DEPARTMENT OF HEALTH AND HUMAN SERVICES

Twentieth Meeting of the
SECRETARY'S ADVISORY COMMITTEE ON
GENETICS, HEALTH, AND SOCIETY
October 8-9, 2009

Meeting Summary

Park Hyatt Hotel
Washington, DC

Prepared by the Office of Biotechnology Activities
National Institutes of Health

Participants

Committee Members Present

Steven Teutsch, M.D., M.P.H., Chair
Mara Aspinall, M.B.A. (by teleconference)
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.
Julio Licinio, M.D.
Barbara Burns McGrath, R.N., Ph.D.
Samuel R. Nussbaum, M.D.
Charmaine Dawn Marie Royal, Ph.D.
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)
Alan E. Guttmacher, M.D. (HHS/NIH/National Human Genome Research Institute)
Adam B. Kanis, Lt. Col., M.D., Ph.D. (Medical Corps, U.S. Army, Department of Defense)
Muin Khoury, M.D., Ph.D. (HHS/Centers for Disease Control and Prevention/Director, National Office of Public Health Genomics)
Kerry Leibig, J.D., on behalf of Stuart Ishimaru, J.D. (Equal Employment Opportunity Commission)
Elizabeth Mansfield, Ph.D. (HHS/Food and Drug Administration)
Douglas Olsen, on behalf of Ellen Fox, M.D. (Department of Veterans Affairs)
Iliana Peters, J.D., L.L.M., on behalf of Robinsue Frohboese, J.D., Ph.D. (HHS/Office for Civil Rights)
Gurvaneet Randhawa, M.D., MPH (HHS/Agency for Healthcare Research and Quality)
Jeffrey Roche, M.D. (HHS/CMS/Office of Clinical Standards and Quality)
Laura L. Rodriguez, Ph.D., on behalf of Alan E. Guttmacher, M.D. (HHS/NIH/National Human Genome Research Institute)
Amy Turner, J.D., on behalf of Phyllis Borzi, J.D., M.A. (Department of Labor)
Jennifer Weisman, Ph.D., on behalf of Robinsue Frohboese, J.D., Ph.D. (HHS/Office for Civil Rights)

SACGHS Staff

Sarah Carr, Executive Secretary, NIH Office of Biotechnology Activities
Kathryn M. Camp, M.S, RD
Tara Hurd Faunteroy
Symma Finn, Ph.D.
Cathy Fomous, Ph.D.

Darren Greninger, J.D.
Linda Silversmith, Ph.D., Consultant
Marianne Tshihamba, The Dixon Group

Special Guests/Consultants

Gregory Downing, D.O., Ph.D. (HHS/Office of the Secretary) (in person & by teleconference)
Kevin T. FitzGerald, S.J., Ph.D., Ph.D. (former SACGHS member) (in person & by teleconference)
Sandra Howard (HHS/Office of the Assistant Secretary for Planning and Evaluation)
Penny Keller (CMS)
Michele Lloyd-Puryear (Secretary's Advisory Committee on Heritable Disorders in Newborns and Children/Executive Secretary)

Speakers

Sylvia Au, M.S., CGC (SACGHS)
Robinsue Frohboese, J.D., Ph.D. (HHS/Office for Civil Rights)
Christina Heide, J.D. (HHS/OCR)
Kerry Leibig, J.D. (Equal Employment Opportunity Commission/Office of Legal Counsel)
James Mayhew, J.D. (HHS/CMS/Division of Private Health Insurance)
Barbara Burns McGrath, Ph.D., RN, (SACGHS)
Charmaine Royal, Ph.D. (SACGHS)
Amy Turner, J.D. (Department of Labor/Employee Benefits Security Administration)
Russ Weinheimer, J.D. (Dept. of Treasury/Internal Revenue Service)

Public Commenters

Tom DiLenge, J.D. (Biotechnology Industry Organization)
Michael Henry (Athena Diagnostics)
Jennifer Leib, Sc.M. (Association for Molecular Pathology)
Amy M. Miller, Ph.D. (Personalized Medicine Coalition)
Lisa Schlager, (Facing Our Risk of Cancer Empowered)
Susan Poland (Kennedy Institute of Ethics)
Michael Remington, J.D. (Wisconsin Alumni Research Foundation)
Luisel Ricks-Santi, Ph.D. (National Human Genome Center)
Ted Rumel (Association of University Technology Managers)
Fay Shamanski, Ph.D. (College of American Pathologists)

October 8, 2009

Opening Remarks

Dr. Steven Teutsch, Chair of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), welcomed attendees and webcast participants to the Committee's 20th meeting. He reviewed the agenda and reported on the following events:

- On September 14, Dr. Teutsch meet with Dr. Francis Collins—the recently appointed director of the National Institutes of Health (NIH) and a former ex-officio member of SACGHS–who asked that the Committee consider (a) implications of the affordable genome and (b) publishing a paper highlighting prior SACGHS recommendations to help gain recognition for the Committee's work.
- In September, SACGHS's letter on genetics-related priorities regarding health care reform was transmitted to Health and Human Services Secretary Kathleen Sebelius.
- The American Recovery and Reinvestment Act (ARRA) makes available funding for a number of grants for comparative effectiveness research (CER) related to genomics. NIH has issued $360 million in ARRA grants to support CER, and several of these grants are focused on genomics.
- In August, the Federal Trade Commission took action on questionable claims by two companies—Genelex Corporation and Sciona, Inc.—that were marketing direct-to-consumer (DTC) nutrigenetic tests and services, and both have now stopped marketing the products.
- In August, the NIH held a state-of-the science conference that concluded substantial research is needed before a systematically collected family history for common diseases can become an evidence-based tool in primary care.
- The Centers for Disease Control and Prevention (CDC) and NIH have announced a new initiative called the Genomic Applications in Practice and Prevention Network (GAPPNeT) The aim of this initiative is to accelerate and streamline effective and responsible use of validated genomic knowledge and applications in clinical and public health practices.

Dr. Teutsch also introduced Dr. Symma Finn, a new staff member of the Office of Biotechnology Activities who will assist SACGHS. Ms. Sarah Carr, SACGHS Executive Secretary, reviewed relevant conflict-of-interest and privacy rules.

Genetics Information Nondiscrimination Act (GINA)–Update on Implementation

Title I—Genetic Nondiscrimination in Health Insurance

Employment-based Group Market Protection. Amy Turner, Department of Labor (DOL), and Russ Weinheimer, Internal Revenue Service (IRS), spoke about regulations for GINA, Title I. Ms. Turner explained that sections 101-104 in GINA address nondiscrimination in health coverage and will be administered by the Centers for Medicare & Medicaid Services (CMS), IRS and DOL collaboratively with the States; section 105 addresses privacy, a responsibility of the Office for Civil Rights (OCR). Title II includes responsibilities for the Equal Employment Opportunities Commission (EEOC). The interim final rules implementing Title I in GINA were published on October 7, 2009. Ms. Turner noted that a plain-English summary, press release, facts sheets, and information on the regulations were available on the DOL website (www.dol.gov/ebsa).

The provisions in GINA for group health insurance coverage (or group market) build on requirements of the Healthcare Insurance Portability and Accountability Act of 1996 (HIPAA). HIPAA prohibits a group

health plan or group health insurance issues from imposing a pre-existing condition exclusion based solely on genetic information or discriminating against an individual in eligibility, benefits, or premiums based on genetic information. For example, individuals with a mutation in the BRCA1 or BRCA2 gene are predisposed to developing breast and/or ovarian cancer, but they cannot have a pre-existing condition exclusion imposed upon them based solely on that genetic predisposition, in the absence of a diagnosis of disease.

Mr. Weinheimer explained that GINA provides additional protections to HIPAA through three substantive rules. In brief, plans and insurance companies cannot: (1) discriminate on the basis of the group rate (i.e., a group cannot be charged a higher rate based on genetic information); (2) request or require an individual to undergo a genetic test (with certain exceptions for providing medical care, genetic testing to determine the appropriateness of particular treatments if the plan is going to pay for that treatment, and research); and (3) require or purchase genetic information for underwriting purposes prior to enrollment. Ms. Turner noted that GINA has a broad definition of "underwriting purposes." The statutory definition states that any change in eligibility, benefits, or premiums is an underwriting purpose.

GINA's rules apply if two or more current employees participate on day one of the plan year, and the rules also apply to plans for retired employees. Under GINA, the IRS can impose an excise tax for noncompliance, and DOL can impose a civil monetary penalty against the plan administrator or insurance companies and impose an excise tax against an employment-based plan not in compliance with GINA. Mr. Mayhew explained that state and local plans that are self-funded can opt out of HIPAA provisions annually; however, all state and local plans are required to comply with GINA.

Discussion. When asked if a health plan could single out people with a family history for a particular condition such as diabetes and offer incentives to participate in a health and wellness program, Mr. Weinheimer clarified that a plan cannot require genetic information, including family medical history, as a condition for any benefit, nor can it provide an incentive (e.g., cash, reduced co-payments) in exchange for genetic information. A plan can ask an individual to volunteer genetic information but cannot tie any benefits or penalties to that request. SACGHS members responding to this explanation expressed concerns that the inability to assess health risks based on family history could lead to higher health care costs and other unintended consequences (e.g., preventive activities related to chronic illness). The speakers indicated that the regulations just follow the law. Ms. Turner noted that a wellness exception is included in the Title II employment provision of GINA, but not into Title I.

