussed the agenda for the second day of the meeting, which included genomic data sharing and finalizing the letter to the Secretary. The meeting was then adjourned for the day.

## Wednesday, October 6, 2010

### Opening Remarks

Dr. Teutsch welcomed everyone to the second day of the SACGHS meeting, thanked the Committee for the progress made thus far, and summarized the day's agenda.

### Genomic Data Sharing

Dr. Charmaine Royal, Chair of the Genomic Data Sharing (GDS) Task Force introduced the session on perspectives of group risks and benefits related to GDS by reviewing the work to date on this topic and providing definitions and background information on the session's focus. She gave an overview of the four session speakers and central questions that would be addressed. The goals of the session were to: (1) explore issues related to the involvement of indigenous, racial, and ethnic groups in genomic research ad broad sharing of data for secondary research purposes, (2) explore best practices in the United States and other countries to address risks of group harms, (3) determine whether there are policy gaps that should be addressed, and (4) come to consensus regarding specific policy issues that should be related to group harms when genomic data are shared.

Perspectives of Indigenous People Regarding Participation in Genomic Research and Data Sharing

Rebecca Tsosie, J.D., Professor of Law, Distinguished Research Scholar, and Executive Director of the Indian Legal Program and the Sandra Day O'Connor College of Law in Arizona State University, cited historical wrong doings that have shaped the attitudes of many indigenous populations and challenges when attempting to obtain genetic-related information. She also stressed the importance of recognizing both cultural and individual harms involved in this process. Several points to consider when creating policies surrounding indigenous people included the current legal structure relevant to genomic data collection, tangible and intangible property rights, privacy interests and concerns, and cultural exploitation.

Perspectives of Ethnic and Racial Groups on Genomic Research and Data Sharing

Vence Bonham, J.D., Senior Advisor to the Director on Societal Implications of Genomics and Head of the Education and Community Involvement Branch, National Human Genome Research Institute, discussed research studies and a national public forum that have yielded both qualitative and quantitative data relevant to addressing the perspectives of ethnic and racial groups in relation to genomic research and data sharing. Perceived risks of participation in genetic and genomic research included ethics violations, unequal treatment in health care system, stereotyping, and invasion of privacy. Common benefits included the improvement of the overall health of families and communities as well as a better understanding of specific diseases that burden these communities. The research also demonstrated an overall willingness to share genomic data. Dr. Bonham also briefly covered the U.S. Public Health Services Syphilis Study (i.e., the Tuskegee Syphilis Study), and stressed the importance of addressing the distrust of government research in a way that is appropriate and respectful to move forward and assure that such deceptive practices will not occur again. He closed by posing the idea of tailoring policies to fit the needs of individual communities and ethnic and racial groups and stressed the importance maintaining an open dialogue and upholding a moral contract between researcher and participant.

Canadian Policies Involving the Genomic Research and Data Sharing of Aboriginal Peoples

Laura Arbour, M.D., Associate Professor of Medical Genetics at the University of British Columbia and Head of Medical Genetics with the Vancouver Island Health Authority, discussed issues related to data collection and use, such as research funding that did not address health disparities, commercial and academic exploitation, trust, and stigmatization. She noted in the last decade steps had been taken to build relationships between policymakers, researchers, and Aboriginal groups to determine the most culturally appropriate way to carry out biomedical genomic research of these peoples. Guidelines adopted by the Canadian Institute for Health Research in 2007 pertain to secondary use of data and samples, interpretation and acknowledgement of data, and knowledge dissemination. Dr. Arbour explained that Aboriginal partners are able to collaborate with researchers and have access to all DNA data. She finished by noting the success researchers have had thus far with this methodology.

Perspectives on U.S. Policies that Address the Involvement of Groups in Genomic Data Sharing

Morris Foster, Ph.D., Professor and Acting Chair for the Department of Anthropology at the University of Oklahoma, noted that social groups are not constituted based on biological characteristics; however, they tend to have nonrandom frequency distributions of biological characteristics, which are often are mistaken for biological definitions of group membership. In addition, all social groups are to some extent heterogeneous in viewpoint; however, group labels have been used as a monolithic representation of viewpoint. Thus far, societal concerns about group risks have outweighed group benefits. Dr. Foster addressed health disparities at the group level, noting that such disparities are a product of nonrandom social factors and nonrandom frequency distribution of biological factors. To ensure equal access and distribution of genetic-based diagnostics and therapeutics, Dr. Foster said it is important to pay more attention to social justice issues and collective clinical benefits (not just risks). In discussing group identities, he noted that identities that are legally and politically relevant may not be biologically or biomedically relevant. Using genotypic data rather than self-reported social identity appears more accurate for population stratification; however, there is a risk that genotypes could become surrogates for social and ancestral identities. He concluded with policy suggestions that included using social labels in association with biological data only when scientifically meaningful and disentangling legal and regulatory requirements for inclusiveness from scientific design and evaluation.

