

February 26, 2008

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 American Health Information Community 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 issue of Newborn Screening (NBS) can be addressed in the next twelve months.

BACKGROUND

As one of the more common applications of genetic tests in public health, newborn screening for congenital (inherited) disorders is performed near the time of birth for nearly every newborn, accounting for more than four million infants each year. Finding these conditions in the newborn, before symptoms appear, has been shown not only to save lives, but also, for certain disorders, to save costs for the health care system. Various

conditions can be screened for by collecting a dried blood spot sample on filter paper or conducting a physiologic test, as with newborn hearing screening.

Newborn screening illustrates the critical need for standardized key data elements and terminologies in order to advance our understanding of genetic and other congenital conditions on a population basis. Issues include standardization of case definitions; recognizing the relationship between screening and diagnosis; and tracking the newborn to ensure proper follow-up of out of range (abnormal) test results. Standard representations of newborn screening (and confirmatory) data must be developed to enable sharing of data with individuals involved in the infants' care, for research purposes, and for improved patient and population health.

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. Newborn Screening Information Exchange

The capability to exchange newborn screening test results via electronic means (interoperability) is not widespread among public health and health care providers, despite the availability and use of electronic data exchange for other types of test results. Information flows are complicated by the fact that the provider who needs to receive results and import them into an electronic health record (EHR) is not always the provider who orders them. Provision should be made to allow parents to authorize delivery of test results to any primary care provider of their choice. Additionally, results should be available from a secure web site for authorized providers who do not use an EHR and for access by hospital Emergency Department personnel who may have to evaluate an infant in crisis. Appropriate direction and exchange of screening data is crucial to ensure proper follow-up and management of affected infants. Clinical vocabularies for newborn screening tests are needed to code the tests that were performed, the categorical results produced, and the conditions that were tested. The use of existing standards and approaches for the exchange of electronic information with Certification Commission for Healthcare Information Technology (CCHIT) certified EHRs could also facilitate the inclusion of newborn screening data, but significant challenges remain at other interfaces between the public and private parties involved.

The legal issues, data flows, actors, and stakeholders for newborn screening are complex and the actions take place over a longer period of time than other types of screenings and genetic testing. The initial screening test is typically mandated by state law and often does not require patient consent. Full diagnostic test evaluation, follow-up, and treatment may take place over prolonged periods of time and involve the participation of specialists and laboratories or audiologic (hearing test) centers, as with hearing screening, not involved in the initial testing.

Many of the conditions detected by newborn dried blood spot screening are rare and seen by a primary care provider only once during his/her career. Approximately 8,000 of the more than four million infants screened each year are identified with certain medical conditions through these screening programs. Some children become acutely and seriously ill as a consequence of their disorder and are taken to Emergency Departments even before the results of newborn screening are available. Information and guidance must accompany the test results to save time by educating providers and families about the rare conditions. The ACT sheets developed by the American College of Medical Genetics¹ are an important example of this type of resource, but information regarding local resources and recommended procedures vary depending on the state screening programs. Additionally, updates to these advisory documents occur over time and it is appropriate to check for updates on an identified condition as the child grows older. Concurrent to reporting the results to the primary care provider, some states report abnormal results directly to parents or through designated specialist centers to improve timeliness of follow-up testing or start of appropriate therapy.

Newborn hearing screening, or more commonly referred to as Early Hearing Detection and Intervention (EHDI) at the state and federal level, has proven to be an outstanding public health success story. With two to three of every 1,000 babies being born with a hearing loss, it is the most frequently occurring condition screened in the newborn period. It is estimated that more than 50% of congenital hearing loss has a genetic basis. In 1993 only eleven hospitals in the United States were screening a significant number of their newborns for hearing loss. Now, the hearing screening results of over 92% of the infants born in the U.S. can be documented. Unfortunately, a documented diagnosis cannot be confirmed for nearly two-thirds of the infants not passing their final hearing screen. Moreover, for those infants with a confirmed diagnosis of permanent hearing loss, over 20% cannot be confirmed as having obtained early intervention services. Left undetected, hearing loss in infants can negatively impact communication, social and emotional development, as well as academic achievement. If a child with hearing loss is identified early and given appropriate educational, medical, and audiological services, significant special education and societal costs savings can be realized.