Individual insurance market provisions. James Mayhew, CMS director of the Division of Private Health Insurance, indicated that GINA is groundbreaking in providing protections to individuals who purchase their own health care insurance. Provisions for the individual health insurance market parallel those for group markets. A company insuring an individual cannot determine eligibility, impose pre-existing condition exclusions, or rate the premium based on genetic information, including family history. Also, GINA prohibits issuers of insurance for individuals from requesting or requiring genetic tests. Furthermore, GINA provides protections when individuals apply for coverage and fill out a release so the insurance company can obtain their medical records. GINA requires insurance companies to include a disclaimer in requests to health providers stating that genetic information, including family history, should be removed from copies of the medical records that are sent to the insurer. CMS provides insurance companies with specific language for the disclaimer.

Regarding enforcement of GINA provisions for the individual market, states will be the primary enforcement authority through the state's Department of Insurance. CMS has the authority to step in if a state substantially fails to enforce any HIPAA provision, including GINA.

Mr. Mayhew continued with a discussion of Section 104 of GINA, which addresses Medicare Supplemental Insurance policies, also known as Medigap. GINA prohibits MediGap insurance companies from discriminating on the basis of genetic information in rating the premiums, determining eligibility, or imposing pre-existing condition exclusions, and it also prohibits requirements for genetic tests and collecting family history. Although these Medigap requirements are not addressed in the GINA regulations, states are required as of July 1, 2009, to adopt National Association of Insurance Company (NAIC) model regulations that cover these policies. If a state does not adopt the regulations, then CMS can step in and regulate the MediGap policies in that particular state.

Privacy and confidentiality of genetic information. Robinsue Frohboese, OCR Principal Deputy Director, and Christine Heide, OCR senior health information privacy policy specialist, explained that GINA, Title I, Section 105 amends the HIPAA Title II Privacy Rule by clarifying that protected health information includes genetic information and by prohibiting certain health plans from using or disclosing genetic information for underwriting purposes. The Privacy Rule covers several categories of health plans such as group health plans, health insurance issuers, health maintenance organizations (HMOs), and MediGap issuers as well as other categories such as certain public benefit plans (e.g., Medicare, state Medicaid agencies, the military and veterans health programs, the Indian Health Service program), long-term care insurers (excluding nursing home fixed indemnity policies), and certain limited vision and dental plans that are separate from group health plans. Also, under the Privacy Rule, an individual has the right to receive a Notice of Privacy Practices for protected health information from covered entities such as health plans. For health plans that use or disclose protected health information for underwriting purposes, the proposed regulations would require that the plans amend their Notice of Privacy Practices to state explicitly that they may not use or disclose an individual's genetic information for those purposes.

Discussion. When asked if all genetic information is considered medical information, Ms. Heide stated that the HHS has always considered genetic information as protected health information. Not all health information, however, is protected by the Privacy Rule. To be protected, the information must be maintained by a HIPAA covered entity (e.g., a health plan or HIPAA-covered health care provider) and must be individually identifiable. Another inquiry concerned whether information gathered through direct-to-consumer (DTC) genetic testing is protected health information. Ms. Heide explained that companies providing DTC genetic testing may not be HIPAA covered entities, but if they were, the companies would be subject to the Privacy Rule. The Committee also asked if the identifiability of DNA samples was considered in the definition of identifiable health information. Ms. Heide replied that HHS has not yet addressed the issue of a genetic sequence as individually identifiable information.

Title II—Prohibiting Employment Discrimination on the Basis of Genetic Information

Kerry Leibig, senior attorney advisor in the EEOC Office of Legal Counsel, addressed the Title II prohibition of employment discrimination based on genetic information and topics in the proposed regulations that received the most public comments. She noted that Title II will be effective on November 21, 2009, and the final regulations will become part of 29 CFR 1635. Ms. Leibig explained that Title II has three basic rules that (1) prohibit the use of genetic information to make employment decisions, (2) restrict the acquisition of genetic information by employers and other entities covered by GINA (with six exceptions), and (3) require a basic confidentiality rule (with six limited exceptions similar to the exceptions under the American with Disabilities Act). Genetic information is defined as including genetic test results of individuals and their family members (extending to fourth-degree relatives) as well as family medical histories. Public commenters requested that more examples of genetic tests be included in the regulation. Genetic information also includes the request for or receipt of genetic services (e.g., genetic counseling) by an individual or a family member and the genetic information of a fetus carried by an individual or family member or of an embryo legally held by the individual or family member using assisted reproductive technology.

Ms. Leibig continued with an explanation of the narrow exceptions to the rule that restricts the acquisition of genetic information by employers and other entities covered by GINA There is no liability for (1) inadvertent acquisition of information (e.g., in response to a general inquiry such as "How are you?"), (2) genetic information obtained through employer-sponsored health services that meet specific requirements (e.g., the service and voluntary and the employer gets the information only in aggregate form), (3) family medical histories acquired when individuals request leave under the Family Medical Leave Act and the information is kept in a separate, confidential medical file, (4) genetic information obtained from a public source (e.g., an obituary), (5) genetic information acquired from genetic monitoring programs (e.g., job site exposure monitoring programs) that meet certain requirements (e.g., the covered entity receives only aggregate information), and (6) employers that engage in DNA testing for law enforcement purposes or to identify human remains and request genetic information from their employees for the purpose of quality control or analysis of DNA markers. Several public commenters requested additional examples to clarify these exceptions. In particular, EEOC received a number of comments about whether social networking sites (e.g, Facebook) and other sort of Internet-based information are considered public sources of information. Ms. Leibig expected these comments would be addressed in the final regulations.

Ms. Leibig also noted that there is a firewall between GINA Title I and Title II to prevent double liability. The intent of the firewall is to ensure that health plan or issuer provisions or actions are addressed and remedied through the Public Health Service Act, Employee Retirement Income Security Act, or the Internal Revenue Code, while actions taken by the employer are remedied through GINA Title II.

Discussion. A Committee member asked if Title I and Title II definitions are consistent. Ms. Leibig noted that the agencies coordinated their efforts regarding definitions and there were only minor differences. A question was raised about the use of genetic information to prevent harm. If an employee had a genetic variant that increased the risk of adverse outcome upon exposure to a particular substance that is likely to be encountered in the work place, the employer may not know about this risk. If there is a monitoring program in place, the employer would receive aggregate information, genetic information for individuals. Could the employee sue if he/she were harmed from the exposure? Ms. Leibig replied that the proposed rule did not address this situation, but the final rule might speak to it.

Public Comments

Association for Molecular Pathology (AMP). Speaking on behalf of AMP, Jennifer Leib commented on a number of topics on the SACGHS meeting agenda. First, with regard to gene patents, she noted that AMP opposes the patenting of DNA and is a lead plaintiff in lawsuit by the American Civil Liberties Union challenging various DNA patents held by Myriad Genetics. Second, with regard to DTC genetic tests, Ms. Leib noted that these tests have the potential to do harm, mislead consumers about the significance of the results, and promote the purchase of products not proven to be medically useful. She proposed that SACGHS review the practices of companies that market these tests and contact consumers who have used them as a means of assessing the benefits and harms of the tests. Third and finally, Ms. Leib asked SACGHS to join AMP in working to ensure that now that GINA protections are not weakened or otherwise undermined as implementation moves forward.

Biotechnology Industry Organization (BIO). The remarks of Tom DiLenge, BIO General Counsel and Vice President for Legal & Intellectual Property, focused on BIO's differences with SACGHS concerning its draft report on gene patents and licensing. Among the points made by Mr. DiLenge were the following: the SACGHS case studies do not support the report's conclusion that patents and licensing cause harms in terms of patient access to genetic tests; patents and exclusive licenses may sometimes be needed for the development of a genetic test; and getting major insurers to pay for genetic testing is the best way to improve access. He also noted that the proposed policy options would undermine the enforceability of patent rights, would chill patent private sector investment, and throw a monkey wrench

into the very successful Bayh-Dole Act, which has fostered technology transfer, spurring economic growth in all of the states—innovation that has benefited patients worldwide. Mr. Dilenge also urged the Committee to consult with the U.S. Trade Representative's Office as he was concerned that some of the report's policy options would violate international obligations under the Tripps Agreement.

College of American Pathologists (CAP). Dr. Fay Shamanski, CAP Assistant Director of Public Health and Scientific Affairs, reaffirmed CAP's view that human health-related gene patents have an inhibitory effect on pathologists and other laboratory physicians' ability to practice medicine, which in turn affects patients' access to important medical testing services. She elaborated that, since 2000, CAP has opposed gene patents and noted that CAP supports the legislative changes proposed by the Committee. Dr. Shamanski also indicated that for pathologists, clinical need—not intellectual property—spurs medical innovation. She asked that the committee recognize the vast amounts of innovation occurring through the work of pathologists in clinical laboratories, who have introduced and improved the majority of molecular tests largely without patent protection.

Athena Diagnostics. Michael Henry, Athena Vice President for Business Development, described Athena as a clinical diagnostics laboratory that offers more than 200 genetic tests for patient care, including some for rare diseases. Mr. Henry remarked that SACGHS has not had sufficient discussion about the benefits of exclusive licensing of genetic tests and the negatives of nonexclusive licensing. He used genetic testing for warfarin metabolism as an example of the negatives of nonexclusinve licensing. He stated that the warfarin metabolism test is one of the most widely known but least-ordered tests in the United States, even though the FDA recommends the test for all patients started on warfarin. Mr. Henry explained that the test's patent owners decided to nonexclusively license the test, and about 10 laboratories—including Athena Diagnostics—began to offer it two or three years ago. However, Athena Diagnostics discontinued testing because no one ordered the test. He said if there had been an exclusive license for the warfarin genetic test, that license holder would have invested in widespread marketing to educate physicians about the test. In concluding, Mr. Henry noted that the report's policy options are inconsistent with its findings. For example, one of the preliminary conclusions states: "Thus far, patents covering genetic tests and related licensing practices do not appear to be causing wide or lasting barriers to patient or clinical access." However, the policy options propose a number of mechanisms to promote access to genetic tests. He said that it is critical to the integrity of the Committee's credibility and success that the policy options reflect the findings.

