DEPARTMENT OF HEALTH & HUMAN SERVICES

Public Health Service

Secretary's Advisory Committee on
Genetics, Health, and Society
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RADM Penelope Slade-Sawyer, P.T., M.S.W.
Director
Office of Disease Prevention and Health Promotion (ODPHP)
U.S. Department of Health and Human Services
1101 Wootton Parkway, Suite LL100
Rockville, MD 20852

Dear RADM Slade-Sawyer:

I am writing on behalf of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) in support of the recommendations forwarded by the Healthy People 2020 Genomics Working Group and to urge ODPHP to consider incorporating genomics more broadly as a component of a range of objectives proposed for Healthy People 2020. The incorporation of genomics into Healthy People 2020 signals the emergence of the field's relevance to public health. The specific objectives that the Working Group identified—implementation of the recommendations from the U.S. Preventive Service Task Force on genetic risk assessment and BRCA mutation testing for breast and ovarian susceptibility and from the Evaluation of Genomic Applications in Practice and Prevention Working Group on genetic testing strategies in newly diagnosed individuals with colorectal cancer—have the potential to improve public health strategies in combating these three cancers.

Genetic and genomic tests are expected to become increasingly prevalent in the coming years. The hope is that these tests will not only help in the early diagnosis of common diseases, but also identify genetic risk factors that can in turn be used to guide preventive measures tailored to each individual's genotype. Similarly, pharmacogenomics tests promise more safe and effective prescribing of drug treatments for all segments of the population. Even without genetic tests, collection of family health history can provide clues to a patient's inherited risk for common diseases. Although many providers are using family history tools in clinical care, they lack evidence-based guidelines on how to use the information in clinical decisionmaking. SACGHS has made recommendations to the Secretary of Health and Human Services about the importance of assuring the safety of genetic tests; establishing research programs to determine the analytical validity, clinical validity, and clinical utility of genetic and genomic tests; using family health history tools; and incorporating genetic and genomic information in electronic health records. These recommendations would help provide needed guidance for clinical practice and disease and risk assessment and ensure the widespread translation of genomic discoveries into public health practice.

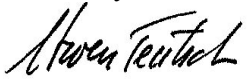
Given the promise and potential value of genomic tests in reducing the burden of common diseases and, based on previous SACGHS recommendations to the Secretary of Health and Human Services relating to oversight of genetic testing and pharmacogenomics, we urge ODPHP to consider adding objectives to

address gaps preventing the full realization of these potential public health benefits. These objectives would address the need for research in the areas of family history and the analytical validity, clinical utility, and comparative effectiveness of genetic and genomic tests, and resources for the dissemination of research findings. We also recommend that several existing objectives be clarified to ensure the safety and validity of genetic/genomics tests and the integration of genetic and genomic data as a component of electronic health records and Health Information Exchange. Our proposed objectives and objective clarifications are relevant to, and could be incorporated into, the Healthy People categories of Genomics, Educational and Community-Based Programs, Medical Product Safety, and Health Communication and Health IT, respectively. The rationale and evidence supporting these revisions are discussed in more detail in the enclosed paper.

In addition, we also recommend that ODPHP consider adding information about genomic and genetic technologies that are likely to affect public health in the future. To accomplish this task, the Genomic Working Group could consider the relevance of genomics to other categories of Healthy People 2020 and include a forward-looking narrative discussion of the relevance of genomics to many areas of public health. Including such information will help raise awareness of the role of genomics in clinical care and public health practice and establish genomics as a viable category for the development of future Healthy People objectives. The Healthy People categories in which genetic/genomic references would be appropriate are Access to Health Services, Cancer, Educational and Community-Based Programs, Environmental Health, Family Planning, Health Communication and Health IT, Public Health Infrastructure, Oral Health, Respiratory Diseases and Social Determinants of Health.

SACGHS commends ODPHP for including genomics as a new category in Healthy People 2020, and we appreciate this opportunity to call further attention to the role that genetics and genomics will play in the future of Healthy People. Thank you for considering our proposals and best wishes in carrying out your important work.

Sincerely,



Steven Teutsch, M.D., M.P.H.
SACGHS Chair

Enclosure: *SACGHS Proposals for Healthy People 2020*

Secretary's Advisory Committee on Genetics, Health, and Society
Proposals for Healthy People 2020

HP Category: Genomics

Proposed New Objective 1: Increase the number of comparative effectiveness research (CER) studies that address the clinical utility of genetic and genomic tests and the collection of family history. In addition, CER studies should incorporate genomic analysis of study participants. Specifically:

- CER studies should include examination of the clinical validity and clinical utility of genetic and genomic tests. While new studies will be needed to help establish how genetic testing affects health outcomes, there are existing data from a variety of retrospective studies that could be used to demonstrate clinical utility of some tests.
- CER studies should include genomic analyses of study participants so that any guidelines disseminated after a study can be made specific to particular genetic subpopulations.
- CER studies on family health history should be conducted towards development of professional guidelines on how to use family history in clinical decisionmaking.

