

July 31, 2007

The Honorable Michael O. Leavitt

Chairman

American Health Information Community

200 Independence Avenue, S.W.

Washington, D.C. 20201

Dear Mr. Chairman:

The American Health Information Community (AHIC) has given the following broad charge to the Personalized Health Care Workgroup:

**Broad Charge for the Workgroup:** Make recommendations to the AHIC for a process to foster a broad, community-based approach to establish a common pathway based on common data standards to facilitate the incorporation of interoperable, clinically useful genetic/genomic information and analytical tools into electronic health records to support clinical decision-making for the clinician and consumer.

The Workgroup's deliberations have highlighted a number of key issues regarding the broad charge, including the following:

1. Genetic/Genomic Tests
2. Family Health History
3. Clinical Decision Support
4. Confidentiality, Privacy, and Security

This letter provides both context and recommendations for how the issues of genetic/genomic tests and family health history can be addressed in the next twelve months.

## BACKGROUND

The Workgroup's vision of Personalized Health Care (PHC) is a consumer-centric system in which clinicians and consumers work together to customize diagnostic, treatment, and management plans based on a variety of factors, including the consumer's culture, personal behaviors, preferences, family health history, and the individual's unique genetic/genomic makeup. In this desirable future, consumers and clinicians both have ready access to information needed to identify and assess individualized treatment

options as well as the resources and reimbursement mechanisms necessary to support implementation of a more extensive menu of tests and treatments.

Underpinning this vision is the confluence of two powerful forces, the development of Health Information Technology (HIT) and the rapid advances in the basic understanding of the relationships between health, disease, genetics/genomics, and prevention and treatment options. Knowledge of an individual's genetic/genomic makeup appears to have an exceptionally powerful ability to assist with disease prediction, diagnostic accuracy, targeted treatments, medication dosing, and health management.

The PHC Workgroup has held six meetings since its formation in October 2006. Testimony from a wide variety of experts in standards development, genetics/genomics, laboratory testing procedures and systems, privacy concerns, tools and standards for family health history, and commercial and government electronic health record (EHR) systems has informed the Workgroup's discussions. In March 2007, the Workgroup developed a vision of PHC from four perspectives: the consumer; the clinician; the researcher; and the health plan/payer. Following this visioning session, the Workgroup outlined its priorities in the areas of: genetic/genomic tests; family health history; clinical decision support; and confidentiality, privacy, and security. The vision summary and priorities documents were presented to the AHIC on April 24, 2007. Subgroups of the Workgroup were formed to address each of these four priority areas. Two of these subgroups, genetic/genomic tests and family health history, have developed recommendations that are being advanced to the AHIC by the PHC Workgroup.

If accepted by the AHIC, these recommendations should be considered for adoption by the Department of Health and Human Services (HHS) as HHS policy regarding current and future federal activities as they relate to the Workgroup's charge.

## INITIAL RECOMMENDATIONS

### 1. Overarching

With the completed sequence of the human genome, genetic/genomic testing and its possibilities have moved from the sidelines into mainstream medicine. There are over 1,400 diseases for which genetic/genomic tests are used in current clinical practice, and several hundreds more are available in a research setting.[FN1] A genetic/genomic test can be performed on a wide variety of tissue samples and across the human lifespan, providing information on predispositions for a disease, presence of a disease, the risk of passing a disease onto offspring, and potential positive or adverse responses to therapeutic interventions.

In addition to the increasing adoption of genetic/genomic testing in medical practice, clinicians have always used a basic and important genetic/genomic tool in everyday practice: family health history. Combined with the power of genetic/genomic testing results, family health history adds value and provides useful predictive information.

Broadly stated, genetic/genomic information has the potential to identify and predict the health outcomes of individuals and their families.

Consumers today are concerned that their health information may be used for unintended purposes or without their authorization. Compounding this concern are the limited understanding of new genetic/genomic tests for heritable disorders, the immutability of this information across the consumer's entire lifetime, the predictive abilities attributed to genetic/genomic information, and the potential for unintended informing of relatives because of a common genetic/genomic background. However, if consumers avoid genetic/genomic tests because of fear, they are potentially at risk by not having information available to them that could substantially and beneficially alter their health care. Therefore, maintaining the public's trust in the use of their personal health and genetic/genomic information, by developing technical and policy guidelines to ensure the security of their genetic/genomic data, is key to maximizing utility and health benefits. Consumer authorization of access to their genetic/genomic information should be taken into consideration as these use cases are developed. Therefore, the PHC Workgroup will work with the Confidentiality, Privacy, and Security (CPS) Workgroup to consider if aspects of genetic/genomic test results and family health history information may raise special concerns about confidentiality, privacy, and security relative to other types of medical data.