Wisconsin Alumni Research Foundation (WARF). Speaking on behalf of WARF, Michael Remington, counsel for WARF, made a number of remarks about the SACGHS gene patents report. He noted that WARF would like to associate itself with the comments of the Council on Governmental Relations and the Association of American Universities about the report, specifically (1) the lack of support for policy options, (2) the lack of understanding that licensing is a complex process requiring substantial flexibility, and (3) too much focus on regulation without consideration of possible incentives. He also remarked that the evidence of patent thickets was fairly meager and the success of the Bayh-Dole Act was given short shrift.

Gene Patents and Licensing Practices

While introducing Dr. James Evans, Chair of the SACGHS Task Force on Gene Patents and Licensing Practices, Dr. Teutsch spoke of the extraordinary work of the Task Force, noting the depth of its investigation.

Report overview, summary of public comments, session goals. Dr. Evans began his presentation by reviewing the history of SACGHS' exploration of gene patents and their impact on patient access to

genetic tests, noting that in March 2009 SACGHS had released a public consultation draft report on the subject.

He then turned to a discussion of the public comments that were received in response to the public consultation draft report. He reported that 77 public comments were received and that among those providing comments were professional associations, technology transfer offices and professionals, academics, health and disease advocacy groups, industry organizations, life science companies, health care providers, commercial laboratories, and private citizens. Dr. Evans stated that the Task Force, during conference calls, reviewed all the public comments.

Dr. Evans noted that the public comments revealed that patient access issues identified in the case studies were not isolated problems. He added that some comments called for more discussion in the report of the impact of patents on whole genome sequencing, multiplex testing, and other emerging testing innovations, while other comments offered opinions and perspectives on the impact of patents on test development. The Task Force kept in mind these comments, Dr. Evans explained, when it revised the report's preliminary conclusions. Dr. Evans next highlighted the report's main points and presented its eight proposed recommendations.

Before turning to a Committee discussion of the proposed recommendations, Dr. Evans made several remarks. He indicated that while a large majority of the Task Force were in favor of the draft final report and the proposed recommendations, two members dissented—Mara Aspinall, one of the Task Force's five SACGHS members, and Brian Stanton, one of the Task Force's *ad hoc* members. Dr. Evans also disagreed with two particular criticisms of the report—that the Task Force was not charged with looking at the benefits of patents on access to genetic tests and that the Task Force did not find significant harms from patents on patient access to genetic tests. Dr. Evans next noted that in the Task Force's study of gene patents and genetic tests, he was struck by the almost complete lack of evidence of the need for patents in the development of genetic diagnostic tests. He cited examples in which tests were developed without patent rights but then shut down by those holding patent rights. He then explained that the recommendations, in his view, are not dramatic and are narrowly tailored to affect diagnostics but not therapeutics. Finally, Dr. Evans observed that the recommendations are also meant to address the threat that sole-source providers pose to the quality of tests.

General discussion. Dr. Khoury wondered if the principles that the Task Force is espousing for diagnostic genetic tests might be applied to other types of diagnostic testing, a possibility that public commenter Michael Remington noted as well. In response, Dr. Evans speculated that other types of diagnostic tests could be very different in types of upfront costs and require the patent incentive, so it might be the case that it would not be appropriate to extend the recommended actions to other diagnostics. He added that these "narrow" recommendations for the analysis of a DNA sequence would not interfere with patenting a major new molecular method such as polymerase chain reaction. When Dr. Khoury asked which kinds of patents in the realm of genetic testing are the most problematic, Dr. Evans answered that association claims probably present the most difficulties.

Ms. Au next pointed out that Medicaid decisions on coverage and reimbursement for genetic testing are usually made at the state level, that laboratories certainly do not want to have to deal with all 50 states, and that states are not going to contract with a faraway laboratory unless they have a serious public health problem requiring that test.

In response, Ms. Walcoff wondered if legislation could require state Medicaid programs to contract with sole-source laboratories. Jeffrey Roche, *ex offcio* representing CMS, replied that he was not sure whether CMS has the authority to impose such a requirement administratively. Dr. Evans responded that such a requirement would deal with only one problem and would not address concerns about the quality of

testing from a sole-source provider because of the absence of proficiency testing through sample sharing. He also noted that addressing Medicaid issues alone would not solve the problems that patents pose to the development of future approaches to testing. Dr. Ferreira-Gonzalez added that from her 18 years of experience with proficiency testing, one learns a lot more about processes from dealing with multiple laboratories than with a sole source. Drs. Billings and Williams cautioned that not having an independent ability to verify testing does not mean that one can claim the testing is of lower quality.

Noting the difficulty of garnering support from an Administration for legislative changes, Ms. Walcoff asked again if administrative, rather than legislative, changes could be the solution, both to the access problems and the concerns about the quality of testing. Dr. Evans responded that the Task Force had looked into this approach and concluded that statutory changes are necessary and, though difficult to achieve, not far-fetched when the public has indicated a discomfort with the patenting of genes.

Dr. Williams reminded everyone that the SACGHS charter tasks the Committee with addressing disparities in access, so the disparity in access to sole provider genetic tests between Medicaid patients and others should be a definite concern of the Committee's. He then noted a potential problem with sole providers of genetic testing that had not yet been discussed. He elaborated that tumor samples are often too small for testing of various markers, and the need to divide them up and send them to multiple sole providers for different tests would only compound this problem. He also spoke about patent thickets and the evidence that laboratorians are not reporting medically significant results because of the concerns about infringing on other patents. Dr. Williams also wondered if it was misleading to emphasize the Committee's overwhelming support for the recommendations, given that those with an interest in patenting are underrepresented on the Committee.

After speaking of her respect for the Committee and its process, which made sure that dissent was heard, Ms. Aspinall declared that the Task Force was charged only with examining access to genetic tests and disparities, not with looking at the effect of patents on the quality of genetic tests. She also objected to a blanket statement that the quality of testing provided by sole-source laboratories will always be inferior to the quality of testing that could be achieved if multiple laboratories were involved. She also disagreed with the assertion that the costs of developing diagnostics are low. Regarding the report itself, she said that it does not adequately represent the public comments from some large academic institutions that support access and dealing with disparities but state that the recommendations go too far. Ms. Aspinall then asked that the report include requests that HHS make it easier for companies to provide testing to needy patients.

Dr. Mansfield stated that it was her observation that there was no input from the diagnostics industry on the report. She also cautioned that as personalized medicine grows and genetic tests become required before use of various drugs, FDA will require those genetic tests to obtain premarket approval, which involves a process that companies may be unwilling to go through without exclusive rights. In reply, Dr. Ferreira-Gonzalez pointed out that laboratories without exclusive rights are pursuing FDA approval for the *KRAS* gene test. Dr. Mansfield then stated that she thinks SACGHS nevertheless should analyze whether lack of exclusive rights would discourage most diagnostic companies from pursuing FDA approval.

Ms. Dreyfuss made a number of observations. First, she argued that the Committee, in considering public comments from university technology transfer offices, should be mindful that universities only license out, so universities do not see the problems that can arise from trying to obtain license rights to patents, and, if they did, they might have a different position. She also pointed out that patents are only one way to stimulate innovation and that competition is another way. She also questioned the suggestion from others that the proposed statutory changes represent a drastic change. She observed that the research exemption the Committee was proposing existed until about 10 years ago, when it was ended by a court

decision. She then said that what distinguishes DNA patents from other patents is that they cannot be invented around and that the impossibility of inventing around a patent is a new development in the history of patents. Another change, Ms. Dreyfuss explained, is that while the patent system used to be narrowly directed at technological arts, it has expanded in the last 10 years so that it covers business methods and methods of doctors treating their patients. As a result, it seems entirely appropriate, Ms. Dreyfuss said, for the Committee to examine the social impact of these new developments. Finally, it also is now easy to get a patent without being particularly inventive, Ms. Dreyfuss stated, while in the past, when patents were not granted as easily, they encouraged greater advances.

Before the Committee took a break, Dr. Evans addressed clarified that diagnostic manufacturers were represented on the Task Force.

Discussion of the draft final recommendations. Recommendation 1, part 1. Noting that the first three recommendations are probably the most contentious, Dr. Evans encouraged everyone to speak up.

Recommendation 1, Part A:

> The Secretary of Health and Human Services should support and work with the Secretary of Commerce to promote the following statutory changes: (1) The creation of an exemption from liability for infringement of patent claims on genes for anyone making, using, ordering, offering for sale, or selling a test developed under the patent for patient care purposes.

Dr. Evans explained the legislative change this proposed recommendation called for is meant to address patient access problems and quality concerns and to enable laboratories and test kit makers to offer multiplex tests and other innovations.

Dr. Laura Rodriguez of NHGRI then asked why the Task Force proposed an exemption that would be available to anyone. Dr. Evans answered that Dr. Winn-Deen had requested that the exemption apply to test kit makers and not only laboratories that make laboratory-developed tests.

