

# Newborn Screening Interoperability Specification

Alan E Zuckerman MD  
Department of Pediatrics, Georgetown University  
National Library of Medicine, Lister Hill Center  
Washington DC  
aez@georgetown.edu

# Overview

- The completion of the HITSP/IS92 Newborn Screening Interoperability Specification represents the culmination of several years of work on the Newborn Screening Use Case
- The specification focuses on electronic transfer of results from the laboratory to the electronic health record (EHR) building on capabilities that will be required in every certified EHR
- This advisory committee will oversee activities at NLM to maintain and refine the specific coding and content of the messages
- Interoperability will improve the efficiency and accuracy of gathering evidence on newborn screening through long term follow-up
- Comments to ONC on the Interim Final Rule on Standards and to CMS on Meaningful Use and EHR Incentives could introduce newborn screening into phase 2 quality measures

# History and Timeline of the Newborn Screening Use Case

- The use case began as a project of the Newborn Screening Sub-Group of the AHIC Personalized Healthcare Workgroup in Aug, 2007
- The Final Interoperability Specification approval is expected at the HITSP Panel meeting Jan 20, 2010
- The specification is ready for immediate use based on extensive comments, inspection testing, and consensus building
- Several vendors have expressed interest in beginning implementation soon
- Several HRSA grantees will begin using the specification during the next year
- Rapid adoption over the next two years will enable including newborn screening in measures of meaningful use of EHR

# The Use Case Was One of Four AHIC Recommendations

- Development of a use case was only one of the AHIC recommendations on newborn screening
- Development of a Privacy document was also recommended and is available on the [healthit.hhs.gov](http://healthit.hhs.gov) web site
- Development of coding and terminology standards was also recommended and resulted in the NLM [newbornscreeningcodes.nlm.nih.gov](http://newbornscreeningcodes.nlm.nih.gov) web site
- The recommendation on monitoring and promoting adoption of electronic data exchange for newborn screening could become an activity of the ACHDNC through existing data gathering resources

# Key Features of the Final Newborn Screening Use Case

- The scope of the NBS use case is focused primarily on delivery of initial screening results to an EHR
- The use case selected the existing HL7 v2.5.1 based EHR lab results capability that is required for EHR certification
- This kind of message is used in every lab in the US and they are designed for Machine reading. Tools are also available to view the data in familiar format or display them in an EHR
- The HITSP selected the NLM Newborn Screening Codes as the source of codes for conditions and analytes
- The electronic ordering of NBS will use new capabilities and LOINC codes to capture data now entered manually on the filter paper card
- We recognize that EHDI hearing screening may require separate orders and reports from the dried blood spot screening and the messages support combined or separate reporting
- All events and actions in the use case diagrams now have proposed standards including consents and delivery of policies and education materials

# Inspection Testing and Public Comments

- Inspection testing has determined that the HITSP specification will meet the needs of individual states and will be an on-going process as revisions are requested
  - SNOMED codes for conditions
  - LOINC codes for tests and quantitative results
  - LOINC codes for fields on the filter paper
  - Use of existing HL7 message segments and codes for demographics
- The sample messages and the LOINC code panel provided by NLM illustrate how to accommodate variations of existing state NBS reports
- LOINC codes were created for interpretation of newborn screening tests by condition using the same SNOMED codes that will appear on problem lists
- Reporting of all quantitative results is encouraged and supported by LOINC codes
- Both summary and detail reporting by category or specific conditions are possible

# Capturing Information that is Entered on the Filter Paper Cards

- The Interoperability Specification includes electronic ordering of the newborn screening metabolic and hearing tests
- Data which is entered manually on the cards can now be sent electronically from a hospital information system or entered at the laboratory as in current systems
- The data fields from the filter paper card or hearing screening risk factor identification are included in the LOINC code panel and the sample HL7 message
- These have been reviewed carefully and some fields, such as mother's information, appear in standard segments of the HL7 messages

# Developing a Library of Newborn Screening Electronic Report Examples

- The comprehensive LOINC Newborn Screening Panel distributed to the ACHDNC includes all possible codes that may be used in message and the sample message illustrates most of them
- More practical examples of typical reports generated by specific newborn screening labs will be made available to illustrate current paper reports, the equivalent data representation in an HL7 message, and a simulated EHR lab result display
- Electronic messaging can be supplemented by more familiar web and PDF documents, but electronic messages can be imported into the EHR and used for research



# Example of Filter Paper Data



OBR|4|2001178109|10A1912105|57794-0^Newborn screening test results panel^LOINC| Routine| |200907061145|||||200907152000|BLD|000001||||  
|||||||2001178109^10A1912102 ||

