Comments of the Secretary's Advisory Committee on Genetics, Health, and Society on the CMS and ONC Rules to Implement Electronic Health Records March 15, 2010

The Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) appreciates this opportunity to comment on the Centers for Medicare & Medicaid Services (CMS) Proposed Rule (PR) *Medicare & Medicaid Programs; Electronic Health Record Incentive Program* and the Office of the National Coordinator for Health Information Technology (ONC) Interim Final Rule (IFR) *Health Information Technology: Initial Set of Standards, Implementation Specifications, and Certification Criteria for Electronic Health Records*. SACGHS advises the Secretary of Health and Human Services on issues related to the use and development of genetic technologies and has focused particularly on the appropriate integration of genetic technologies into health care and public health. These comments are an extension of comments we provided on the definition of meaningful use to the Health Information Technology (HIT) Policy Committee in June 2009. It is worth noting that in the eight months since SACGHS submitted its comments, there have been a number of developments that further attest to the rapidity with which genetic/genomic information is becoming a part of clinical care.

General Comments

SACGHS wishes to commend ONC for developing standards, implementation specifications, and certification criteria for electronic health record (EHR) technology and CMS for the criteria that must be met by an eligible professional and hospital to qualify for incentive payments. We recognize the complicated nature of this work, particularly given the diverse needs and challenges of reforming our health care system. However, we are concerned that the rules do not go far enough in providing incentives for the incorporation of genetic/genomic information in EHRs. We recognize the interrelated nature of these two rules and are making general comments that apply to both followed by changes and insertions specific to each document.

SACGHS agrees that defining meaningful use of an EHR is critically important to achieve the ultimate goal of enabling significant and measurable improvements in population health. We understand that a phased approach may be necessary in building up to a more robust definition of meaningful use based on anticipated technology and capabilities development. However, we urge CMS and ONC to recognize that the earliest possible incorporation of genetic/genomic information into the EHR will play an important role in realizing the promise of personalized and evidence-based medicine. Nine of the top 10 causes of death in the United States, including the three identified as priority conditions in the CMS PR and the ONC IFR—diabetes, coronary vascular disease, and heart disease—have contributing genetic components.[1] As such the capacity to capture genetic/genomic and family history information in EHRs will facilitate progress on prevention of these diseases.

Any definition of meaningful use of EHRs must be sufficiently flexible to accommodate changes in medical practice that will result from evidence-based research, and EHRs must be dynamic and structurally ready to incorporate new genetic/genomic findings. The importance of newly emerging genetic/genomic data is highlighted by the U.S. Preventative Services Task Force recommendations

[1] Centers for Disease Control and Prevention. *Human Genome Epidemiology Network.* See http://www.cdc.gov/genomics/public/faq.htm. Accessed on March 10, 2010.

regarding genetic risk assessment and *BRCA* mutation testing for breast and ovarian cancer susceptibility and the 2004 formation of the Centers for Disease Control and Prevention Evaluation of Genomic Applications in Practice and Prevention (EGAPP) workgroup. EGAPP's charge is to review the evidence of the validity and utility of genetic/genomic tests that are in transition from research to clinical and public health practice, and the workgroup has published several important studies regarding the efficacy of genetic testing in specific circumstances.[2,3]

As the process moves forward to implement meaningful use, we encourage CMS and ONC to recognize the importance of enabling the incorporation of genetic/genomic information, family history, and newborn screening results in EHRs. Efforts to harmonize HIT data standards and the interoperability of the HIT infrastructure must include these elements to prevent proliferation of fragmented and incompatible databases, increased costs to the health care system, and impediments to knowledge generation, data collection, analysis, and research. In this regard, we call your attention to the work that has already been carried out by the American Health Information Community's (AHIC) Personalized Healthcare and Clinical Decision Support Workgroups during 2007 and 2008 as well as the ongoing initiatives from the Clinical Data Interchange Standards Consortium (CDISC), Healthcare Information Technology Standards Panel (HITSP), and the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC).