Newborn dried blood spot tests and hearing screening testing often differ from other laboratory tests in the practice of reporting results as positive or negative (or in range/out of range) without specific analytical values. Current practice recognizes that these tests are a screen and generally not considered diagnostic in nature. A qualitative value is generally used at the time of assigning a diagnosis. There is also often a need for immediate and long-term follow-up testing, and reporting the quantitative measurements that now underpin the qualitative results and/or interpretative reports, and other test- or method-specific information, may assist in qualitative result interpretation.² As currently deployed, manufacturers of screening devices most often utilize statistical and mathematical techniques to distinguish between individuals who are likely versus those who are not likely to be identified with a target condition, rather than the reporting of quantitative data. However, the capacity to collect, transmit, and analyze quantitative data could potentially improve quality assurance measures, reduce costs, and support clinical decision making for the public health community, clinicians, and consumers.³

A detailed Use Case will clarify the workflows involved, guide the identification and selection of required standards, and determine the electronic reporting and tracking requirements of the entire newborn screening process. The information exchange for newborn screening serves at least two purposes: to ensure timely and accurate delivery of information for clinical decision-making and to facilitate quality assurance within the screening system. The existing Office of the National Coordinator for Health Information Technology (ONC) Harmonized Use Case for EHRs (Laboratory Result Reporting), the Consumer Empowerment: Consumer Access to Clinical Information Use Case, and the Personalized Health Care Use Case provide a foundation that will extend the scope of the existing use cases and build on prior work by including the additional data requirements and the need to share results between multiple providers and public health entities. This should also be considered in the context of the Population Health and Clinical Care Connections Workgroup recommendations regarding laboratory result reporting and a national program to enable public health laboratories to exchange data with other public health laboratories, which were accepted by the AHIC in January 2008.⁴ Additionally, work in progress to develop a Health Level Seven (HL7) Implementation Guide for Newborn Screening will also facilitate and guide development of the Use Case.

Recommendation 1.0: The information flows for Newborn Screening should be prioritized for Use Case Development. All of the multidirectional information flows, stakeholders, and other participants involved in the complete evaluation of newborn screening (i.e., hearing detection, dried blood spot screening, and diagnostic confirmation) should be considered so that appropriate standards and interoperability specifications can be developed to support information exchange.

Recommendation 1.0.1: The Newborn Screening Subgroup of the Personalized Health Care Workgroup should complete development of a reference matrix of tests, analytes, conditions screened for, and associated genomic variants that are used in newborn screening programs.

Recommendation 1.0.2: Based on the reference matrix described in Recommendation 1.0.1, appropriate codes should be identified for use in electronic reports to identify the test ordered, individual test results, and categorical results of these tests (e.g., Logical Observation Identifiers Names and Codes (LOINC), Systematized Nomenclature of Medicine Clinical Terms (SNOMED-CT), HL7, Online Mendelian Inheritance in Man (OMIM), International Classification of Diseases Ninth Edition (ICD-9), and ICD-10 Clinical Modification (CM)).

Recommendation 1.0.3: Long-term maintenance of the reference matrix should be coordinated by the National Library of Medicine (NLM) in collaboration with the Health Resources and Services Administration (HRSA) and the Centers for Disease Control and Prevention (CDC).

Recommendation 1.0.4: For the Use Case development process, ONC should consider the need for documentation of permissions and authentications of users for access and transmittal of results, the need for ongoing collection of information for long-term

follow-up, and integration of existing educational and clinical decision support information.

