Discussion of Proposed Priority Issue Areas

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Priority-Setting Process— A Quick Review of Steps Thus Far

- 73 potential priority issues were generated through
  - a brainstorming session at the February 2008 SACGHS meeting
  - subsequent discussions with *Ex Officios*
  - solicitation of public comments
  - interviews with additional experts in the field
- Issues ranked by Committee using Likert scale; priority areas emerge from “heat map” analysis
- Committee discussion in July leads to the identification of eight priority areas (“clusters”)

Priority-Setting Process— A Quick Review of Steps Thus Far

- Task Force incorporates disparities priority area into other clusters
- Issue briefs developed

Seven Proposed Priority Areas (aka Clusters)

1. Coverage and Reimbursement for Genetic Services
2. Ensuring the Clinical Utility of Genetic Information
3. Genetics Education and Training with Attention to Workforce Diversity
4. Informed Consent, Privacy, and Discrimination Issues in Genomic Data Sharing
5. Implications of Consumer-Initiated Use of Genomic Services
6. Public Health Applications of Genomics Research with Attention to Health Disparities
7. Genetics and the Future of the Health Care System

Today's Session Goals

- Review cluster areas agreed to in July
- Discuss specific policy questions and proposed action steps in each cluster
- Develop an overarching and flexible action plan (recognizing the need for adjustments to reflect the incoming Administration's priorities)

Cluster 1
Coverage and Reimbursement for Genetic Services

Topic Statement

- Problems with coverage and reimbursement limit access to and integration of new genetic tests and services into the health care system.
- SACGHS should
  - Pursue unresolved issues from the February 2006 SACGHS report *Coverage and Reimbursement of Genetic Tests and Services*
  - Identify strategies to address obstacles to implementation
  - Identify any new issues
    - Address concerns of the public, including - -problems within Medicare coding, billing, and payment,
    - reimbursement policies that are barriers

Background

- In 2003, SACGHS identified coverage and reimbursement (C&R) as a high-priority issue; culminated in the 2006 report *Coverage and Reimbursement of Genetic Tests and Services* with 9 recommendations

- Actions following the 2006 report
  - June 2006: SACGHS Chair and C&R Task Force Chair meet with CMS Administrator and staff
  - July 2007: SACGHS sends response to CMS' comments to C&R report
  - December 2007: Conference call with CMS staff to discuss July comments
  - February and August 2008: SACGHS sends letters to HHS Secretary regarding family history and genetic counseling services

Policy Questions

1. What is the best approach to revise payment rates to reflect the true cost of a genetic test?

2. What billing options available to certified genetic counselors? What is the best approach to enable genetic counselors to use the full range of CPT E&M codes to enhance patient accessibility to genetic counseling services and informed decision-making before and after genetic testing?

3. What is the best approach to avoid inappropriate application of reimbursement audits (such as MUEs) to procedure-specific CPT codes that are appropriately used multiple times for molecular diagnostic testing?

Policy Questions (continued)

4. What is the best approach to include family history in “personal history” of a disease so that a beneficiary with a family history of a disease can meet the “reasonable and necessary” standard for Medicare coverage?

   Could EGAPP work with USPSTF to define those cases in which family history should be considered a personal history of disease?

   Could the findings from the August 2009 State-of-the-Science Conference on Family History also inform this process?

   In addition, how can clinicians be adequately reimbursed for the construction and analysis of a pedigree that would be used in medical decision making?

Policy Questions (continued)

5. Can we infer from the fact that CMS sought public comment on whether PGx testing should be a potential NCD topic that CMS considers PGx testing to be diagnostic testing and not screening? How might a PGx NCD be used to address issues raised in questions 1 and 3 above?

6. How can access to genetic services be improved in underserved populations?

Possible Action Steps

Monitor

- Continue monitoring recommendations from the 2006 coverage and reimbursement (C&R) report

Short Term Actions

- Continue discussions with CMS officials regarding
  - C&R of genetic counseling services
  - Revising payment rates for laboratory tests
  - Use of medically unlikely edits
  - How CMS will approach making a national coverage decision for PGx testing.