Dr. Dale next spoke in support of the recommendation, expressing his view that his patients should have access to their genomic information without obstacles as a part of general access to care. Ms. Aspinall then inquired about whether a Patent and Trademark Office (PTO) official was present to provide the PTO's viewpoint on the proposed recommendation, and Dr. Evans replied that a PTO representative had joined in every Task Force conference call. Ms. Dreyfuss added that the proposed legislative change was not PTO's concern, as they focus on patentability and not post-patent issues such as infringement or exemptions from liability. Ms. Aspinall then asked if Dr. Debra Leonard, former SACGHS member and former Task Force Chair, had had any objection to this recommendation. Dr. Evans stated that Dr. Leonard was in support of the proposed recommendation. Ms. Carr then clarified that agency experts, such as the PTO representative, participate in task forces as agency experts and that their role is not to offer their opinions on the advisability of particular recommendations.

When Dr. Billings asked about the language of the recommendation, Dr. Evans explained that one would still be able to patent a test kit if the proposed legislative change were made.

Ms. Darien next suggested that since the purpose of the proposed recommendation is to improve patient care, there should be no objections to the proposed recommendation. In response, Dr. Evans said that those who object to the recommendation believe the legal change will harm patient care. He continued that he does not agree with the assessment that the legal change will cause harms, and he stated that he has explained the reasons for his belief many times before.

By a vote of 11 to 3, SACGHS members approved the recommendation.

Recommendation 1, Part B. Dr. Evans noted that the second part of recommendation 1 is about research exemptions that would enable test developers to conduct research to design new tests. It proposed

> The creation of an exemption from patent infringement liability for those who use patent-protected genes in the pursuit of research. Related health care and research entities also should be covered by this exemption.

Dr. Rodriguez requested a definition of research, and Dr. Williams proposed using already known definitions of "related health care and research entities" as well as of research. Dr. Evans indicated that the Task Force had not wanted to restrict the exemption to only those parties doing basic, clinical, or translational research. He later committed to including definitions in recommendation 1, part 2.

Recommendation 2.

> The Secretary should use her powers to discourage the seeking, the granting, and the invoking of simple association patent claims; it is the Committee's position that these claims represent basic laws of nature that cannot be invented around.

Dr. Evans indicated that this recommendation would address association patents. He explained that although the Task Force had considered whether the exemptions proposed in recommendation 1 should apply to association patents as well as patent claims on genes, the Task Force ultimately decided to make a separate recommendation relating to association patents because it is an active area of debate. The recommendation calls for the HHS Secretary to discourage the seeking, the granting, and the invoking of simple association patent claims. Dr. Evans explained that the Task Force is using the word "simple" to refer to basic associations discovered between a gene and a disease and did not want to imply that complex associations could not be patented. Dr. Evans then asked if the recommendation should stand on its own or be folded into the first recommendation.

Dr. Williams responded that the word "simple" was not sufficiently defined in the report in a way that would permit the reader to distinguish between simple and complex associations. He also wondered if courts will actually consider what SACGHS is saying in rendering decisions that may bear upon association patents. In response to his first point, Ms. Dreyfuss wondered if SACGHS, instead of referring to simple associations, could refer to direct associations between a genotype and a phenotype. But Dr. Williams expressed doubt about this too, wondering what was meant by "direct." Ms. Walcoff suggested defining the meaning of "direct" by providing examples of direct associations since the recommendations would be interpreted by the HHS Secretary, rather than by a court.

Dr. Evans responded by noting that it is not unreasonable for SACGHS to state a position regarding association patents that may carry weight with the courts; he also indicated that the Task Force was aware that this recommendation was nebulous. Other SACGHS members then offered suggestions for potential wording changes to the recommendation. In the end, SACGHS members voted to drop the recommendation in favor of inserting a statement in the report saying substantially the same thing as the recommendation.

Recommendation 3.

> A. The Secretary should develop mechanisms to promote voluntary adherence to the principles reflected in NIH's Best Practices for the Licensing of Genomic Inventions; the Organisation for Economic Co-Operation and Development's (OECD) Guidelines for Licensing of Genetic

Inventions; the NIH Policy for Sharing of Data Obtained in NIH Supported or Conducted Genome-wide Association Studies; and In the Public Interest: Nine Points to Consider in Licensing University Technology. The Secretary of Health and Human Services should also advocate that professional organizations involved in intellectual property policy and practice in this area work together to build on those norms and practices as they relate to gene-based diagnostics by articulating more specific conditions under which exclusive licensing and nonexclusive licensing of uses relevant to genetic testing are appropriate. Professional societies should work cooperatively to forge consensus positions with respect to gene patenting and licensing policies.

B. The Secretary should encourage stakeholders (for example, industry, academic institutions, researchers, patients) to continue their work of developing a code of conduct that will enable broad access to such technologies.

Committee members discussed a number of ways in which the recommendation might be improved. Recognizing that the text of the recommendation would be revised further overnight, SACGHS members then voted to accept the recommendation, with 14 in favor and one abstention.

Recommendation 4.

A. The Secretary should encourage holders of patents associated with genetic tests and their licensees to make information about patent licenses readily available either by making the signed licenses publicly available or by disseminating information about their technology and licensing conditions, including any terms that pertain to the type of license, field of use, and the scope of technologies that are still available.

B. As a means to enhance public access to information about the licensing of patents related to gene-based diagnostics, the Secretary should direct NIH to amend its Best Practices for the Licensing of Genomic Inventions to encourage licensors and licensees to include in their license contracts a provision that allows each party to disclose information about its licenses (including such factors as type of license, field of use, and scope) in order to encourage next-generation innovation.

Dr. Evans noted that the recommendation has been objected to by some, and Ms. Aspinall stated that universities in public comments had opposed the recommendation because they did not want the financial terms of their license deals disclosed.

Dr. Williams wondered about this objection, however, noting that the actions the proposed recommendation called for would not appear to have the effect of requiring the disclosure of financial information. Dr. Ferreira-Gonzalez clarified that what SACGHS would be asking for would be the information on who has licensed what to whom and what field of use the license covers. Committee members then agreed that the language of the recommendation could be modified to make clear that it was not calling for the Secretary to take actions requiring the disclosure of financial information in licenses.

Recommendation 5.

The Secretary should establish an advisory board, which would be available to provide ongoing advice about the public health impact of gene patenting and licensing practices. This advisory board would also be available to receive any reports of problems in patient access to genetic tests

from the public and medical community. The board then could review new data collected on patient access and assess the extent to which access problems are occurring. One of the board's missions would also be to recommend what information should be systematically collected through iEdison so that iEdison can be used to research questions about licensing, including whether the licensing of genomic inventions has been conducted in accordance with NIH's Best Practices for the Licensing of Genomic Inventions. The advisory board also could provide input on the implementation of any future policy changes, including the other proposed recommendations in this report.

Dr. Evans explained the advisory board would be the dedicated contact group for reports of problems in patient access to genetic tests. During the subsequent discussion, Committee members decided to include within the recommendation the option that the new advisory body could be a standing committee of SACGHS. Committee members also decided that the text discussing the recommendation would indicate the need for interdepartmental membership on the board and a broad array of experts. The recommendation was then passed, with 13 members voting in favor of it, one voting against it, and one abstaining from the vote.

Recommendation 6.

The Secretary should encourage Federal agencies within the Department of Health and Human Services to undertake the following actions:

A. Federal agencies should promote wider adoption of the principles reflected in NIH's Best Practices for the Licensing of Genomic Inventions and the OECD Guidelines for Licensing of Genetic Inventions, both of which encourage limited use of exclusive licensing for genetic/genomic inventions.

B. Federal agencies should encourage wider use of the Nine Points to Consider in Licensing University Technology. Points two and nine, including their explanatory text, are particularly relevant for genetic tests. For example, the explanatory text under point two recognizes that "licenses should not hinder clinical research, professional education and training, use by public health authorities, independent validation of test results or quality verification and/or control."

C. Federal agencies should explore whether approaches to addressing patent thickets, including patent pools, clearinghouses, and cross-licensing agreements, could facilitate the development of multiplex tests or whole genome sequencing.

D. Federal agencies should provide more detailed guidance regarding the licensing of patents associated with genetic tests. In particular, this guidance should encourage the use of diligence terms in licensing agreements, particularly those with exclusivity. Increasing the number of insurers that reimburse for the test or improving the specificity and sensitivity of the test are examples of milestones that a licensee could be required to meet to earn or maintain license rights.

When Dr. Evans reviewed the first part of the sixth proposed recommendation, calling for the Secretary to encourage federal efforts to promote broad licensing and patient access, Committee members reached a consensus that this part of this recommendation should be made part of the discussion of the third recommendation. Dr. Evans then reviewed the next part of proposed recommendation 6, which called for federal agencies to explore patent pools, clearinghouses, and cross-licensing agreements as potential approaches to facilitate the development of multiplex tests and whole-genome sequencing. Dr. Williams

suggested that the advisory board called for in recommendation 5 could undertake this work and other Committee members agreed.

Recommendation 7.

> Because it is unclear whether the Bayh-Dole Act gives agencies authority to influence how grantees license patented inventions, the Secretary should seek clarification about this legal question. If it is determined that such authority exists, the Secretary should promulgate regulations that enable the Department's agencies to limit the ability of grantees to exclusively license inventions resulting from government funding when they are licensed for the genetic diagnostic field of use. Exceptions should also be allowed if a grantee can show that an exclusive license is more appropriate in a particular case, e.g., because of the high costs of developing the test. The Secretary should also direct NIH to make compliance with NIH's Best Practices for the Licensing of Genomic Inventions an important consideration in future grants awards.

Committee members agreed to rearrangements involving part of this recommendation and recommendation 3.

Recommendation 8.