To help address recognized gaps in the current EHR landscape relative to representing genetic/genomic information, family history, and newborn screening results, AHIC developed use cases to provide a framework for EHR certification standards in these areas. We would encourage CMS and ONC to ensure that standards previously approved by HITSP are used as standards for certified EHRs. HITSP's Personalized Healthcare Interoperability Specifications were approved and released by HITSP in December 2008. These specifications describe the incorporation of family history and genetic/genomic laboratory ordering mechanisms and results, which can then be used to provide personalized treatment specific to genetic makeup. Many of HITSPs interoperability specifications have gone through the process of approval and recognition by the Secretary, however, the Personalized Healthcare Interoperability Specifications standard has not. We urge CMS and ONC to facilitate adoption of HITSPs interoperability specifications for personalized health care and family history.

With regard to the specific criteria for meaningful use proposed for Stage 1, we agree that the care goal "Apply clinical decision support at the point of care" is especially important in the realm of genetics given the rapid pace at which the field is moving. Traditional educational mechanisms are insufficient to keep providers abreast of genomic medicine and thus clinical decision support for genetic/genomic information in the context of the EHR has the potential to prevent harms due to misinterpretation of genetic test results and help eligible professionals provide adequate and appropriate counseling. Clinical decision support tools, made available at appropriate times, will enhance patient care. However, this goal cannot be met unless genetic/genomic information is available as structured, coded, and computable data in the EHR. As such, we encourage development of technical standards for genetic/genomic and family history data for incorporation in certified EHRs to allow decision support tools to be able to respond to this dynamic and rapidly changing health field. The outcome of these technical standards would enable

[2] Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group. Recommendations from the EGAPP Working Group: genetic testing strategies in newly diagnosed individuals with colorectal cancer aimed at reducing morbidity and mortality from Lynch syndrome in relatives. *Genetics in Medicine* 2009. 11(1):35-41.

[3] Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group. Recommendations from the EGAPP Working Group: can tumor gene expression profiling improve outcomes in patients with breast cancer? *Genetics in Medicine* 2009. 11(1):66-73.

meaningful users to receive appropriate alerts and care suggestions based on genetic/genomic information.

Specific Comments that Apply to Both Rules

The field of pharmacogenomic testing is rapidly developing and an increasing number of validated tests is expected to be available in the near future. Such testing promises to improve patient health and safety through the reduction of adverse drug reactions and enhancement of drug effectiveness, and it may help reduce health care costs through more targeted use of medications. For example, genetic testing is now standard of care prior to prescribing abacavir used to treat HIV/AIDS, and to determine the level of expression of the *ERBB2* gene, to guide decisionmaking in the treatment of breast cancer by identifying which patients should receive the chemotherapeutic agent herceptin. New authorities under the Food and Drug Administration Amendments Act of 2007 (FDAAA) are expected to lead to more post-market safety studies, studies that will increasingly involve genotyping to determine whether a genetic variation is involved in an adverse drug reaction. EHRs with the capacity to incorporate genetic/genomic information will facilitate the conduct of post-market safety studies. The Food and Drug Administration (FDA) will require label changes on medications where known genetic mutations influence the drug's efficacy and safety. For example, FDA now requires that the label for Tegretol® include a warning of serious skin reactions in persons carrying the *HLA-B*1502* allele.[4] CMS is exploring which pharmacogenomic tests have sufficient evidence to warrant Medicare coverage. As clinical applications of pharmacogenomic research continue to emerge, EHRs and meaningful use of EHRs must be able to incorporate these advances. Thus, we would recommend that the objective "Implement drug-drug, drug-allergy, drug-formulary checks" include pharmacogenomic-informed prescribing in the following sections of the Rules:

- ONC IFR: Proposed meaningful use Stage 1 objective to "Implement drug-drug, drug-allergy, drug-formulary checks and pharmacogenomic-informed prescribing," (page 2026).
- CMS NPRM: Stage 1 objective to "Implement drug-drug, drug-allergy, drug-formulary checks and pharmacogenomic-informed prescribing," (page 1867).