Rationale: Clinical utility and comparative effectiveness determinations help guide clinical care, establish clinical guidelines, and inform coverage decisions. Given the growing role that genetic testing is expected to play in the future of health care, assessing the clinical utility and comparative effectiveness of various genetic tests will be a constructive way to ensure high-quality health care and potentially control future health care costs. However, quality improvement processes are needed to ensure that genetic tests are delivered consistently to appropriate patients. Furthermore, an ongoing process is needed to identify opportunities for improving the use of genetic testing, including the collection of post-market outcome data.

Comparative effectiveness studies should recognize that the effectiveness of treatments and preventive interventions may vary among different genetic subpopulations (i.e., an intervention that is ineffective for the general population may be effective for a subpopulation with a particular genotype and vice versa). Therefore, whenever possible depending on the study design, federally funded comparative effectiveness studies should take account of the genomic analysis of the study participants so that any guidelines disseminated after a study can be made specific to particular genetic subpopulations. Incorporating genomic information into comparative effectiveness studies will ensure that particular groups are not denied access to clinical and preventive health services that are effective for them but not others. In addition, all studies involving genomic assessments should have safeguards in place to prevent genetic discrimination, such as denial of life insurance, long-term care insurance, or disability insurance.

In addition to the lack of underlying utility studies (and the lack of standards for judging those studies), there is no existing government or private-sector system capable of efficiently conducting utility assessments for the large number of emerging genetic tests. The practical effect of this series of problems is that public and private insurers and health care providers are unsure of the value of these tests and their appropriate use. Thus, problems in assessing the clinical utility of genetic tests are impeding the appropriate integration of genetic tests into health care.

Like genomic information, family health history can provide clues to a patient's inherited risk for common diseases. Family history tools are already commonly used in clinical care, but for the most part, providers lack evidence-based guidance on how to use the information in clinical decision making.

Comparative effectiveness studies of family health history would provide needed guidance on how to incorporate this low-cost clinical tool into health care practice.

Data Sources: "SACGHS Progress Report and Future Directions, January 2009" http://oba.od.nih.gov/oba/SACGHS/SACGHS_Progress_and_Priorities_Report_to_HHS_Secretary_Jan_2009.pdf; and "SACGHS Letter to the Institute of Medicine's Committee on Comparative Effectiveness Research Priorities, Mar, 2009".

Proposed New Objective 2: Increase awareness among health care providers regarding the distinction between a genetic test that has been found to be clinically useful and a genetic test for which there is little or no evidence of utility. Increase awareness about how genetic testing affects health outcomes. Specifically:

- Develop evidentiary standards, data sources, and evidence-based methods applicable to genetic testing to help establish clinical utility and guide the effective translation of genetic research into practice. Disseminate study findings, including negative findings, through publications, meetings, and an information clearinghouse.
- Increase education and guidance for clinicians, laboratory personnel, and other health care professionals to ensure the accurate use and interpretation of genetic tests.
- Provide training on the effective use of electronic health records and clinical decision support tools in the pre-analytical and post-analytical phases of genetic testing.
- Conduct ongoing public health surveillance such as surveys of patients, providers, and the general population to monitor the uptake and use of genetic tests and the determinants of care.

Rationale: Technical advances in genetic testing must be accompanied by accurate interpretation and communication of genetic test results. Professional recommendations, including those from such groups as the American College of Medical Genetics and the U.S. Preventive Services Task Force, provide information to practitioners about ordering genetic tests and reporting results.[1] Organizations such as the National Coalition for Health Professional Education in Genetics have engaged in efforts to enhance clinician understanding of genetic testing and its appropriate use.[2] Yet there are insufficient data about how well practitioners order, conduct, and interpret genetic tests and the extent to which genetic test results are used appropriately to support clinical decisionmaking. Most practitioners are unfamiliar with guidelines for the appropriate use of genetic tests, and few processes have been implemented, evaluated, or enforced to support practitioners in this regard.

Data Source: "U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services Report of the Secretary's Advisory Committee on Genetics, Health, and Society, April 2008" http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf and "Realizing the Potential of Pharmacogenomics: Opportunities and Challenges: Report of the Secretary's Advisory Committee on Genetics, Health, and Society, May 2008". http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_PGx_report.pdf
Potential Data Sources (and measures): Pre-Market Approval Database, FDA; Electronic Medical Records, CMS.