> The Secretary should recommend that the Secretary of Commerce advise USPTO to establish an advisory committee to provide advice about scientific and technological developments related to genetic tests and technologies that may inform its examination of patent applications in the realm of human genes. The Committee believes experts in the field could help USPTO in its development of guidelines on determinations of nonobviousness and subject matter eligibility in this field once pending court decisions such as Bilski v. Kappos are decided.

Committee members discussed changes to this recommendation and agreed to various modifications to its wording.

Dr. Evans then returned to the final part of proposed recommendation 6, which stated, in part, that federal agencies should provide more detailed guidance regarding the licensing of patents associated with genetic tests. Dr. Evans suggested that this text be moved to the discussion of proposed recommendation 7. In the subsequent discussion, Committee considered a recommendation pertaining to coverage and reimbursement of genetic tests. Ms. Aspinall wanted any such recommendation to call for a solution to problems she asserted exist with regard to laboratories offering discounts on genetic tests. It was decided that Ms. Aspinall would draft text regarding this issue that could be used in the report, rather than in a recommendation. Dr. Wise then stated that issues of access to genetic testing should not be made part of the proposed recommendation under discussion and proposed a separate set of recommendations relating to equity in access to genetic testing as well as an appendix listing past SACGHS recommendations that address the inequitable provision of genetic tests and services.

Closing Remarks

Dr. Teutsch asked Dr. Evans to prepare revised recommendations for Committee discussion on October 9.

October 9, 2009

Opening Remarks—Dr. Teutsch

After welcoming remarks, Dr. Teutsch noted that the first topic of the day would be the ethical implications of data sharing.

Genomic Data Sharing

Discussion of Proposed Steps for the SACGHS Priority Topic on Ethical Implications of Genomic Data Sharing—Dr. Charmaine Royal

Goal. Dr. Royal explained that the goal of the discussion was to come to consensus on steps that SACGHS could take concerning issues that have emerged from large-scale sharing of genomic data—including the NIH requirement that research involving genome-wide associations funded by or conducted at NIH be submitted to the database of Genotypes and Phenotypes (dbGaP). In addition to individual, de-identified genomic data, SACGHS may want to consider genomic data related to groups. Because recent research papers have reported that it is possible to identify individuals from aggregate data, new concerns arise about data being broadly available to researchers. Issues to consider include (1) new ways of thinking about consent; (2) consent for future, unspecified research; (3) the interaction between genomic research (outcomes data) and its clinical relevance (i.e., the blurring line between research and clinical care); (4) concerns about whole-genome sequence data as a unique identifier that can be linked with data that might be obtained or stored in other contexts, which raises issues of privacy protection.

Prior SACGHS activities on this topic. In December 2008, SACGHS identified the ethical implications of genomic data sharing as a priority issue. At the March 2009 SACGHS meeting, the Committee was briefed on the Institute of Medicine report *Beyond the HIPAA Privacy Rule* and heard from the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) about informed consent in the context of newborn screening and potential re-use of residual dried blood spots derived from such screenings. In September 2009, the HHS Office of the Assistant Secretary for Planning and Evaluation (ASPE) awarded a one-year contract to The Lewin Group to develop a report on genomic data sharing based on a literature review and interviews with experts. The contract is designed to provide analytical support for the Committee's efforts.

Proposed action steps. Dr. Royal proposed the formation of a steering group to explore models of genomic data sharing and usage through a session at the February 4-5, 2010, SACGHS meeting. Questions to consider for this action step included whether the February 2010 session should focus on particular elements of data sharing or usage agreements, specific population, or certain types of data (e.g., data from clinical practice, secondary use of research data) as well as expected outcomes.

Invited comments. Dr. Gregory Downing, Program Director, Personalized Health Care Initiative, Office of the Secretary, HHS, agreed with the proposed plan for a SACGHS session genomic data sharing. He remarked that new partnerships are emerging across organizations, and HHS is interested in examining models that address issues related to consent, data sharing, publication. In addition, there is a great deal of interest in how technology supports the movement of data and the applicability of data to solve problems. Dr. Downing noted that it would be advantageous to have public policy perspectives prepared as the mobilization of data continues apace.

Dr. Kevin Fitzgerald, former SACGHS member, noted that Dr. Downing had helped set up a meeting at Georgetown University that examined the consequences for genomic research involving sharing data from vulnerable populations, particularly indigenous communities, which are of interest to genomic researchers due to their somewhat isolated genomic characteristics. Dr. Fitzgerald suggested that the topic fits with the Committee's mandate to explore genetics, health, and society and that it could serve as a microcosm for much broader issues.

Committee Discussion

Ms. Au observed that genomic data sharing is an important topic for states because of their newborn screening programs. Dr. Williams suggested including the DTC aspects of data collection within the scope of the project. Dr. Downing cited an example that provoked a lot attention last summer when a publication reported genetic findings about certain patterns of human behavior associated with substance abuse. The underlying concern was that the findings from population-based studies were not placed in a context that broader communities could understand. Dr. Downing noted that communication issues could be informed by the scientific aspects and take advantage of technologies such as podcasts and videos that provide a social construct for the implications of research findings. Dr. Royal remarked that studies have not been conducted in many vulnerable populations to understand how members of these populations feel about their data being shared, and as a result, researchers are reluctant to participate in data sharing.

Dr. Amos asked if practical aspects, such as information technology tools, would be included in SACGHS' work. If so, the National Institute of Standards and Technology would identify appropriate people to get involved.. Ms. Darien suggested bringing in more health advocates to give voice to the patient communities as the Committee explores this issue. Dr. McGrath recalled that the SACGHS report on large populations studies touched on many of the same issues. Such studies may be the best use of limited resources for addressing population health issues and communicating results from large studies remains a public priority.

Dr. Olsen remarked that the Department of Veterans Affairs (VA) has plans for studies that involve large data collection and improving capabilities for data sharing, and the VA has a strong interest in these issues (e.g., consent and privacy). Dr. Downing mentioned that an important contribution of the Georgetown conference was the different models of community consultation. He noted that different communities have different needs, especially internationally, and the importance of discerning community need and taking into consideration the various cultural perspectives on ownership. Dr. FitzGerald also acknowledged that other countries such as Canada and Mexico are ahead of the United States in development of models and methodologies.

Dr. Michele Lloyd-Puryear, SACHDNC Executive Secretary, remarked that SACHDNC would be interested in collaborating with SACGHS. SACHDNC has examined communication issues in the context of long-term follow-up when a genetic disorder is diagnosed through newborn screening. She noted the importance of engaging communities and developing standards that enable information exchange. Dr. Dale mentioned an international registry of a relatively rare set of conditions that he has overseen for 20 years and a key feature is linking clinical data to genetic data. He noted that one challenge has been dealing with the continued evolution of requirements for informed consent.. The registry offers a framework or foundation for building understanding in a community of interested people about the value of genetic studies as they relate to long-term health and could serve as a model for considering common conditions in larger populations.

Dr. Carome, HHS Office for Human Research Protections (OHRP), mentioned that the blurring between clinical practice and research dates back three decades when the Belmont Report was issued by the National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research, and

it is an ongoing concern of OHRP. He wondered if genetics further blurs the line in a way that is different from other data and, if so, SACGHS' work in this area could help inform OHRP. Dr. Carome also asked if OHRP needs to revisit its longstanding policy positions regarding de-identified tissue or coded tissue samples in light of new technologies. He reported that the Secretary's Advisory Committee on Human Research Protections is focused on research uses of biospecimens that have been banked for clinical or research reasons and related consent issues.

Drs. Teutsch and Royal concluded that there is enough interest to plan for a February 2010 sessionon genomic data sharing, and the following individuals agreed to be on a steering committee: Drs. Royal, Amos, Carome, Dale, Fitzgerald, Olsen, and Lloyd-Puryear and Ms. Au and Ms. Walcoff, as well as Ms. Dreyfuss, if needed. Dr. Royal said that she would ask the steering committee to address the rest of the questions raised today on genomic data sharing.

Genetics Education and Training

Dr. Teutsch noted that the Genetics Education and Training Task Force has made considerable progress under the leadership of Barbara Burns McGrath. He stated that the purpose of this session is to review the findings of the Task Force and get initial input from the Committee on the draft recommendations, which will be presented in a more final form at the February 2010 meeting..

Literature and Survey Findings

Dr. McGrath, Chair of the Genetics Education and Training Task Force, explained that the Task Force was composed of three workgroups: (1) health care professionals, (2) public health providers, and (3) consumers and patients. The workgroups collected data about genetics education and training needs through literature reviews, surveys, and interviews with experts. Although survey data analysis was ongoing, Task Force members had begun to consolidate findings and develop recommendations, which they recognized must be actionable and forward-looking.

Health care professionals. A major finding from the literature review is that integration of genetics into health care is limited by a lack of or inappropriate genetics education. Licensure, certification, and accreditation requirements for health care professionals have not kept up-to-date with advances in genetics and genomics, and based on data of the American College of Medical Genetics, the existing number of medical geneticists in the U.S. workforce is only 41 percent of what is needed.

The workgroup surveyed Federal agencies and health professional organizations. The latter survey indicated that 70 percent of respondents viewed genetics education as part of the role of their organization, but more funding, program evaluation, and greater interest in genetics education within their own organizations' leadership, is needed. Competing priorities are a major barrier to providing genetics and genomics education.

Public health providers. From a literature review and survey data, the public health provider workgroup found that the current public health workforce is not well prepared to receive and assimilate genetic and genomic information into public health. Among the identified barriers were: (1) diverse roles of the public health workforce, (2) varied education and training paths represented by that diverse group, (3) out-of-date formal training, and (4) a general sense within the workforce that the utility of genetics is not clear. When asked to rate 12 genetics-related competencies, respondents from the public health workforce chose "Demonstration of basic knowledge of the role of genetics in the development of disease, and in screening and interventions for programs of disease prevention and health promotion" as the most important. The competency for conducting outcome evaluations was ranked least important.