Recommendation 1.1: Requirements for electronic reporting of newborn screening results should include specifications for reporting the quantitative measurements that now underpin the qualitative results and/or interpretations. Allowance should be made for accompanying qualitative and/or interpretive reports, and other test- or method-specific information that may assist in qualitative result interpretation.

Recommendation 1.1.1: HHS should work with the National Governors Association (NGA) and the National Conference of State Legislatures (NCSL) to support electronic reporting of quantitative, qualitative, and/or interpretive reports.

Recommendation 1.1.2: HHS should convene a workgroup with participation from the Centers for Medicare and Medicaid Services (CMS), HRSA, Substance Abuse and Mental Health Services Administration (SAMHSA), Administration for Children and Families (ACF) and other agencies that provide grants or reimbursement to health care providers, in order to determine the most appropriate ways to facilitate the adoption and development of electronic systems that conform to the concepts and standards identified in the Use Case. Special attention should be given to funding opportunities provided by existing authorities associated with the Early, Periodic, Screening, Diagnostic and Testing (EPSDT) requirements under Title XIX for Medicaid beneficiaries; e.g., enhanced match for the Medicaid Management Information System (MMIS) and in a manner consistent with the emerging architectures described within the Medicaid Information Technology Architecture (MITA).

Recommendation 1.2: An action plan, timetable, and metrics for the implementation and tracking of these recommendations should be developed by HRSA to measure uptake of electronic transmission of test results that conform to the standards identified through the Use Case development process. HRSA Newborn Screening technical support centers should conduct annual surveys to monitor the pace of implementing these recommendations, standards, and transmission of newborn tests results by electronic means (EHRs and repositories).

2. Confidentiality, Privacy, and Security Issues Specific to Newborn Screening

Secure communication is critical to the patient-family/physician relationship, contributing to the quality of care and improved health outcomes. Sharing of newborn screening results is common and necessary for effective and timely use of newborn screening results and directing appropriate responses to those results. Several aspects of the newborn screening process present unique challenges with respect to appropriate sharing and transmission of results. First, it is common that a newborn's name changes between when the test is performed and when the results need to be reported. Second, the clinician ordering the newborn screening tests is usually not the same clinician who will be acting as the infant's primary care provider. Third, situations commonly arise when infants are born in one state while their family's primary residence and the location of the

primary care provider may be in a different state. Private and secure solutions need to be developed that facilitate electronic reporting of or web access to screening results by the parents, and/or parent-authorized health care providers regardless of whether they are the original ordering provider or if they practice in a state other than the one where the infant was born and the newborn screening test was conducted. While there may not be overt aspects of the various privacy regulations that appear to be an impediment, the lack of understanding amongst health care providers around their application to newborn screening results impedes timely exchange. A January 2008 analysis prepared by the HRSA-funded National Newborn Screening and Genetics Resource Center for the NBS subgroup of the Personalized Health Care Workgroup suggests that newborn screening program officials in many states are uncertain about the privacy requirements for electronic reporting of newborn screening results. While a majority of the respondents confirmed that privacy concerns are an important consideration for electronic NBS test result reporting, almost half of the respondents were uncertain about which privacy laws or regulations are relevant to electronic reporting, and were equally uncertain if their specific states were developing new laws that would affect electronic sharing of NBS results. When considering electronic reporting of NBS results, both the need for timely communication and sharing of screening results among appropriate clinicians, and protections against inappropriate disclosure of screening results, should be considered.

Recommendation 2.0: HHS should work with state stakeholders to accurately identify, analyze, and develop solutions to address any misperceptions or misapplications of state privacy laws that may affect the timely transmission of newborn screening results. This work should also include an analysis of whether clarifying guidance from HHS related to the Health Insurance Portability and Accountability Act (HIPAA) Privacy and Security Rules, the Clinical Laboratory Improvement Amendments (CLIA), and other regulations under HHS' authority would be appropriate.

