

# Newborn Screening as the Model for Interoperable Genetic Data

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## Abstract

Each year over 4 million infants undergo mandated Newborn Screening to identify a wide range of genetic disorders. The Newborn Screening Use Case is introducing standards for interoperability to this important and challenging juncture of inpatient, public health and ambulatory practice settings. Special coding and terminology issues for these rare disorders have led to development of new LOINC codes and integration of work of the Personalized Healthcare Workgroup into the NLM UMLS. The birth of a new infant is an ideal time for families to provide interoperable family health histories collected using the Internet based Surgeon General's Family Health Portrait Tool that can be incorporated into a lifetime child health record beginning at the time of newborn hospital discharge. Privacy issues surrounding the transmission of genetic data must be addressed and the policy implications for newborn screening of the Genetic Information Nondiscrimination Act of 2008 (GINA) as well as HIPAA, CLIA, and state laws will be discussed. Effective newborn screening requires sharing of results with non-ordering providers and sharing results across state lines. Research uses of newborn screening are fundamental to population health improvement through program evaluation and gathering evidence about effective approaches to newborn screening and assessment of outcomes.

## Description

Personalized Healthcare and Systems Based Medicine are transforming the next generation of healthcare through integration of genomics and genetic differences into clinical practice. Informatics must prepare and participate in the transformation of medical sciences and interoperable newborn screening data and the American Health Information Community (AHIC) Newborn Screening Use Case will be an important challenge for the informatics community to address the adequacy of current methods and standards to handle new types of data and interoperability amongst a wide range of stakeholders. By the time of the AMIA 2009 Fall Symposium, the Health Information Technology Standards Panel (HITSP) will complete work on the

newborn screening interoperability specification and state health departments and EHR vendors will be preparing to implement their work. The practice of newborn screening creates a mandatory need for interoperability that is not being met today and opportunities to introduce new standards for electronic data transfer and integration of family history with genetic testing that can form the basis for a lifetime electronic record for every new person born in the United States. The panel will address a range of informatics issues and challenges that can use the Newborn Screening Use Case as an example of solving problems that will be encountered in other areas of integrating genetic data into medical practice. The panel will include discussion of five central issues in newborn screening interoperability. This panel will address important cross cutting issues between the application domains of providers, public health, and government health policy. It requires attention to controlled vocabularies and terminologies as well as data exchange, communication, and integration across care settings.

## Presentations

**Health Information Exchange of Newborn Screening Data** – This presentation will give an overview of the interoperability requirements and standards proposed for newborn screening.

**Coding and Terminology Requirements for Newborn Screening** – This presentation will discuss the need for attention to terminology and the challenge of reporting data in an area where the focus is on qualitative results based on conditions screened with additional need for quantitative results of the actual methods and tests performed.

**Integration of Family Health History with Newborn Screening Data** – This presentation will present new tools for combining family health history with genetic testing and the need to create lifelong records that will follow the infant through childhood and into adulthood.

**Privacy Issues in Newborn Screening Health Information Exchange** – This presentation will

review regulations and ethical considerations that require attention as newborn screening moves from manual methods to electronic health information exchange and the special considerations of working in the public health domain.

**Public Health Issues and Long Term Follow-up of Newborn Screening** – This presentation will give an introduction to the population health perspective including research and public health considerations in a rapidly changing field.

**Participants**

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All participants have agreed to take part in the panel