Two-thirds of survey respondents felt that their genomic resources were inadequate for implementing the competencies within their area of public health.

Consumers and patients. The workgroup acknowledged that sources of genetics information are not lacking. Consumers generally recognize that genes and behavior are related to health outcomes but know less about complex traits and multifactorial conditions. Consumers express continued concern about confidentiality and disclosure of genetic information.

An online survey was developed based on interviews with subject matter experts and sent to consumer organizations. More than 300 responses were received and analyzed. Because the survey participants were not representative of the general U.S. population, the workgroup reviewed data from a consumer survey conducted by Cogent Research. From these data, three clear findings emerged: (1) consumers want to get information about genetic testing from primary care providers but are not confident that providers have adequate knowledge, (2) the government is seen as a trusted source for genetic and genomic information and should act as a clearinghouse for this information, and (3) family history is seen as an important tool to understand health and disease.

Draft Recommendations

Dr. McGrath invited the Committee to help shape 13 draft recommendations and ensure that nothing important from prior discussions had been left out. She explained that some recommendations are workgroup-specific and some are more general.

Health care professionals. The draft recommendations proposed actions that HHS could take to (1) encourage the integration of genetics and genomic content into health professional education; (2) stimulates creative, innovative, collaborative care delivery; and (3) facilitate interdisciplinary collaboration.

Public health providers. The draft recommendations proposed that HHS: (1) assess the public health workforce regarding genetic and genomic responsibilities, (2) facilitate the development of core competencies, and (3) promote collaborative education and training.

Consumers and patients. The draft recommendations proposed that HHS should (1) coordinate with other Federal agencies and community organizations to improve literacy in genetics and genomics, (2) support expanding educational resources for the public, (3) support continued efforts related to family health history tools;,and (4) increase public understanding of the risks and benefits of participating in genetic research.

Draft recommendations applicable all three workgroups proposed that HHS should (1) consult with other agencies and ensure funding of national strategic planning for genetics and genomics education and training, (2) facilitate increased health professional faculty training, and (3) support research to develop effective methods of translating genetic and genomic science to information that can be incorporated into health care practice. Dr. McGrath also presented four relevant recommendations from prior SACGHS reports regarding health provider education and training, coverage and reimbursement of genetic services, regulation of genetic testing, and the need for greater public awareness.

Committee Discussion

Dr. Evans suggested that the second health care professional recommendation recognize that formal didactic mechanisms for teaching are inadequate and that genomic education must be directly integrated into patient care through just-in-time tools and resources. Dr. Nussbaum added that a logical step would

be inclusion of genetics and genomics in the practice improvement modules (PIMs) that have become part of the process of recertification in health professional organizations. Reimbursements could be an incentive to use these PIMs.

Dr. Lloyd-Puryear reported that ACHDNC had developed a recommendation on education of primary care providers that she would share with SACGHS. Dr. Khoury agreed with the recommendation to assess the public health workforce because of its heterogeneity and varied education needs. He also noted that passive learning is not effective and advocated for actively involving the public to help them understand their own genome. Family history is one way to engage the public.

Regarding health provider education, Dr. Williams noted the importance of moving beyond traditional educational methods and embracing innovative approaches such as using electronic health records for just-in-time learning. He also remarked that it is critical to incorporate genetics and genomics education in clinical training to build life-long practice patterns. Dr. Williams suggested that HHS could fund an evaluation of health professional education and support unconventional methods for education and training.

Dr. Dale proposed engaging with the Institute of Medicine and others interested in health literacy to gain a better understanding of the public's knowledge of genetics. Drawing on his experience with Mexican Americans in Los Angeles, Dr. Licino observed that it is hard to engage a community in something that its members know nothing about; therefore, it is necessary to do education and training in parallel with engagement.

Ms. Dreyfuss wondered if the education process could take advantage of people's interest in DNA analysis for forensic purposes, as often seen in television programs. Using HIV/AIDS education as an example, Dr. Lloyd-Puryear agreed that for genetics education to succeed it must be tied to something tangible. She also noted that enthusiasm for the 1999 Genetics in Primary Care Project—sponsored by SACHDNC, NIH, and the Agency for Healthcare Research and Quality—waned because health care providers struggled to tie genetics knowledge to everyday practice. Mr. Bonham commented that members of the public do not need to know everything about genetics, only the information necessary to help them make good decisions.

Dr. Amos remarked that an important role for HHS is to serve as a clearinghouse for genetic and genomic information to help consumers identify credible information. Dr. Guttmacher said that public education is critical and that the Surgeon General's family health history initiative and web-based tool facilitate consumer education. He also suggested that the Secretary of HHS talk with the Secretary of the Department of Education.

Dr. Teutsch thanked everyone for contributing to a valuable discussion.

Public Comments

Personalized Medicine Coalition (PMC). Amy Miller, Ph.D., PMC Public Policy Director, reported on an educational effort in which PMC has worked with consumer genomic companies over the past 18 months to address some concerns raised by SACGHS and others regarding their products. She explained that PMC partnered with Medco a pharmacy benefit manager, to develop a consumer guide to DTC genetic tests. Ms. Miller added that PMC believes that a government-sponsored guide is needed as well.

National Human Genome Center at Howard University. Luisel Ricks, Ph.D., a research associate at the National Human Genome Center at Howard University, commented that some companies offering DTC personal genomic testing claim the ability to predict risk of diseases such as cancer, cardiovascular

disease, and diabetes. She cautioned if these tests are not used equitably by all groups, they may actually widen the health disparities gap because they concern diseases that affect minorities at alarmingly higher rates than other members of the U.S. population. Dr. Ricks noted that national health surveys, such as HealthStyles, the National Health Interview Survey, and the Behavioral Risk Factor Surveillance System, show that ethnicity, education, and socioeconomic status significantly predicted awareness and use of personal genome tests. Specifically, these surveys revealed that African Americans and Hispanic Americans are less aware of DTC personal genome tests than Caucasians. She added that another barrier to the use of DTC tests is affordability. In 2006, the U.S. Census Bureau reported that African American, American Indian, Alaska Native, and Hispanic households earn less than 75 percent of Caucasian households. Cost has the potential to exclude minority populations from using personal genome tests and exacerbate inequalities in health and health care. Dr. Ricks concluded by stating that is it of paramount importance to educate consumers to ensure awareness of genetics and personalized medicine and adopt policies that ensure affordability and equal access by all groups to genomic applications.

Association of University Technology Managers (AUTM). Speaking on behalf of AUTM, Ted Rumel, Vice President for Research Innovation and Commercialization at the University of Maryland Biotechnology Institute, made remarks regarding the Committee's report on gene patents. Mr. Rumel indicated that AUTM does not support additional regulations, clarification of the USPTO role, or any statutory changes to the Bayh-Dole Act but does recommend further data analysis and expert testimony, on which appropriate recommendations could be based. He also said that AUTM recognizes the concerns related to gene patents and supports additional guidelines to augment those developed by NIH and AUTM.

Susan Poland, Susan Poland, J.D., Legal Research Associate at the Kennedy Institute of Ethics at Georgetown University, spoke about family history and public education. She proposed that the meaning of family history needs to be defined more clearly and environmental factors also need to be considered.

Facing Our Risk of Cancer Empowered (FORCE). Speaking on behalf of FORCE, Lisa Schlager stated that FORCE has mounting concerns about the marketing used by genetic laboratories, particularly by Myriad Genetics. It is FORCE's belief that Myriad's sales representatives have discouraged doctors and other health care providers from referring patients to genetics experts. She also stated that in a recent publication Myriad's CEO was quoted as saying that "the sales force at Myriad provides doctors with the tools to do counseling in-house, and as a result, physicians can bill the insurers directly." Ms. Schlager asserted that these practices have had negative consequences. She stated that FORCE has learned of patients who have had the wrong tests ordered for them and of one patient who had an unnecessary surgery to remove her ovaries based on Myriad test results that were misinterpreted. In light of problems such as these, Ms. Schlager urged SACGHS to recommend federal action to track adverse events resulting from marketing of genetic testing by laboratories and to require doctors, prior to ordering a genetic test, to inform their patients that they should receive genetic counseling.

During the discussion, Dr. Kanis, an *ex officio* member of SACGHS, spoke of seeing similar problems in his practice. Dr. Mansfield, *ex officio* representing FDA, observed that anyone experiencing adverse events can report them to the FDA. Dr. Billings suggested inviting Myriad to respond to the statements made by Ms. Schlager.

DTC Genetic Testing

Presentation of Revised Draft Paper on DTC Genetic Testing

Ms. Au, Chair of the DTC Genetic Testing Task Force, explained that the goals of the session were to (1) reach consensus on key areas for the Secretary's attention, prior SACGHS recommendations and action steps that address these areas, and remaining concerns that may require additional action and (2) approve the paper for transmission to the HHS Secretary. The Task Force started its work in March 2009, presented an initial draft paper at the June 2009 meeting, and revised the paper in response to comments from SACGHS members.