3. Reporting of Newborn Screening Results to Improve Population Health

There are several uses of patient-based informational registries and other health record systems, including research, program evaluation, and monitoring of health outcomes. Program evaluation improves the efficiency and effectiveness of newborn screening programs. Positive predictive values of similar tests may vary from state to state based on cut-off values or variations in methods that are used. The high social cost and unnecessary diagnostic testing cost of false positive results, as well as the potentially tragic costs of a false negative result, justify appropriate use of newborn screening data to improve screening programs. In parallel with enhanced infrastructure and technical capabilities for information sharing, ongoing efforts to address emerging ethical, legal, and social implications of these capabilities on newborn screening programs will need to be addressed.

Use of newborn screening data to advance population health should be facilitated to improve the efficiency and effectiveness of these tests. One example of such a project is a HRSA-funded program at the Region 4 Genetics Collaborative ⁵ that has been established to improve access to high-quality genetic and newborn screening services to children and

families. Findings from this project's efforts include the creation of a multi-state database that could be used as a model for a national program to improve the early identification and management of infants with metabolic, genetic, and other physiological disorders. Another example is a Region 3 Genetics Collaborative⁶. project that is developing a long-term follow-up information system as a resource for care coordination, research, and information among clinicians, researchers, and consumers within that region. Both the Region 4 and 3 projects necessitate the transfer of information between the public and private sectors and multiple public health and health care providers in a multidirectional fashion. The development of policies and procedures that address confidentiality and privacy issues to guide the appropriate use of patient-based informational registries and other health record systems should consider guidelines for secondary data use previously developed by the National Committee on Vital and Health Statistics (NCVHS)⁷ and the American Medical Informatics Association (AMIA)⁸.

Recommendation 3.0: A taskforce that includes representatives from appropriate federal and state agencies, professional and public organizations, and the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC) should be formed to develop a plan for and descriptions of a patient-based information registry of newborn screening data within twelve months. Public review of the findings of this taskforce will be essential to address any ethical, legal, and social implications of any proposed research that will be facilitated by the development of electronic test reporting and national standards for identifying the tests performed and results obtained.

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/>.

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, PhD
Co-Chair, Personalized Health Care
Workgroup

Sincerely yours,
Douglas E. Henley, MD
Co-Chair, Personalized Health Care
Workgroup

1 <http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>. ACTION (ACT) sheets describe the short term actions a health professional should follow in communicating with the family and determining the appropriate steps in the follow-up of the infant that has screened positive.

2 Quantitative newborn dried blood spot screening reports may include numeric values that represent analytic values, percentiles, and/or ratios and should be accompanied by expected ranges. Qualitative reports may include testing observations (e.g., fluorescence or no fluorescence) and subjective evaluations (e.g., Hb FA present). Interpretive reports

may include probability information (e.g., probable Hb S,S anemia), or other reporting information (e.g., T4 out-of-range, TSH out-of-range, please refer for serum testing).

3 Quantitative newborn hearing screening reports may include data collection parameters such as type of stimulus delivery transducer (e.g., circumaural, supraaural and tubal-insert earphone), stimulus parameters (e.g., type [transient/tonal envelope], intensity [reference equivalent threshold sound pressure level], number, rate, duration, polarity), and elicited waveform response measurements (acoustic as in otoacoustic and distortion product measurement [frequency analysis, response level, baseline noise and contamination level, reproducibility, correlated non-linearity] or electrical as in auditory brainstem response [analysis of intensity, frequency, absolute and inter-peak latencies, morphology]), and test conditions (e.g., test time, signal-to-noise ratio, calibration date).

4 http://hhs.gov/healthit/documents/m20080115/06-phccc_recs_ltr.html

5 <http://region4genetics.org/>

6 <http://region3collaborative.org/>

7 <http://www.ncvhs.hhs.gov/071221lt.pdf>

8 “Toward a National Framework for the Secondary Use of Health Data: An American Medical Informatics Association White Paper.” Journal of the American Medical Informatics Association, Volume 14, Number 1, 2007.