Discussion. Areas of discussion included the controversial use of genetic tests to determine Native American ancestry, the definition of ownership and cultural property rights of genomic data, whether resources spent on emerging genetic technologies are best serving ethnic and racial groups, how to convey the long-term general health benefits of genetic research, and the challenges of implementing policies specific to individual groups. Dr. Royal reviewed proposed recommendations for the final letter to the Secretary, and Committee members provided comments. Revised recommendations were reviewed later that day.

## Public Comment

Joann Boughman, Ph.D., Executive Vice President of the American Society of Human Genetics (ASHG) publicly thanked the Committee and its staff, on behalf of ASHG, for all of their time and dedication to human genetics interests. She stated that ASHG would have to find another venue for continued dialogue about genetic policy issues and noted that replacing SACGHS would be difficult.

## Concluding Business

Final Recommendations for the Report on Genetics Education and Training

After extensive discussion from SACGHS members and *ex officios*, the Committee approved the following recommendations for genetics education and training:

*Recommendation 1*
Evidence from the United States and abroad suggests inadequate genetics education of health care professionals as a significant factor limiting the integration of genetics into clinical care. Specific inadequacies include the amount and type of genetics content included in undergraduate professional school curricula and the small amount of genetics-related knowledge and skills of physicians, nurses, and other health professionals once they enter clinical practice. Modifications in medical, dental, nursing, public health, and pharmacy school curricula and in medical residency training programs are needed to ensure that health care professionals entering the workforce are well-trained in genetics.

1. Innovative approaches that coordinate the efforts of entities involved in health professional education and training are required to address these gaps. Therefore, HHS should convene a task force of stakeholders to identify:
   A. Outcomes-based education and training guidelines and models;
   B. Best practices for enhancing and expanding the content needed to prepare health care professionals for personalized genomic health care;
   C. Mechanisms to assure the incorporation of up-to-date genetic content in standards, certification, accreditation, electronic health records, and continuing education activities; and
   D. Funding sources for developing and promoting genetics education for relevant health care professionals.

*Recommendation 2*
The inherent diversity of the public health workforce makes it difficult to target educational efforts that are relevant across groups. A systematic effort is needed to evaluate the composition of the public health workforce with current job responsibilities related to genetics and genomics and to identify future priorities, such as the potential impact of affordable genomic analysis.

2. HHS and its public health agencies should:
   A. Assess the public health workforce to determine the number and type of public health providers with responsibilities in genetics and genomics and to ascertain current trends and future education and training needs;
   B. Identify and engage exemplary public health genomic programs to identify critical workforce information not captured in the assessment; and
   C. Using the results of these assessments and to address identified gaps, HHS should:
      - Support development of skills, competencies, and leadership in genetics and genomics that specifically address the identified needs; and
      - Based on these skills and competencies, fund the development and implementation of accessible educational programs and continuing education in genetics and genomics for the public health workforce.

*Recommendation 3*
Findings in the literature and SACGHS surveys indicate that health care professionals and public health providers serving underserved and underrepresented groups and populations face significant challenges.

3. To increase services and access to care in underserved communities, HHS should:

A. Identify existing effective educational models for health care professionals and public health providers in underserved communities;
B. Identify and support programs to increase the diversity and genetic competencies of the health care workforce serving underserved communities; and
C. Incentivize organizations and ensure that consumers and representatives of rural, minority, and underserved communities participate in the process of developing education and training models and materials. Assure that these materials are culturally and linguistically appropriate and tailored to the unique needs of these diverse communities.

*Recommendation 4*

With the vast increase in scientific knowledge stemming from genetics research, the development of new technologies, and the increase in direct-to-consumer genetic services, educational efforts are needed to translate this information to reach consumers of all literacy levels.

4. HHS should identify effective communication strategies for translating genetics knowledge into information that consumers and patients can use to make health decisions. Specifically, HHS should:
   A. Support multidisciplinary research that identifies effective methods of patient and consumer communication;
   B. Based on this research, and to reach diverse people and communities, HHS should develop educational programs that use a wide array of media and community-based learning and provide culturally and linguistically appropriate materials; and
   C. In collaboration with the Department of Education and the National Science Foundation, support the incorporation of genetics and genomics in K-12 education.

*Recommendation 5*

A significant amount of genetic-related information directed to consumers and patients exists in a variety of formats and from a number of sources, but the quality of the content is variable. Consumers have consistently expressed the desire for accessible, web-based genetic information that they can trust and consider provision of these resources as a role of the Federal Government.

5. HHS should create and maintain a state-of-the-art Internet portal to facilitate access to comprehensive, accessible, and trustworthy web-based genetic information and resources for consumers.