Recent Federal Trade Commission (FTC) actions. Noting relevance to the report, Ms. Au asked Sarah Botha, FTC staff attorney in the Division of Advertising Practices, to brief the Committee on recent FTC actions against two companies offering DTC genetic testing that culminated in closing letters on August 14, 2009. Ms. Botha explained that one company—Sciona, a manufacturer, processor, and marketer of neurogenetic testing—offered a test kit and consultation service (the MyCellf Program or the Cellf Test), and Genelex Corporation marketed and distributed the test and forwarded test samples to Sciona for processing while also marketing its own ancestry and paternity tests. FTC's concerns were that both companies made marketing claims (expressed and implied) that the diet and lifestyle recommendations that were given as part of the program could significantly impact consumers' health outcomes, including their risk of developing serious diseases. Another unsubstantiated claim was that having a neutrogenetic test could help you lose weight and keep off the weight. This information went directly to consumers with no physician involvement. FTC viewed all these claims as unsubstantiated scientifically and consulted FDA and others to verify its view. Eventually Genelex agreed to stop all neurogenetic testing, and Sciona, having gone into bankruptcy, stopped as well and destroyed consumers' DNA samples and lifestyle questionnaires. Ms. Botha said that she could not share the number of consumers involved and noted that the companies' strong confidentiality agreements made it easier for FTC to apply pressure.

Review of the revised paper—key areas for the Secretary's attention. Ms. Au stated that the objectives of the paper were to (1) outline the benefits and concerns related to DTC genetic testing, (2) highlight prior SACGHS recommendations that address these concerns, and (3) identify issues that are not adequately addressed by prior recommendations and may require further action. She noted that some concerns identified in the paper are not unique to DTC genetic testing but may apply more broadly to provider-based laboratory tests. Other issues are unique to DTC genetic testing if a consumer's personal health provider is not involved in health decisions or government regulations do not apply to entities providing DTC services.

Ms. Au explained that the revised draft paper includes three key areas for the attention of the Secretary and five specific action steps. The first key area concerned (1) gaps in the federal oversight of DTC genetic testing, particularly the absence of FDA review of DTC genetic testing promotional materials and claims due to limitations under current regulatory practices and (2) lack of evidence of clinical validity and utility for most health-related DTC genetic tests. Ms. Au noted that DTC companies may claim that their tests are not health related.

The second key area involves gaps in privacy and research protections for consumers using DTC testing. As most companies do not receive federal funding, they are not subject to federal regulation. State-level protections may be inadequate. The third key area concerned the limited genetic knowledge among consumers and health care providers as well as limited involvement of the consumer's personal health care provider in assisting consumers to select genetic tests and make health care decisions based on DTC genetic test results.

Recommendations and action steps. Ms. Au noted that nine prior SACGHS recommendations form the basis for the five action steps recommended to the HHS Secretary in this paper. These recommendations from SACGHS reports on the oversight of genetic testing and the coverage and reimbursement of genetic tests and service, address concerns related to oversight gaps, marketing claims, promotional materials, analytical validity, clinical validity, clinical utility, standardization, privacy, and consumer and provider education. Based on these prior recommendations, the following actions that the Secretary can take were proposed:

Action Step 1: Direct the FDA Commissioner and CMS Administrator to solicit broad stakeholder input through a series of public hearings, then convene jointly to draft and publish an advanced notice of proposed rulemaking that (1) analyzes gaps, inconsistencies, and duplications in regulations related to DTC genetic testing and (2) identifies specific proposals to address them within relevant statutory authority.

Action Step 2: Include laboratories that provide DTC genetic testing and services, if HHS establishes a laboratory registry.

Action Step 3: Convene a joint HHS-FTC task force—with industry, consumer, academic, and government stakeholders—to propose specific guidelines for DTC genetic testing, advertising, promotion, and claims consistent with existing statutory authority. The task force would also identify gaps in the authority relevant to the emergent industry. These guidelines, which will form the basis of more targeted federal enforcement of claims that are misleading and/or not truthful, should be grounded in evolving evidence standards—which are accepted by experts in relevant fields—for identifying and evaluating competent and reliable scientific evidence of DTC genetic test performance consistent with the claims made by DTC companies related to these tests.

Action Step 4: Direct the HHS OCR, with support from OHRP and other relevant HHS agencies, to identify specific gaps in state and federal privacy protections for personal health information that may be generated through DTC genetic testing and propose to the HHS secretary specific strategies the Federal Government can undertake consistent with its existing authority to address these gaps and inform consumers of potential risks to privacy.

Action Step 5: Develop an initiative within ASPE focused on genetics education, including information specific to DTC genetic testing and links to HHS educational resources for consumers and health practitioners. ASPE should also follow up its March 2009 report, *Consumer Use of Computerized Applications to Address Health and Health Care Needs* by conducting research and evaluating studies specific to DTC genetic testing, developing policy analyses, and estimating the costs and benefits of policy alternatives and potential regulations under consideration by HHS.

Ms. Au cited the following additional areas of concern that are not adequately addressed by prior SACGHS recommendations and may benefit from further evaluation by SACGHS and/or appropriate federal agencies: nonconsensual testing, limited data on the psychosocial impact of DTC genetic testing, the impact of DTC genetic testing in children, potential exacerbation of health disparities, inadequate protection for the research use of specimens and data derived from specimens, and the impact of DTC testing on the health care system.

Committee Discussion

Referring to the third action step, Dr. Billings asked to what extent the recommendation for a new task force furthered the previous SACGHS oversight recommendations. He also wondered if FDA decided to regulate these tests, would the task force be needed. Dr. Mansfield said that most DTC genetic tests

appear to fall under the rubric of medical device, and FDA would have authority to regulate them. Currently, however, FDA does not conduct premarket review or postmarket control of these tests. Dr. Williams asked about FTC's role in the oversight of claims. Is oversight under the sole purview of FTC or FDA, or does each agency play a role? If both agencies are involved, then a joint task force would be valuable. Ms. Botha explained that FTC has a broad statutory authority to address unfair and deceptive acts and practices affecting commerce and more specific authority to go after false advertising for health care products, including devices. FTC also has a longstanding memorandum of understanding with FDA regarding overlapping authority. FDA takes primary jurisdiction for product labeling, and FTC takes primary jurisdiction for product advertising, with exceptions for prescription drug advertising and restricted medical devices. She said that DTC genetic testing clearly falls under the Federal Trade Commission Act, but one problem is the lack of agreed-upon evidentiary standards for clinical validity and clinical utility.

Dr. Evans suggested adding a short preamble to the third action step about reconciling claims with the reality of the evidence, which would help explain the rationale for the FDA-FTC task force. Ms. Botha clarified that FTC is primarily an enforcement agency, not a regulatory agency, and it is unlikely to issue regulations in an area where the science is emerging and evolving. She also wondered how one could write guidelines in a field that is continuing to change, but Dr. Evans remarked, and Dr. Amos agreed, that the types of issues that guidelines would address are not contingent on the technology. Ms. Au added that stakeholders with the necessary expertise would be members of the task force and could provide advice for the development of guidelines for DTC genetic test advertising, promotion, and claims.

Dr. Billings noted that it sounds as if, rather than a task force, what the agencies need is access to appropriate expertise. Dr. Williams suggested that the task force's main priority should be to deal with the evidentiary standards issue, and the action item needs to reflect this focus. Dr. Khoury mentioned that the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) working group, will likely looking at diabetes and cardiogenomic profiles. He also mentioned that the topic of scientific standards for personal genomics was discussed at a December 2008 NIH-CDC workshop, and the recommendations from the workshop were published in the August 2009 issue of *Genetics and Medicine* (see http://journals.lww.com/geneticsinmedicine/toc/2009/08000). Dr. Khoury noted that there is limited evidence for clinical validity and clinical utility for gene variants based on genome-wide association studies. He stated that the time for action now is now because so many people are already availing themselves of the DTC services.

Regarding the prior SACGHS oversight recommendation that FDA take a risk-based approach in evaluating laboratory tests, Dr. Mansfield said it would be of interest to know where the DTC genetic tests fall on a continuum of risk—since any FDA regulation of laboratory-developed tests would likely be on a risk basis. Dr. Teutsch remarked that DTC tests have an intrinsically higher risk than tests ordered through a knowledgeable provider. That is, if the same test is offered DTC or through a knowledgeable health care provider, the DTC test is likely to have higher risk. Dr. Mansfield replied that she did not think FDA would assign risk based on who orders the test.

Dr. Randhawa stated that the focus of the evidentiary standards needs to be narrowly defined since clinical guideline developers have a different perspective from those who make reimbursement coverage or regulatory decisions. Dr. Amos remarked that he would like to see the Committee use stronger wording about whether the tests are bad or good and perhaps ask the Secretary to make a statement about these tests to help educate the public.

Dr. McGrath applauded the even tone of the report and the way that it recognizes that DTC genetic testing will continue to exist. The Committee's focus on evidence is appropriate. Ms. Au returned to one of the

goals of the paper—to use DTC testing as a means for the Secretary to consider prior SACGHS recommendations that apply to a broad spectrum of genetic tests, not just DTC tests.

Dr. Williams proposed adding a definitive statement that companies cannot escape scrutiny because they claim their tests do not provide health information. Dr. Ferreira-Gonzalez added that some companies contract with CLIA-certified laboratories to provide data, which is transferred back to the company. The companies are not subject to CLIA regulation because they do not produce the data. Ms. Au explained that these issues fall under the first action step. Dr. Ferreira-Gonzalez suggested adding the prior SACGHS oversight recommendation that addressed certain health-related tests that fall outside the scope of CLIA.

Dr. Penny Keller of CMS described how the agency monitors genetics/genomic companies. CMS attempts to contact DTC companies and whenever it a response, the company is asked for information about their tests—including their requisition forms, testing descriptions, and the test reports that they generate and send to consumers or providers. If the information can be used for health assessment by the provider, then CMS indicates whether it falls under CLIA. If it does, the company would need to meet CLIA requirements or requirements of one of the accrediting agencies. One example of a genetic test that does not fall under CLIA is testing for bitter tasting, which does not lead to treatment or health assessment. Ms. Keller added that even if a company claims that a test is not a diagnostic, CMS still asks for information and tries to educate. She cautioned, however, that unless a company applies for CLIA, CMA does not have the force in the statute to go after the company.