The objective to "Incorporate clinical lab test results into EHR as structured data" should explicitly reference genetic/genomic test results. In addition to enhancing the quality and safety of care and ensuring easy access to genetic test results, incorporating genetic tests results—coupled with clinical decision support tools that alert clinicians to prior tests—would eliminate unnecessary duplicative testing for heritable mutations, which contributes to increased healthcare costs.[5] Interpretation of genetic test results is also a well recognized problem. Including genetic test results in the EHR allows providers to access point-of-care education and interpretation aids that would address this issue as well as support efforts by the CDC to create an interactive genetic test report. There are currently more than 1,600 genetic tests available from clinical laboratories.[6] International data standards for genetic/genomic test results are emerging and to ensure that these tests are well documented in the EHR, they must be included in the infrastructure of the EHR. As such, SACGHS recommends that genetic/genomic test results be explicitly referenced in the following sections of the Rules:

[4] Food and Drug Administration. See http://www.accessdata.fda.gov/scripts/cder/drugsatfda/index.cfm?fuseaction=Search.Label_ApprovalHistory. Accessed on March 5, 2010.
[5] Riegert-Johnson DL, Macaya D, Hefferon TW, and Boardman LA. The incidence of duplicate genetic testing. *Genetics in Medicine* 2009. 10(2):114-116.
[6] GeneTests website. See http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests. Accessed on March 8, 2010.

- ONC IFR: Proposed meaningful use objective to "Incorporate clinical lab and genetic/genomic test results into EHR as structured data," (page 2026).
- CMS NPRM: Stage 1 objective to "Incorporate clinical lab and genetic/genomic test results into EHR as structured data," (page 1868).

There is also a need to optimize the use of family history in clinical care to include development of clinical decision support tools and the ability to utilize pedigrees within the EHR. These steps would complement ongoing efforts underway across the Department of Health and Human Services as well as the Department of Defense, Indian Health Service, and the Department of Veterans Affairs to deploy robust family history collection tools in the clinical environment. Interest in the use of family history has increased and the weight of evidence linking the collection and use of family history information and improved health is growing.[7] Family history information is also critical if not essential for the appropriate use and accurate interpretation of genetic tests used for risk assessment.[8,9] A detailed family history is now recognized as an important tool in primary prevention and early intervention with particular relevance for sub-populations at highest risk of genetic disease and underserved communities.[10] Its incorporation into the medical record will require focused mechanisms to provide a tool that is easy to interpret by eligible professionals. EHRs should have the capability to incorporate both family history and genetic/genomic data at their inception, and the definition of meaningful use in 2011 should be expanded to include use of family history to identify high-risk populations in need of screening. Other uses of family history can be designated as meaningful use in subsequent years. As such, SACGHS recommends that family history be included in the following sections of the Rules:

- ONC IFR: Proposed meaningful use objective, to "Record demographics" "Enable user to electronically record, modify, and retrieve patient demographic and family history data including …" (page 2026).
- CMS NPRM: Stage 1 objective "Record Demographics" insert family history as a bullet (page 1867) and Stage 1 Criteria for Meaningful Use, The health outcomes policy priority, "Engage patients and families in their health care," "Provide patients and families with timely access to data, knowledge, and tools, including family history tools, to make informed decisions and to manage their health," (page 1868)

We commend ONC for including an objective (Table 1, p. 2028, fifth objective) that supports the "capability to provide electronic submission of reportable lab results (as required by state or local law) to public health agencies and actual submission where it can be received." This objective would include laboratory results such as those provided through newborn screening. The same objective is included in the CMS stage 1 objective for hospitals but not for eligible professionals. SACGHS recommends that the same objective also be added to the CMS stage 1 objective for eligible professionals.