Possible Action Steps (continued)

Short Term Actions (continued)

- Confer with the laboratory community to gauge support for the application of CMS' inherent reasonableness authority to the entire Clinical Laboratory Fee Schedule
- Analyze the implementation of P.L. 110-275, the Medicare Improvements for Patients and Providers Act of 2008, which authorizes CMS to cover clinical preventive services if the U.S. Preventive Services Task Force recommends them and MedCAC concurs
- Encourage collection of demographic data for better assessment of access to and utilization of genetic services in underserved populations

Cluster 2
Ensuring the Clinical Utility of Genetic Information

Topic Statement

Multiple Challenges in Establishing the Clinical Utility of Genetic Tests

- Lack of underlying clinical studies
- Lack of defined/accepted standards against which to judge studies
- Lack of dedicated organization for performing utility assessments

An Important Issue on the Horizon

- Whole genome sequencing may soon be available in a clinical setting
- A group or organization will be needed to perform utility assessment for this information

Background

Other Groups Working on This Issue

- Evaluation of Genomic Applications in Practice and Prevention (EGAPP)
  - Working to define standards; performs assessments as well
- Hayes and Blue Cross Blue Shield's Technology Evaluation Center
  - These private organizations perform utility assessments
- Institute of Medicine Roundtable on Translating Genomic-based Research for Health

Background (continued)

SACGHS's Previous Work on This Issue

- Reports on Oversight, PGx, and Coverage and Reimbursement examined need for clinical utility information
- Recommended that Secretary create public/private group that would define the types of underlying studies needed for assessments and the standards by which to judge these studies; group would carry out a number of other functions, including performing assessments and disseminating clinical guidelines based on assessments

Policy Questions

1. What group(s) would be most effective in defining the evidentiary needs and standard of review for evaluating the clinical utility of an affordable genome sequence?

2. How might Government better inform those involved in research and development about the evidentiary needs for clinical utility determinations in general and in the case of specific technologies and specific conditions?

Policy Questions (continued)

3. Should the Government create a public/private organization responsible for assessing the clinical utility of genetic tests?

4. How can the Government facilitate the use of information to enhance the delivery of care [See Issue Brief 7]

Possible Action Steps

Short Term Actions

- Provide a forum for discussion to help define the evidentiary needs and standard of review for evaluating the clinical utility of an affordable genome sequence.
- Recommend that any governmental organization or group tasked with assessing clinical utility apply different clinical utility assessment methods for different clinical uses of genetic tests.

Possible Action Steps (continued)

Brief Reports on

- How clinical utility assessments can incorporate contextual issues (cost, cost effectiveness, ethics, legal issues, feasibility, acceptability, etc.) to better inform users (decision makers such as patients, regulators, public and private payers, health care providers, and performance measurement specialists).
- Particular procedure or system that should be used to inform those involved in research and development about the evidentiary needs for clinical utility determinations.

Possible Action Steps (continued)

Brief Reports on (continued)

- Government organizations or groups other than a public/private group of stakeholders that can establish evidentiary standards for clinical utility assessments of genetic tests, that can create methods for assessing the clinical utility of genetic tests, and that can perform utility assessments for genetic tests.

Cluster 3
Genetics Education and Training

Topic Statement

- Advances in genetics and genomics are leading to better insights into disease processes and improved applications of genetic testing to inform health decisions
- Health community and the general public challenged to keep pace with these advances.
- SACGHS Genetics Education and Training Task Force, established in November 2007, will examine the education and training needs of:
  - point-of-care health professionals with and without training or expertise in genetics
  - public health providers
  - patients and consumers

Background

- Many clinicians are unprepared to interpret genetic risk information and/or discuss details of the condition with patients
- Only 15% of schools of public health require genomics as part of core curriculum (2004 study)
- More than half of consumers are aware of genetic risk factors in multifactorial disease but do not necessarily have clear understanding of how these factors influence health
- Need to be vigilant that the benefits of genetics and genomics are realized equitably across diverse population groups
- In spite of efforts by federal agencies, professional societies, and advocacy groups, deficiencies in genetics education and training remain

Policy Questions

1. What initiatives or programs are available to primary care clinicians, public health providers, and consumers to enhance genetics education and training? Are these programs adequate and current? Can these programs serve as models for training other groups?