*Recommendation 6*

6. Because family health history tools are a potentially powerful asset for consumers and health care professionals to use in risk assessment and health promotion, HHS should:
   A. Support efforts to educate health care professionals, public health providers, and consumers about the importance of family health history;
   B. Promote research on how consumers and diverse communities use family history to make health care decisions and incorporate those research findings into consumer educational materials;
   C. Support the use of family history in clinical care through development of point-of-care educational materials and clinical decision support tools in electronic health records that utilize coded and computable family history, genetic, and genomic information; and
   D. Promote embedding educational materials in family history collection tools and personal health records directed to consumers and ensure for all by providing these tools in various formats.

SACGHS Letter to Secretary Sebelius

The Committee developed a final letter to the Secretary and discussed the language for the guiding principles as well as the inclusion of the need for public policy deliberations around the integration of genetics and genomics in public health efforts, a topic of importance the Committee had planned to explore in the near future. The Committee then reviewed the following revised recommendations:

*Affordable Whole Genome-Sequencing*

SACGHS Recommendation: Experts and stakeholders should be convened on a regular basis to advise policy makers on the efficient adoption and clinical use of whole-genome sequencing technologies to improve health outcomes.

*Genomic Data Sharing*

SACGHS Recommendation: A mechanism(s) is needed to address the issues surrounding the risks and benefits to indigenous, racial, and ethnic groups that participate in genetic and genomic research and data sharing. A federal entity or entities should be charged with the ongoing consideration of the unresolved issues identified here. For example, advisory bodies, such as The Presidential Commission for the Study of Bioethics Issues and the Secretary's Advisory Committee on Human Research Protections, or federal agencies, such as the Office for Human Research Protections and the National Institutes of Health (NIH) and its individual institutes, could be assigned to take up these important matters.

*Clinical Utility and Comparative Effectiveness*

SACGHS Recommendations: Based on these gaps and a review of the priorities defined by the Institute of Medicine (IOM) and Federal Coordinating Council for Comparative Effectiveness Research (FCCCER) and an analysis of CER projects funded through September 30, 2010, the steps below should be taken.

- In order to achieve the goals of health care reform, the administration and Congress have invested significant resources in CER. Family history (FH), genetic, and genomic information are critically important factors to consider if the results of CER studies are to yield fully valid information. Further research is needed to ensure the appropriate translation of genomics into health care. Necessary programmatic direction is needed to ensure that:
    - The initiative "Identification of New and Emerging Issues for CER (AHRQ-10-10003)" includes a focus on FH, genetic, and genomic issues;
    - At least one of the eight Centers charged with identifying evidence gaps focuses on issues relating to CER and clinical utility of FH, genetic, and genomics in health care;
    - The inventory of CER that will be carried out (Solicitation Number: 10OS32990) explicitly collects and includes information related to the use of FH, genetics, and genomics in all inventoried projects;
    - The evaluation of CER studies to be carried out under the initiative "Evaluation and Impact Assessment of ARRA CER (Solicitation Number: 10-233-SOL-00191)" describes the extent, if any, to which the funded studies included FH, genetic, genomic information and assesses the impact of the decision to include or exclude such information;
    - The Comprehensive Informatics Framework for CER Dissemination supports the use of FH, genetics, and genomics; and
    - FH, genetic, and genomic issues are given priority consideration as funds become available (e.g., through the Agency for Healthcare Research and Quality's Unfunded

Meritorious Applications program), particularly studies of the translation of personalized medicine into clinical practice.

- The development and use of fully functional EHRs is another key element to health care reform. Current informatics systems and EHRs are not capable of capturing FH, genetic, and genomic information in a coded computable fashion. This deficiency will impede CER studies and postmarket data collection for conditions where these data are critical. It will also affect the inclusion of point-of-care educational resources for clinical decision support. As such, the Office of the National Coordinator of Health Information Technology should be directed to explore options to facilitate the development of EHRs capable of handling FH, genetic, and genomic information and clinical decision support systems.

- The reform of the health care system is dependent on the development of evidence of best practices. While HHS agencies are supporting some efforts to develop evidence-based recommendations for genetics and genomics (e.g., the Evaluation of Genomic Applications in Practice and Prevention at the Centers for Disease Control and Prevention), HHS should be dedicating additional resources to expand the development of systematic evidence-based recommendations.

- Evidence-based genomics is critically important in ensuring that CER studies develop and achieve meaningful comparative effectiveness data. As such, individuals with specific expertise in evidence-based genomics should be nominated for membership on the methodology committee of the Patient-Centered Outcomes Research Institute.

Discussion. The Committee reviewed and approved the letter and voted unanimously to send a copy edited version forward to the Secretary.

## Presentation of Awards and Appreciation

At the conclusion of the meeting Dr. Francis Collins, NIH Director, joined the Committee to thank SACGHS members and *ex officios* for their work, note the highlights and achievements of SACGHS, and present the members with certificates of appreciation.

## Closing Remarks

Dr. Teutsch thanked Dr. Collins, the Committee members, *ex officios*, and staff.

## Adjournment

The meeting was adjourned at 3:20 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., M.P.H.

Sarah Carr