



Update on Newborn Screening Use Case Advisory Committee on Heritable Diseases in Newborns and Children

**- Advisory Committee on Heritable Diseases in
Newborns and Children -**

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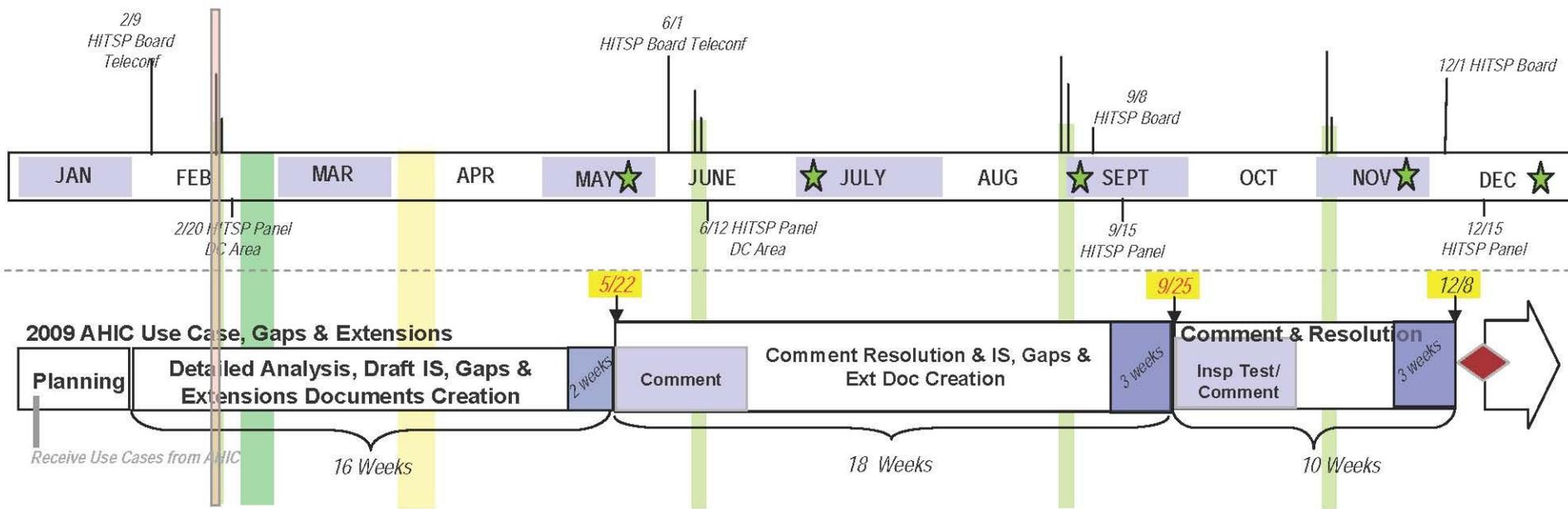
Status of the Newborn Screening Use Case

- The Use Case and companion Coding and Terminology Guide were released to the public on December 31, 2008
<http://www.hhs.gov/healthit/usecases/documents/NBSDetailedUseCase.pdf>
<http://www.hhs.gov/healthit/usecases/documents/NBSTerminology.pdf>
- Standards harmonization is underway at the Health Information Standards Panel HITSP Population Perspective Technical Committee
 - Participation in the weekly TC is open and is encouraged
 - Review and public comment on the HITSP documents will be extremely important to assure that the Interoperability Specification meets the needs of NBS labs and programs
- We presented an APHL teleconference on Interoperable Electronic Newborn Screening Lab Reports to encourage NBS labs to plan for implementation of the Use Case
- A document is in preparation to advise the use case on privacy considerations under HIPAA, CLIA, and state laws regarding reports to non-ordering providers and sharing data between states
- Today's presentation will focus on progress with terminology and migration of the work of the PHCWG into NLM UMLS

Components of the HITSP Interoperability Specification - Reuse Existing Standards