2. What are the barriers to adequate genetics education and training and what role can the Federal government take to eliminate these barriers?

Policy Questions (continued)

3. What steps can the Federal government take to promote and support diversity and cultural competency of health care and public health professionals?

4. How can the Federal government work effectively with State-level organizations to address incorporation of human genetics and genomics into accreditation, licensure, and certification processes?

Policy Questions (continued)

5. What steps should the Federal government take to assure that patients and consumers are receiving appropriate education in genetics and genomics?

   What can we learn from a range of approaches to genetics education, including the work of health communication researchers, clinician-educators, lay health educators, and industry?

Policy Questions (continued)

6. Promotional materials for genetic services that are directed to consumers and health professionals are currently being used, intentionally or unintentionally, as consumer and provider educational materials. Do FDA requirements for medical device promotional materials also apply to these cases of laboratory developed genetic tests? Should there be Federal oversight?

Possible Action Steps

Short Term Action

- Confer with FDA officials to discuss requirements for medical device promotional materials and whether these requirements would be enforced for laboratory-developed tests
- Work with the Personalized Medicine Coalition and industry representatives to encourage the development of voluntary standards for promotional materials

In Depth Report on

- Genetics education and training needs, in progress; report expected in 2010
- Upon completion, assess whether the report's findings and recommendations also apply to a broader constituency, such as laboratorians, health care administrators, payers, and policymakers. If not, an addendum report should be considered

Cluster 4
Informed Consent, Privacy, and Discrimination Issues that Relate to Genomic Data Sharing

Topic Statement

- Genetic technologies and genetic/genomic research are posing challenges for informed consent and privacy
  - large-scale, population-based research studies may uncover clinically meaningful information for individual participants and may raise group harm issues
  - identifiability of whole genome data is evolving
- Evolving research paradigms and growth of personalized medicine may necessitate new conceptualization of informed consent
  - application in both research and clinical practice
  - additional vigilance regarding privacy protections and discrimination

Background

- Emerging scientific and technical opportunities
- Current ethical requirements and safeguards
- Comprehension and public attitudes
- Relevant activities
  - NBAC (1999)
  - PRIM&R white paper (2007)
  - SACGHS LPS report (2007)
  - AHIC recommendations (2008)
  - ACHDNC planned study related to newborn screening (2009)
  - Agency efforts
    - AHRQ toolkit in development
    - NIH public communication plans

Policy Questions

1. Are there new issues regarding privacy and discrimination raised by the increase in acquiring, collecting, and storing (including in electronic health records) genetic and genomic information (including epigenetic and metabolic data)?

2. How can the public be engaged to understand and to delineate the issues of importance to them so that the consent process can be adequate and meaningful?

3. What can we learn about consent needs from existing large-scale population studies?

4. How can the consent process be improved, e.g., by employing strategies such as “teach-back”? How will information obtained in research be entered into personal health records and what are the implications for consent?

Policy Questions (continued)

5. What is the role of SACGHS and HHS in these discussions and review of existing policies and practices?

6. What are the implications for informed consent and privacy/confidentiality of recent developments in using a statistical method for resolving individual genotypes within a mix of DNA samples or data sets containing aggregate single-nucleotide polymorphism data?