The Workgroup identified the following actionable recommendations for the next twelve months that begin to address one aspect of the broad charge, incorporating clinically useful genetic/genomic information into the EHR.

Recommendation 1.0: The Community should advance the area of Personalized Health Care as a Priority for Use Case Development.

Recommendation 1.1: Priorities for use cases in the area of Personalized Health Care should be developed in conjunction with work performed by the genetic/genomic test workgroup and the family health history workgroup described in Recommendations 2 and 3. The use cases should additionally leverage the work in related activities including: the AHIC EHR, CPS, and Consumer Empowerment (CE) Workgroups; the Harmonized Use Case for Electronic Health Records (Laboratory Results Reporting); the Consumer Access to Clinical Information Use Case; and others.

## 2. Genetic/Genomic Tests

Inclusion of genetic/genomic test results in the EHR or personal health record (PHR) could enable the personalization of health care decisions through avoidance of adverse reactions, selection of optimal interventions, and beginning the transition of the health care sector from a reactive to a predictive enterprise. Standardized electronic recording of data associated with laboratory performance of genetic/genomic tests and, in parallel, inclusion of relevant results from genetic/genomic tests in the EHR have been identified as immediate priorities for recommendation by the PHC Workgroup.

Genetic/genomic testing in humans generally falls into two categories: molecular and biochemical. A molecular genetic/genomic or cytogenetic test may be defined as an analysis performed on human DNA, RNA, and chromosomes to detect heritable or acquired disease-related genotypes, mutations, or karyotypes for clinical purposes. A biochemical genetic/genomic test may be defined as the analysis of human proteins and certain metabolites, which are predominantly used to detect inborn errors of metabolism, heritable genotypes, or mutations for clinical purposes. Tests that are used primarily for other purposes, but may contribute to diagnosing a genetic/genomic disease (e.g., blood smear, certain serum chemistries), would not be covered by this definition.[FN2]

The process of performing a genetic/genomic test can be segmented into three distinct phases with each having different information collection requirements. The three phases include: (1) the pre-analytic phase, which encompasses such events as determining which genetic/genomic test, if any, is appropriate to answer the clinical question being asked, collecting clinical information that is necessary to interpret the test, and collecting an appropriate sample and transporting it to the test site; (2) the analytic phase, which involves steps taken to perform the analysis and analyze the results; and (3) the post-analytic phase, which includes reporting and interpretation of the results.[FN2]

As the area of genetic/genomic tests is relatively new to the medical community, and there are a growing number of different types of tests that are captured by the broad definition of a genetic/genomic test, standards development in some areas of this diverse category may be immature. Therefore, an iterative process should be pursued where use case development is performed in parallel with standards identification/creation. Gaps in available standard reference materials, protocols, metrics, IT standards (terminology, coding, messaging, instrument integration, and implementation guides) will therefore be highlighted early in the process and brought to the attention of the appropriate standards development organizations. Standards that address communication between EHRs and Laboratory Information Systems (LIS) are crucial to ensure comprehensive bidirectional transfer of information between the EHR and LIS in the pre- and post-analytic phases.

The many different information requirements for incorporation of genetic/genomic test information in the EHR is an issue of immediate concern to the PHC Workgroup. Longer term goals of this Workgroup include supporting the development of accompanying information about benefits, risks, analytical validity, clinical validity, and clinical utility to ensure the development of robust clinical decision support concerning genetic/genomic test results. Additionally, incentives to develop new genetic/genomic tests that provide new or added value to clinical care and the corresponding reimbursement strategies to ensure their widespread use need to be addressed. These longer term goals would be facilitated by the development of means and standard materials and processes for capturing laboratory data and test results identified as the immediate concerns for Healthcare Information Technology Standards Panel (HITSP) use case development. Future recommendations by the PHC Workgroup may address these longer term issues.

**Recommendation 2.0:** An extension to the Harmonized Use Case for EHRs (Laboratory Results Reporting) should be developed to address the specific information needs in the

pre-analytic, analytic, and post-analytic phases of genetic/genomic tests. This extension to the use case should additionally address the need for integrated data flow across the pre-analytic, analytic, and post-analytic phases of genetic/genomic testing and address both the EHR and Laboratory Information Systems.