- Initial Screening
 - Electronic ordering with birth history and newborn data
 - Lab result messages to EHR (HL7 v2.3.1)
 - Lab report documents (HL7 CDA) for web viewing
 - Capture of hearing screening results
- Consult and Referral Document (CCD)
 - Summarize initial screening
 - Summarize confirmatory testing
 - Confirmed diagnosis
 - Serve as an emergency information form
- Public Health Reporting and Registries
 - Identify the medical home
- Quality Measurement and Follow-up
 - Based on Quality Use case and templates
- Context Specific Information Distribution (such as ACT sheets)
- Timeline – accept Dec 2009, recognize Dec 2010, certify July 2011

HITSP 2009 TC Timeline Overview



WE ARE HERE

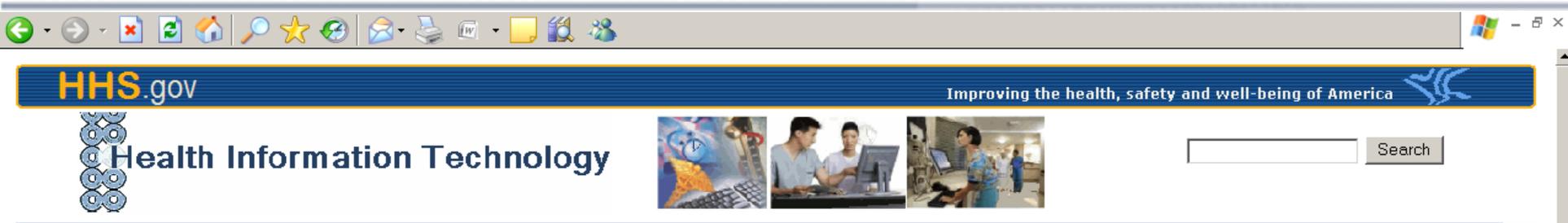
- ★ Holiday
- Full TC Meetings
- IRT Content, Editorial and Quality Review
- Connectathon
- HIMSS Conference

◆ Panel Approval

Update on Coding and Terminology

- A revised version of Coding and Terminology Guide was published with the use case on Dec 31, 2008
- On-line version remains available for comment at
 - <http://transparency.cit.nih.gov/screening>
- LOINC code revisions were included in the Jan 2009 v2.6
- Clarification of the “ACMG Codes” has led to plans for SNOMED revisions and UMLS integration
- Dataset development activities for result reporting are underway at HITSP
- Dataset development activities for Long Term Follow-up are underway at ACHDNC and ACMG
- Opportunities for public comment on datasets and codes will continue during the next six months

NBS Coding and Terminology Guide - online



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Analyte Detail	Condition Detail
MS/MS Analyte to Condition Mapping	MS/MS Condition to Analyte Mapping
Email Your Comments	

This is a web-based tool to allow the review of proposed standards for newborn screening condition and analyte terminology, codes, and mapping for the purpose of supporting the generation and exchange of electronic laboratory reports for newborn screening. Based on a recommendation approved by the American Health Information Community (AHIC) these data are provided for clinical conditions identified and the quantitative analytes measured.

In addition, data include a variety of coding systems that may be required for rare disorders genetic disorders and provide LOINC codes to assist in identifying results included in a newborn screening report and documenting the methods used in the laboratory.

Early Hearing Detection and Intervention (EHDI) testing is included in addition to newborn dried blood spot testing. The aim is to accurately document and encode the data in order to identify the conditions screened for or identified as well as the test results that are associated with those conditions. Maintenance of the newborn screening guide and codes will be an on-going activity as the field of newborn screening changes.

The queries listed illustrate the types of coding and terminology that will be available for use in electronic reporting of newborn screening results. Use of a standard framework for coding and terminology will assist in the comparison of data from different laboratories and help identify gaps in coding that to support the exchange of electronic newborn screening reports.

Conditions:

The Conditions query lists the conditions that can be identified by newborn screening tests along with appropriate diagnostic codes. The report includes conditions that are tested for by tandem mass spectrometry (MS/MS) on newborn dried blood spots, and conditions indentified by non-tandem mass spectrometry tests performed on newborn dried blood spots, as well as hearing loss detected through early hearing detection and intervention (EHDI) programs.