7. How do HIPAA and GINA affect the informational elements that should be included in the informed consent process?

Possible Action Steps

Monitor

- Monitor the rulemaking process for implementation of GINA for issues relevant to informed consent related to genomic data sharing

Short Term Action

- Solicit public input on concerns pertaining to privacy and confidentiality of data collected and stored in large-scale genomics studies

In-depth Report on

- Recommended elements of disclosure for use in the informed consent process for research involving the sharing of genome datasets, either through SACGHS or in cooperation with NIH, OHRP, and FDA
- Mechanisms for risk mitigation (i.e., methods for ensuring risks are minimal)

Cluster 5
Implications of Consumer-Initiated Use of Genomic Services

Topic Statement

- Number of personal genomic services marketed directly to the public has increased over past few years
- Without direct involvement of a personal health care provider, a comprehensive consumer protection strategy may be needed
- Concerns include
  - the relative value of the information provided, will these tests improve health
  - the level of consumer understanding
  - the provider community's ability to understand and translate information for patients
  - the potential risks of misuse of information by consumers or third parties (e.g., insurers, employers)

Background

- Genome services companies differ vastly in their practices and intentions; offer diverse DTC genomics services ranging from risk assessment for chronic disease to recreational uses (e.g., matchmaking)
- Past SACGHS activities
  - 2004 and 2006 letters to the Secretary expressing concerns about advertising claims made by companies offering DTC genetic services
  - information-gathering session at July 2008 SACGHS meeting to explore current landscape of genomic services
- Considerable number of U.S. and international activities related to consumer-initiated genomic services including research studies, development of educational resources, and workshops to explore issues such as evaluation of the added value of genomic profiles to promote health and prevent disease

Policy Questions

Oversight

1. Should genomic tests be regulated similarly to other complex laboratory tests?
2. Previous SACGHS reports have assessed oversight needs in genetic testing. Are these recommendations sufficient for the issues related to direct-to-consumer genomic services?

Concerns about Clinical Validity and Utility

3. What are the best formulas for calculating risk?
4. What criteria should be used to determine whether the association between a particular genetic marker and phenotype is strong enough for that marker to be included in genetic testing?

Policy Questions (continued)

Concerns about Clinical Validity and Utility

5. Should there be standards for formatting raw genetic data from whole genome scans (e.g., reporting the NCBI dbSNP build, rs number, strand direction for each variant, or location of variants within reference sequence from the NCBI refseq database)?
6. How will clinical validity and utility of such tests be assessed and communicated to consumers?
7. When have sufficient data been produced to change previously recommended risk calculations?

Policy Questions (continued)

Issues for Consumers and Health Care Professionals

8. Are requirements for public education and informed consent needed before testing?
9. What are the appropriate roles and responsibilities of health care providers, consumers, and public health programs in this nontraditional approach to testing?
10. Do personal genome services fill a specific health care and/or public health need?
11. Are providers and consumers adequately prepared for the information provided by such services?
12. What are the benefits and potential drawbacks of DTC personal genome services?

Policy Questions (continued)

Issues for Consumers and Health-Care Professionals (cont'd)

13. How will the health care system and health care providers be affected by the availability of personal genome services?
14. What is known about consumer interest in personal genome services and consumers' understanding of these services?
15. What criteria should be considered in determining the value of personal genome services?
16. What are the criteria for determining whether previously tested individuals should be contacted to inform them of a modified risk and when should it be left up to individuals to initiate such follow-up?

Policy Questions (continued)

DTC Marketing and Advertising

17. What criteria do companies offering personal genome services consider before marketing services?

Privacy and Discrimination Concerns

18. What are the privacy concerns?
19. What cautions/benefits should consumers consider when sharing their genomic information with others such as family members, a social network, or clinicians?
20. Does GINA apply to the genetic risk information provided by companies offering personal genome services? Are these companies covered by GINA?