Recommendation 2.1: A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to identify what types of data and information are generated when performing genetic/genomic tests, and to identify standard metrics, terminology, language, and processes. This work should inform the extension to the Harmonized Use Case for EHRs (Laboratory Results Reporting) developed for genetic/genomic tests.

Recommendation 2.2: Research activities that increase the knowledge base regarding genetic/genomic test results need to be supported. The National Institutes of Health (NIH) should continue to work with public and private partners to support, develop, and enhance public reference databases that enable more effective and efficient genetic/genomic testing and incorporation of test results that can be aggregated in electronic health records.[FN3]

### 3. Family Health History

Health care professionals and the general public have widely accepted the importance of family health history for predicting increased risk for a number of common diseases, including cancer, heart disease, and diabetes. As our scientific understanding of the molecular and genetic/genomic basis for health and disease improves, the importance of family health history as a valuable predictive tool has only increased. This has been highlighted throughout HHS by the Surgeon General's online web portal for collecting family health history information, the 'My Family Health Portrait', developed in conjunction with the NIH and the Centers for Disease Control and Prevention. The Family Health History priority area for the PHC Workgroup includes activities of immediate concern related to use case development by HITSP. The use case should represent the continuum of information collection, from consumer entry of family health history in the PHR to clinician entry of family health history in the EHR, with the longer term goal of interoperability between the PHR and EHR. Health care providers involved in any pilots of this use case should examine the merits of developing a modular family history tool, where collection of family health history is performed within the EHR, followed by messaging of this information to a variety of richer family history tools that perform risk analyses. In these tools, family history data can continue to be extended with new family history information as well as analyzed using the latest risk assessment algorithms. The enhanced family history and results of these algorithmic calculations could then be returned to the EHR, allowing for the ongoing curation of novel risk assessment algorithms and use of these tools in concert with well established family health history collection tools.

Additionally, the longer term goals of the Family Health History priority include: infrastructure and incentives to use PHRs to improve consumer-clinician communication;

and characterization of the validity and utility of use of family health history in making clinical decisions. An overarching theme across the Family Health History priority area is how the clinician can use the family health history information, and this should be considered in short and long term activities. These longer term goals are contingent on the development of means and standards to capture the family health history information identified as the immediate concerns for HITSP use case development. Future recommendations by the PHC Workgroup may address these longer term issues.

Recommendation 3.0: A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to develop a core minimum data set and common data definition available for primary care collection of family health history information.

Recommendation 3.1: Additionally, studies should be performed as part of this collaboration as an evidence-base to determine the validity and utility of family health history risk assessment and management tools, clinical decision support tools, and how clinicians view this information as helpful for informing their medical decisions.

Recommendation 3.2: Federal agencies in conjunction with private health care organizations with similar interests and expertise sponsoring pilots in the area of family health history should be used to evaluate the core minimum data set and evidence-base developed through Recommendations 3.0 and 3.1. Health care providers involved in these pilots should also examine the feasibility of consumer-clinician exchange of family health history information between PHR and EHR systems. When possible, the pilots should test and implement the standards and architecture identified in the HITSP developed use case.

These recommendations are supported by information obtained through research and testimony to the Personalized Health Care Workgroup, which is contained in the supporting documents available at <http://www.hhs.gov/healthit/ahic>.

Thank you for giving us the opportunity to submit these recommendations. We look forward to discussing these recommendations with you and the members of the American Health Information Community.

Sincerely yours,

John Glaser  
Co-chair  
PHC Workgroup

Douglas E. Henley  
Co-chair  
PHC Workgroup

---

1 [www.genetests.org](http://www.genetests.org)

2 CDC definition, Federal Register, Vol 65, No 87, 5/4/2000, 25928.

3 Specifically, NIH, and the National Library of Medicine (NLM) in particular, should continue to: (1) enhance its collection of mutation data; (2) expand a National Center for Biotechnology Information (NCBI) clinical reference sequence database (RefSeqGene); (3) expand coverage of genetic/genomic tests in Logical Observations Identifiers Names Codes (LOINC) in collaboration with other HHS agencies, state public health laboratories, and the American Society of Human Genetics; and (4) provide more integrated access to genetic/genomic information for the public through NCBI portal developments, the Genetics Home Reference, Online Mendelian Inheritance in Man (OMIM), and MedlinePlus in cooperation with other HHS agencies, the Genetic Alliance, the American College of Medical Genetics, and other professional and disease advocacy groups.