[1] American College of Medical Genetics Web site. "American College of Medical Genetics Practice Guidelines." http://www.acmg.net/AM/Template.cfm?Section=Practice_Guidelines&Template=/CM/HTMLDisplay.cfm&ContentID=2257. Accessed on March 20, 2008

[2] National Coalition for Health Professional Education in Genetics Web site. "Contracts and Grants." See http://www.nchpeg.org/content.asp?dbsection=contracts#1. Accessed on March 20, 2008.

HP Category: Medical Product Safety

MPS HP2020-4: (Developmental) Increase the utilization of safe and effective medical products that are associated with predictive biomarkers.

Objective Clarification: It is important for this objective to recognize that genetic and genomic tests that use predictive biomarkers (e.g., pharmacogenomic tests) are also medical products, and their safety and effectiveness need to be assured.

Rationale: Analytical validity of a genetic test refers to a test's ability to measure the analyte or genotype of interest accurately and reliably, and clinical validity refers to a test's ability to detect or predict the associated disorder (phenotype). Analytical and clinical validity must be established for genetic testing technologies through the development of assay validation tools, improved data sharing among researchers, and establishment of evidentiary standards. Prospective data of a test's analytical and/or clinical validity, however, are often unavailable or incomplete. In addition, there are numerous challenges to demonstrate analytical and clinical validity, such as the lack of materials for proficiency testing and quality assurance. Funding is required for the development and characterization of reference materials, methods, and samples (e.g., positive and negative controls and samples from different ethnic/geographic populations) for assay, analyte, and platform validation; for quality control and performance assessment; and for standardization. Collection of post-market data and sharing information among laboratories would assist with establishing the clinical validity of genetic tests. Better coordination of public and private sector activities has the potential to strengthen the oversight of genetic testing through complementary and consistent State and Federal requirements for establishing analytical validity, quality assurance, and clinical validity.

Data Source: "U.S. System of Oversight of Genetic Testing: A Response to the Charge of the Secretary of Health and Human Services Report of the Secretary's Advisory Committee on Genetics, Health, and Society, April 2008" http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf
Potential Data Sources (and measures): Pre-Market Approval Database, FDA; Electronic Medical Records, CMS.

HP Category: Health Communication and Health IT

HC/HIT HP2020-11: (Developmental) Increase the proportion of providers who use health information technology to improve individual and population health.

HC/HIT HP2020-12: (Developmental): Increase the proportion of providers and governmental health agencies that use advanced connectivity to optimize electronic health information exchange to improve individual and population health.

Objective Clarification for HP2020-11 and 12: Genetic and genomic information is fundamental information that will need to be integrated into general health care practice rather than as ad hoc specialty information. During the development of software to support electronic health records (EHRs) and adoption of electronic connectivity in health care systems and private practices, architecture should assure the ability of the EHR to incorporate and facilitate the use of validated genetic/genomic information across the record. We recommend that:

- Any definition and certification of meaningful use of the EHR must be sufficiently flexible to accommodate changes in medical practice that will result from evidence-based practice research.

- EHRs must be dynamic and structurally ready to incorporate genetic/genomic information such as laboratory test results and pharmacogenomic-informed prescribing as future technologies reveal advances that are not currently recognized.
- EHRs should incorporate family history and newborn screening results.
- EHRs should also acquire and incorporate clinical data to support comparative effectiveness research in genetics, genomics, and personalized medicine.
- EHRs should optimize the use of family history in clinical care through the incorporation of appropriate clinical decision support tools and pedigrees within the EHR. This effort would complement ongoing activities across the Department of Health and Human Services as well as the Department of Defense and the Department of Veterans Affairs to deploy robust family history collection tools in the clinical environment.

Rationale: SACGHS supports the further development of clinical decision support tools for point-of-care use, particularly for dynamic health fields such as genetics. Additional resources will be needed to design and support programmatic and research efforts for clinical decision support in the ordering, interpretation, and application of genetic tests. Clinical decision support for genetic/genomic information in the context of the EHR has the power to prevent potential harms to patients due to misinterpretation of genetic test results and help primary care physicians provide adequate and appropriate counseling, thus enhancing patient care. This goal cannot be met unless genetic/genomic information is available in EHRs.

Proposed Data Source: "Comments of the Secretary's Advisory Committee on Genetics, Health, and Society to the Health Information Technology Policy Committee on the Definition of Meaningful Use of an Electronic Health Record, June 26, 2009"
http://oba.od.nih.gov/oba/SACGHS/SACGHS%20Comments%20on%20MU%20EHR%206-26-09.pdf