Disparities

21. Could personal genome services exacerbate health disparities?

Possible Action Steps

Monitor

- Monitor outcomes of federal and nonfederal workshops and upcoming NIH GeneScans website, then assess possible SACGHS actions

Short Term Actions

- Promote the development of a checklist that patients could use to evaluate genomic services

Possible Action Steps (continued)

Brief Report on

- Selected key issues not addressed by other federal and nonfederal efforts, such as the effect of direct-to-consumer testing on health disparities

In-Depth Report on

- The implications of consumer-initiated use of genomic services

Cluster 6
Public Health Applications of Genomics Research

Topic Statement

- **Public health genomics**: a multidisciplinary field concerned with the effective and responsible translation of genome-based knowledge and technology to improve population health
- **Focus**: policies and actions needed to promote health and prevent and control disease in populations; consider interplay of genes, environment, and behaviors; ensure that the benefits of genetics and genomics are realized equitably across diverse population groups
- **Core functions**: assessment, policy development, and assurance; research forms the center of this core

Background

- **Assessment**: systematic collection, analysis, and dissemination of information on the health of community – epidemiologic and laboratory research, investigation and monitoring of identified community health problems and risk factors

- **Policy development**: promotes translation of advances in human genetics into disease prevention opportunities – policy and communications, public education and health promotion and disease prevention policies for clinical and population settings

- **Assurance**: bolsters public's confidence that genetic information is used appropriately and genetic services meet agreed-upon goals for effectiveness, accessibility, and quality –research; impact evaluation and quality; enforcing laws, policies, and standards; and assuring a competent workforce

Policy Questions

1. How do characteristics of diverse systems of health care management and delivery influence the provision of genetic tests and subsequent clinical or preventive services?

2. What are the leading opportunities and/or responsibilities for the public health system to contribute to the development and implementation of new genomic knowledge and technologies to improve health, prevent disease, and address health disparities? Specifically, what are the:

Policy Questions (continued)

2. (continued)

   a) Opportunities, challenges, and benefits of incorporating genomics into existing and future public health investigations and surveillance systems to advance knowledge?

   b) Opportunities, challenges, and responsibilities for incorporating evidence-based genomics knowledge and technologies into public health programs to improve health and prevent disease?

   c) Opportunities for the governmental public health infrastructure to partner with the health delivery system, employers and businesses, communities, academia, the media and others?

Policy Questions (continued)

3. What steps must be taken to address ethical, legal, and social issues in public health genomic research and practice?
   a) How does informed consent for genetic testing in public health differ from informed consent for other public health services and in clinical practice? Under what circumstances is new consent for archived specimens needed for public health investigators?
   b) What are the immediate and long-term benefits and risks of population-based disease registries?
   c) How can concerns about potential stigmatization of population groups that results from research on testing programs be addressed?

Policy Questions (continued)

3. (continued)

   d) What policies should be in place to share large amounts of data collected through gene-environment-disease association studies?

   e) An emerging concern is that as technologies evolve, it will become possible to test for multiple layers of biological changes, which reveal chinks in bodily integrity before classic clinical symptoms emerge. Will advances in technology and knowledge shift current conceptions of “injury” in toxic tort suits or the “pre-existing condition” exclusion in the Genetic Information Nondiscrimination Act (GINA)?

Policy Questions (continued)

4. What tools are needed to understand how genes and environmental factors interact to perturb biological pathways and cause injury or disease?
5. How does the federal investment in genomics encourage translation into population health benefits?
6. Is it cost effective to tailor interventions based on genetic information?

Policy Questions (continued)

7. What steps must be taken to assure a competent public health workforce with a sufficient knowledge base and skill to ensure the appropriate use of genetic information to promote health and prevent disease, as well educate the general public to be informed consumers of genomic applications?

   How can public health agencies prepare their workforce and constituencies to ensure that information about gene-environment interactions is used appropriately?