Analytes /Results:

The Analytes query lists the analytes or chemical entities that are measured by newborn screening tests along with appropriate LOINC codes that are used to identify specific laboratory result fields on electronic laboratory reports. The report includes conditions that are tested for by tandem mass spectrometry

Need for NBS Coding and Terminology

- Coding and terminology drive modern standards by defining data fields and value sets - not just about labeling diagnoses
- Newborn screening presents many challenges to maintain consistent terminology
 - Names based on clinical syndromes, enzyme deficiencies, lab abnormalities, or specific genetic variants
 - The latest version of LOINC includes the suggestions of the PHCWG newborn screening sub-group
- Essential for electronic result ordering and reporting
- Essential for quality assurance and long term follow-up and outcomes assessment
- Enable population health activities including research and program evaluation

Clarification of “ACMG codes”

- During the selection of the 29 test core panel, ACMG used a set of acronyms of convenience to label the over 80 conditions that were considered as screening targets
- Reports from the NNSGRC and NNSIS on current state activities are organized by those terms
- The “ACMG codes” are not a formal vocabulary and are not codes like ICD9 even though they are often used that way
- The granularity of the ACMG acronyms is not perfectly aligned with SNOMED or ICD10
- NLM has agreed to request SNOMED revisions and use their own MeSH terms to provide necessary short term additions or clarifications and facilitate literature searches
- NLM will create or find Unique Concept Identifiers (CUI) for each term and the ACMG acronym will be the short name

Dataset and Code Development at HITSP

- It is important for all stakeholders to participate in or monitor the interoperability specification development at HITSP
- Fields and value sets for newborn screening test ordering will capture birth history
- LOINC codes for qualitative results based on “condition screened for” and quantitative results based on “analytes measured” will enable electronic report messages and documents
- Electronic hearing screening reports will be needed
- ACMG acronyms will become short names for SNOMED diagnosis codes on EHR problem lists and public health reports to data systems and registries
- A standard consult and referral document will be used

Dataset Development for Long Term Follow-up at ACHDNC and Elsewhere

- Understanding the effectiveness and impact of newborn screening will require long term follow-up and outcomes assessment
- Building the quality measures and outcomes datasets will be the next important newborn screening coding and terminology activity and these activities are underway at ACHDNC and elsewhere
- Codes for outcome measures aligned with newborn screening conditions will enable the use case messages and documents to carry this information
- Section 1139A(d)(1)(D) and 1139A(f) of the Children's Health Insurance Program Reauthorization Act of 2009 call for a demonstration project of the impact of model electronic health record format for children as part of quality measures for child health
 - IT will “allow interoperable exchanges that conform with Federal and State privacy and security requirements”
 - be “structured in a manner that permits parents and caregivers to view and understand the extent to which the care their children receive is clinically appropriate and of high quality”
 - and “capable of being incorporated into, and otherwise compatible with, other standards developed for electronic health records.”
 - Newborn screening could be a foundational component

Next Steps and Migration to NLM UMLS

- Migration to the National Library of Medicine Unified Medical Language System will provide a permanent and publically accessible home for the terminology work of the PHCWG
 - Will include new tests and methods of screening as well as associated new codes through collaboration with ACHDNC
 - Will address harmonization across different standards and coding systems
 - May expand to include more genomic information and links to other databases
- Final interoperability specification will include coding requirements
- Quality assurance measures are under development
- Monitoring adoption of standards including terminology will be critical to enabling interoperability

Collaboration between NLM and ACHDNC

- To facilitate the development of UMLS unique concept identifiers for all ACMG conditions under consideration at ACHDNC, NLM will require a fixed starting point and regular six month updates to UMLS
- The Coding and Terminology Guide developed by the Personalized Healthcare workgroup is proposed for use as that starting point along with the current LOINC subset for newborn screening which was also a product of the PHCWG
- Additional conditions under consideration should be added
- The precision and granularity of codes should be affirmed
- The UMLS CUI and MeSH terms will enable literature searching, clinical problem list entries, uniform laboratory reports, and collection of research data to support NBS decision making

Questions and Comments

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