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[7] Quillin JM, Ramakrishnan V, Borzelleca J, Bodurtha J, Bowen D, Baer Wilson D. Paternal relatives and family history of breast cancer. *Am J Prev Med* 2006. 31(3):265-8.

[8] Valdez R, Yoon PW, Qureshi N, Green RF, Khoury MJ. Family history in public health practice: a genomic tool for disease prevention and health promotion. *Annu Rev Public Health* 2010. 31:25.125.19.

[9] Vos YJ, de Walle HE, Bos KK, Stegeman JA, Ten Berge AM, Bruining M, van Maarle MC, Elting MW, den Hollander NS, Hamel B, Fortuna AM, Sunde LE, Stolte-Dijkstra I, Schrander-Stumpel CT, Hofstra RM. Genotype-phenotype correlations in L1 syndrome: a guide for genetic counselling and mutation analysis. *J Med Genet* 2009. Published on October 20, 2009 as 10.1136/jmg.2009.071688.

[10] Suchindran S, Vana AM, Shaffer RA, Alcaraz JE, McCarthy JJ. Racial differences in the interaction between family history and risk factors associated with diabetes in the National Health and Nutritional Examination Survey, 1999-2004. *Genet Med* 2009. 11(7):542-7.

- CMS NPRM: Care Goal to "communicate with public health agencies," add the following stage 1 objective for eligible professionals: Capability to provide electronic submission of reportable lab results (as required by state or local law) to public health agencies and actual submission where it can be received.

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Finally, there is a significant inconsistency between the CMS and ONC rules that should be rectified. The CMS rule includes "research" in the stage 1 care goal of improving quality, safety, efficiency and reducing health disparities. The specific objective reads: "Generate lists of patients by specific conditions to use for quality improvement, reduction of disparities, **research,** and outreach" (pages 1855 and 1862) (note that the same language, i.e., including research, should also appear on page 1868). We commend CMS for recognizing the importance of enabling EHRs to facilitate the conduct of research. However, the corresponding certification criteria in the ONC rule (page 2026) should also include "research." This policy approach is consistent with, and particularly important if we are to realize, the priority currently placed on comparative effectiveness research (CER). The Federal Coordinating Council for Comparative Effectiveness Research (FCCCER) Report to the President and Congress[11] recognized the complementary nature of CER and personalized medicine, noting that comparative effectiveness studies can identify different responses in different groups of patients; and identify sub-groups for whom effective therapies do not yet exist and steer research efforts towards strategies for areas of need. Pharmacogenomics was cited as a hallmark of this approach. In the strategic framework for CER, FCCCER identified CER data infrastructure as one of four major activity and investment categories that can be made by the government or other institutions, and specifically identified EHR databases linked to practice-based data networks as an example activity. As such, the SACGHS recommends that "research" be included in:

- ONC IFR: Proposed meaningful use Stage 1 objective to "Generate lists of patients by specific conditions to use for quality improvement, reduction of disparities, and outreach," modify the objective to read: "Enable the generation of lists of patients by specific conditions or individual clinical level data to use for quality improvement, reduction of disparities, research and outreach," (page 2026).
- CMS NPRM: Page 1856, 2nd column, 4th bullet and Stage 1 objectives "Generate lists of patients by specific conditions to use for quality improvement, reduction of disparities, research and outreach," (page 1868) for both eligible professionals and hospitals.

We appreciate the opportunity to comment on these rules and we would welcome the opportunity to provide further input as you move forward to advance the development and adoption of meaningful use of EHRs.

[11] Department of Health and Human Services. Federal Coordinating Council for Comparative Effectiveness Research. *Report to the President and Congress*, June 2009. See www.hhs.gov/recovery/programs/cer/cerannualrpt.pdf. Accessed on February 3, 2010.