



Department of Health & Human Services
Office of the National Coordinator for
Health Information Technology

American Health Information Community

**Personalized Health Care Workgroup
Newborn Screening Draft Reference Matrix**

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PHC Newborn Screening (NBS) Subgroup Member List

- **Co-Chairs:**

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- **Members:**

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- **Members:**

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PHC NBS Reference Matrix Subgroup Member List

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Background

- Relevant newborn screening recommendations accepted by the AHIC on February 26, 2008:
 - 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)).

Background (cont.)

- Relevant newborn screening recommendations accepted by the AHIC on February 26, 2008 (cont.):
 - 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).
- June 3, 2008 AHIC Meeting:
 - Full NBS use case identified as a priority for HHS and advanced for the next round of formal use case development (2009)

NBS Reference Matrix Subgroup Activities to Date

- Four meetings held April through May 2008
 - Convened core task group of stakeholders and domain experts
 - Reviewed goals for implementing reference matrix recommendation
 - Discussed proposed database approach and format
 - Developed plan for completing matrix
 - Identified external reviewers for feedback on draft matrices
- Ongoing:
 - External review and vetting of matrices

Overview of the NBS Draft Reference Matrix

- A set of tables that will be used to:
 - Support development of interoperability specifications for the NBS use case
 - Guide specifications for a uniform electronic newborn screening report that can be used for patient focused care as well as for population health
 - Serve as the source for all terminology and coding for both the initial screening report and the consultation and referral summary document
 - Initial focus has been on the initial screening report based on newborn dried blood spot screening tests and newborn hearing screening
 - Expedite implementation of the use case by facilitating early involvement of standards development organizations to address gaps in coding and standards

Overview of the NBS Draft Reference Matrix (cont.)

- Important feature of the reference matrix is an attempt to merge together the wide range of tests and conditions covered in newborn screening programs
 - Included Early Hearing Detection and Intervention (EHDI), along with other forms of newborn screening for metabolic, endocrine, hemoglobin, congenital infections, and other conditions
 - Although level of alternate terminology and coding varies among categories of newborn screening, the reference matrix demonstrates strong similarities in the data requirements of all types of newborn screening and the ability to represent this data in a single matrix
- The matrix remains a work in progress that will continue to expand and change as the methods of newborn screening evolve

Roadmap to the NBS Draft Reference Matrix

- Matrix was developed separately for each screening domain and then reassembled into a single final document with the following core worksheet tabs:
 1. Tandem Mass Spectrometry (MS/MS) Analytes
 2. MS/MS Conditions
 3. MS/MS Analyte to Condition
 4. Non-MS/MS Analytes
 5. Non-MS/MS Conditions
- Each section followed comparable goals for content, which vary considerably in detail and granularity by domain

Next Steps

- NBS reference matrix completed by June 16, 2008
- Long-term maintenance to be coordinated by NLM
- Alignment with formal NBS use case development

- Questions?