Dr. Ferreira-Gonzalez asked Dr. Keller to clarify whether companies that contract with a CLIA-certified laboratory for testing are subject to CLIA regulations. Using 23andMe as an example, Dr. Keller explained that 23andMe does not perform laboratory testing but provides interpretation of data generated in a CLIA-certified laboratory. Current CLIA regulations do not extend to interpretation services, so 23andMe does not need to meet CLIA requirements. She added that some state laws cover this regulatory gap. Several SACGHS members agreed that this gap in oversight needs to be addressed, which could be done through the prior oversight recommendation about health-related tests that fall outside the scope of CLIA.

When Dr. Williams asked whether CMS provides any information to FTC about companies that state that they are not providing health information, Dr.. Keller replied that currently CMS is concentrating on interacting with FDA. She said that CMS has not been gathering information long enough to notify FTC about companies that have not responded. It sometimes takes 3 months for a company to respond, so CMS is not yet at a point to involve FTC.

Dr. Khoury proposed that Appendix B be brought back to a more prominent position in the report because it lists prior recommendations that apply to DTC and indicates that more specific steps need to be taken. When others explained that Appendix B was created so as not to dilute key recommendations, Dr. Khoury commented that no one will read it and added that the information needs to be in the executive summary. Ms. Au said that she and SACGHS staff will make some revisions to add discussion of prior recommendations in the body of the paper.

SACGHS members approved the report (14 to 0). Revisions based on the Committee's comments will be made by Thanksgiving, and the Committee will be asked to review the report by Christmas. The final paper can then be ready to forward to the Secretary in early 2010.

Gene Patents and Licensing Practices

The Committee reviewed the revised proposed recommendations for the gene patents report. Before turning to that task, Dr. Teutsch requested that Committee members with perspectives different from those in the report prepare written statements that could be included in the report. He also emphasized that the rationale for the recommendations must be clearly articulated in the report.

Dr. Evans reviewed the revised recommendations and asked for comments.

Recommendation 1: Supporting the Creation of Exemptions from Infringement Liability (13 members supported, 2 opposed, 1 abstained)

> The Secretary of Health and Human Services should support and work with the Secretary of Commerce to promote the following statutory changes:
>
> A. The creation of an exemption from liability for infringement of patent claims on genes for anyone making, using, ordering, offering for sale, or selling a test developed under the patent for patient care purposes.
> B. The creation of an exemption from patent infringement liability for those who use patent-protected genes in the pursuit of research. Related health care and research entities also should be covered by this exemption.

No changes were made to part A. For part B, there was a proposal to drop the last sentence—"Related health care and research entities also should be covered by this exemption." Commenting on this proposal, Ms. Dreyfuss indicated that there was a possibility that hospitals could be sued for contributory infringement if the exemption did not extend to them. After further discussion, the Committee agreed to drop the sentence from the recommendation and add the text to the report that discusses this issue.

Recommendation 2: Promoting Adherence to Norms Designed to Ensure Access (14 members supported, 1 opposed, 1 abstained)

> Using relevant authorities and necessary resources, the Secretary should explore, identify, and implement mechanisms that will promote more than mere voluntary adherence to current guidelines that promote nonexclusivity in licensing of diagnostic genetic/genomic technologies.
>
> The Secretary shall convene stakeholders—for example, representatives from industry and academic institutions, researchers, and patients—to develop a code of conduct that will further broad access to such technologies.

Dr. Evans explained that the recommendation was shortened, and the text that was removed will be folded into the body of the report. Ms. Dreyfuss remarked that representatives from "academic institutions" will likely be technology transfer professionals and academic researchers should explicitly be included. Although the recommendation mentions "researchers," they could be from industry or academia. Dr. Evans replied that an explanation can be added to the report that "academic institutions" includes researchers.

Recommendation 3: Enhancing Transparency in Licensing (13 members supported, 2 abstanined)

> Using relevant authorities and resources as necessary, the Secretary should explore, identify, and implement mechanisms that will make particular information about patent licenses readily

available to the public. The specific licensing terms that should be available are those that pertain to the type of license, the field of use, and the scope of technologies.

Committee members agreed that the text discussing the recommendation would state that the HHS Secretary should (rather than "could") direct NIH to amend guidelines to promote disclosure of information about licenses.

Revised Recommendation 4: Establish an Advisory Body on the Health Impact of Gene Patenting and Licensing Practices (13 members supported, 1 abstained)

> The Secretary should establish an advisory body to provide ongoing advice about the public health impact of gene patenting and licensing practices. The advisory body could provide input on the implementation of any future policy changes, including the other proposed recommendations in this report.

The Committee agreed with Dr. Evans' suggestion that "public health impact" should be changed to "health impact." In the report's discussion of this recommendation, the Committee agreed that the advisory body could be established within an existing committee.

Recommendation 5: Providing Needed Expertise to USPTO

> The Secretary, working with the Secretary of Commerce, should designate a liaison between this Committee and the USPTO. This liaison, along with technical advisors the SACGHS could recommend, would provide input to the USPTO about scientific and technological developments related to genetic testing and technologies. This input would help inform the USPTO's examination of patent applications in the realm of human genes.

Committee members decided that it was unnecessary to spell out the particular mechanism (i.e., a liaison between SACGHS and USPTO, along with other technical advisors) through which the PTO would be provided with scientific experts. The simplified recommendation stated:

> The Secretary should work with the Secretary of Commerce to ensure that the USPTO is kept apprised of scientific and technological developments related to genetic testing and technology.

Recommendation 6: Ensuring Equal Access to Clinically Useful Genetic Tests

> Given that genetic tests will be increasingly incorporated into medical care, the Secretary should ensure that those tests shown to have clinical utility are uniformly covered by governmental and nongovernmental payers.

Dr. Wise suggested adding a paragraph at the beginning of the report to provide a foundation for this recommendation. He proposed stating that SACGHS has long been concerned about the rapid evidence-based implementation of the equitable provision of genetic and genomic capabilities and then reference prior Committee reports. Ms. Dreyfuss agreed and suggested including the reduction of health care cost in addition to the equitable provision of genetic tests. Dr. Teutsch remarked that a process is needed for individuals who do not have insurance coverage to be assured access to genetic services and proposed adding that the Secretary should take step to identify and remove barriers to access. He suggested reframing the recommendation so that it addresses access to tests, and other issues such as uniform insurance policies and removing barriers to access could be discussed in the body of the report. The recommendation was revised as follows:

Given that genetic tests will be increasingly incorporated into medical care, the Secretary should ensure that those tests shown to have clinical utility are equitably available and accessible to patients.

The Committee approved the recommendations (12 to 1, with 1 abstention), but the members stopped short of approving the entire draft report. They called for particular revisions to the report's background sections, including a more extensive incorporation of public comments received at the meeting and during the prior public consultation process. A subgroup of the Committee will guide the revision process, and the revised report will be reviewed again at the next Committee meeting.

Concluding Discussion and Adjournment—Dr. Teutsch

Before adjourning the meeting, Dr. Teutsch asked for suggestions on addressing three issues that had been raised by Dr. Collins, the recently appointed director of NIH. The first is incorporating the economic value of technological innovations into the activities of the Clinical Utility and Comparative Effectiveness Task Force. The second is considering whether the Committee should address the implications of an affordable genome as a discrete topic. The third is to write a commentary for publication highlighting prior SACGHS recommendations. .

Drs. Dale, Evans, Ferreira-Gonzalez, and Licinio volunteered to help Dr. Teutsch and staff prepare the commentary article. Dr. McGrath cautioned to think carefully about the intended audience to prevent a narrow focus. Dr. Teutsch suggested a broad audience could be achieved by aiming for publication in journals such as *Health Affairs*, *The New England Journal of Medicine,* or *The Journal of the American Medical Association.* Ms. Darien offered to help prepare a lay version after the journal article is ready.

Concerning incorporating the economic value of technological innovations, Dr. Teutsch then inquired whether this topic is already being addressed by the Clinical Utility and Comparative Effectiveness Task Force. Dr. Williams responded that any rational view of comparative effectiveness has to include issues around costs, including cost-effectiveness in the traditional sense, opportunity costs, comparative costs, and cost minimization. Task Force members are still reviewing documents from Federal agencies and other organizations, and we will be able to comment at the February 2010 SACGHS meeting. Dr. Williams added that the Task Force wants to be responsive to the Secretary's role in the realm of comparative effectiveness.

With regard to the idea of addressing the implications of an affordable genome, Dr. Ferreira –Gonzalez said that an in-depth look is needed, not only at analytical and clinical validity and clinical utility, but also the related ethical issues. Ms. Walcoff and Ms. Au agreed, noting the importance of family history to help understand the meaning of the presence of genetic variants. Dr. Williams felt that an educational session on the affordable genome at the June 2010 meeting would be highly useful. Dr. McGrath remarked that the Committee should be sure that it examines how genetics can help health and society generally, not just a privileged few. She also cautioned that an educational session on the affordable genome would be a great step, but the Committee must be careful not to push aside other important issues. Dr. Teutsch proposed scheduling some discussion at the February meeting to help prepare for a more extended session.

Dr. Teutsch briefly reviewed the topics and action steps that were covered during the meeting and thanked everyone for their valuable input.

Adjournment

The meeting was adjourned at 2:25 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