Possible Action Steps

Short Term Actions

- Organize sessions at SACGHS meetings to explore the field of public health genomics policy questions associated with advances in understanding gene-environment interactions and for an in-depth discussion of the potential for genetic and genomic testing to exacerbate or lessen health disparities

- Perform a systems review of relevant agencies to assess mechanisms to disseminate information about the distribution of genotypes in different populations and assure the effectiveness, accessibility, and quality of services

- Use SACGHS as a forum to promote collaboration within and between DHHS agencies for efforts such as preventing the stigmatization of individuals, families, or populations at-risk for or with genetic conditions and implementing an assessment process that will provide guidance for how and when genetic tests can be used to promote health and prevent disease

Possible Action Steps (continued)

Brief Report on

- Selected public health topics such as the impact of genetic/genomic testing on health disparities, how characteristics of different health care systems influence the provision of genetic tests and subsequent clinical or preventive services, building a competent public health workforce to ensure the appropriate use of genetic information to promote health and prevent disease, or whether it is cost effective to tailor interventions based on genetic information

In-depth Report on

- Issues related to public health genomics, particularly in the areas of disparities, gene-environment interactions, and population-level testing

Cluster 7
Genetics and the Future of the Health Care System

Topic Statement

- Focus on Two Broad Inquiries Concerning Genomic Medicine/Personalized Health Care
  - Achievability, costs, and value of personalized health care
  - Infrastructure changes needed to foster personalized medicine

Background

- U.S. Health Care: Its Present and Possible Future
  - Present system has high costs and poor outcomes:
    - U.S. spends more per capita on health care than any other country
    - Current trend in spending is unsustainable
    - U.S. is far behind other developed nations in measures of population health

Background (continued)

- Value, costs, and achievability of personalized health care
  - Genomic medicine stands to benefit public health
  - New technologies may decrease or increase costs
  - Considerable research will be needed to understand molecular basis of disease and to translate this understanding to health care
  - Need for greater understanding of genetics/epigenetics of various subpopulations for genomic medicine to benefit all people

Background (continued)

- Needed infrastructure changes
  - More cost-effective health care delivery and increased genetic counseling
  - Increased clinical lab workforce
  - Deployment of health information technology
  - Systems to monitor and evaluate effectiveness of genomic medicine
  - Government incentives for business pursuit of diagnostics and genetic therapies

Policy Questions

1. What type of process or procedure should the Government employ in deciding how best to invest resources in genomic medicine?

2. Should the Government promote the adoption of a new approach to health care delivery? What particular system should the government promote? What other changes in health care infrastructure should the government promote to favor personalized health care?

3. Should financial incentives be offered to attract more students to clinical laboratory careers? If so, what mechanisms might the Government use to create these incentives?

Policy Questions (continued)

4. Apart from existing efforts by the Government to develop health information technology, what else should the Government do to promote the development of electronic health records and the digital storage of health data?

5. How would quality improvement systems be used to improve genetic health technologies, to ensure their delivery to the appropriate patients, and to monitor their public health benefit? What surveillance systems are needed?

Policy Questions (continued)

6. What steps can the Government take to assist businesses that wish to pursue the development of genetic diagnostics and targeted therapeutics?

7. How best can the Government use it resources to learn about differences in genetic and epigenetic variations among U.S. subpopulations?

Possible Action Steps

Short Term Actions

- Organize a panel of chief medical officers from prominent health plans and other experts to discuss how they see the future of health care unfolding

Brief Reports on

- Procedures the Government could use in deciding how best to invest resources in genomic medicine
- Incentives to attract more students to clinical lab careers

Possible Action Steps (continued)

Brief Reports on

- Tools and systems to improve genetic health technologies, to ensure their delivery to the appropriate patients, and to monitor their public health benefit; examination of how genetics may improve the effectiveness and efficiency of the health care system
- Possible incentives the Government could provide for businesses to pursue genetic diagnostics and targeted therapeutics
- Steps that the Government may take to accelerate the development, adoption and integration of health information technology

Possible Action Steps (continued)

In-depth Reports on

- Proposed reforms to health care delivery, including the medical home model, customized care centers, and genetic counseling as a customary part of health care

- How the Government could help to ensure that appropriate resources and infrastructure are available to learn about differences in genetic and epigenetic variations among various geographic and ethnic U.S. subpopulations