OBR|5|2001178109|10A1912106|53261-4^Amino Acid NBS panel^LOINC^3500^AMINO REP^AMINO REP^Amino Acid Profile\*MAIN^LOCAL| Routine| |200907061145|||||  
200907152000|BLD|000001|||||||2001178109^10A1912105 ||

OBX|1|CE|46733-2 Amino acidemias NBS interpretation ^LOINC ^3500^AMINO REP^Amino Acid Profile^ LOCAL|1|Normal|N| |||F|||20090714074206

OBX|2|TX|46733-2 Amino acidemias NBS comment-discussion^LOINC |1| Any baby with clinical features suggestive of a metabolic disorder requires clinical and diagnostic follow-up regardless of whether the NMS result is normal or abnormal. || |||F|||20090714074206

# Example of Specimen Quality

2010-01-07 NLM HL7 NBS example.pdf - Adobe Acrobat Pro

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OBX|2|CE|57718-9^Specimen quality of dried blood spot^LOINC|1|LA12432-3  
^Acceptable^LOINC^ Await **New-SNOMED CT** code^New SNOMED-CT  
name^SNOMED CT|| |||F||| 20090714143552

## Full Answer List for 57718-9:

SEQ#	Answer	Code	Answer ID
1	Acceptable		LA12432-3
2	No sample received		LA12433-1
3	Specimen quantity insufficient for testing		LA12443-0
4	Specimen appears scratched or abraded		LA12682-3
5	Specimen not dry before mailing		LA12683-1
6	Specimen appears supersaturated		LA12684-9
7	Specimen appears diluted, discolored or contaminated		LA12685-6
8	Specimen exhibits serum rings		LA12686-4
9	Specimen appears clotted or layered		LA12435-6
10	No blood		LA12687-2
11	Sample too old		LA12441-4

**\*\* required \*\***

OBX|3|CE|57130-7^Newborn screening report - overall interpretation^LOINC|1|  
LA12430-7^Not normal requiring further filter paper testing for at least one  
condition^ LOINC| ||A|||F||| 20090714143552

# Example of Test Interpretation



**\*\* required \*\***

OBX|3|CE|57130-7^Newborn screening report - overall interpretation^LOINC|1|  
LA12430-7^Not normal requiring further filter paper testing for at least one  
condition^ LOINC| ||A||F||| 20090714143552

**Full Answer List for 57130-7:**

SEQ#	Answer	Code	Answer ID
1	All screening is normal		LA12428-1
2	Screen is borderline for at least one condition		LA12429-9
3	Not normal requiring further filter paper testing for at least one condition		LA12430-7
4	Not normal requiring immediate non-filter paper follow-up for at least one condition		LA12431-5

**\*\* required \*\***

OBX|4|CE|57131-5^Newborn conditions with positive markers^LOINC|1|  
128596003 ^Medium-chain acyl-coenzyme A dehydrogenase deficiency  
^SNOMED CT^ LA12509-8^MCAD^LOINC | |A||F|||20090714143552

**\*\* required\*\***

OBX|5|CE|57131-5^Newborn conditions with positive markers^LOINC|2|  
124214007^Deficiency of steroid 11-beta-monooxygenase (disorder)^SNOMED  
CT ^LA12533-8^ CAH ^LOINC|| |A||F||| 20090714143552

# Example of Summary Reporting



OBR|4|2001178109|10A1912105|57794-0^Newborn screening test results panel^LOINC| Routine| |200907061145|||||200907152000|BLD|000001||||  
|||||||2001178109^10A1912102 ||

OBR|5|2001178109|10A1912106|53261-4^Amino Acid NBS panel^LOINC^3500^AMINO REP^AMINO REP^Amino Acid Profile\*MAIN^LOCAL| Routine| |200907061145||||||  
200907152000|BLD|000001|||||||||||2001178109^10A1912105 ||

OBX|1|CE|46733-2 Amino acidemias NBS interpretation ^LOINC ^3500^AMINO REP^Amino Acid Profile^ LOCAL|1|Normal|N| |||F|||20090714074206

OBX|2|TX|46733-2 Amino acidemias NBS comment-discussion^LOINC |1| Any baby with clinical features suggestive of a metabolic disorder requires clinical and diagnostic follow-up regardless of whether the NMS result is normal or abnormal. || |||F|||20090714074206

# Example of Quantitative Results

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OBX|3|ST|53150-9^Alanine+Beta Alanine+Sarcosine  
^LOINC^3501^ALA^LOCAL|1|1236.06|umol/L| |||F||| 20090714074206

OBX|4|ST|47562-4^Arginine^LOINC^3502^ARG^LOCAL |1|5.89| umol/L|<90|  
|N||F|||20090714074206

OBX|5|ST|42892-0^Citrulline^LOINC^3503^CIT^LOCAL|1| 19.4| umol/L|<55|N|  
||F|||20090714074206

OBX|6|ST|47679-6^Leucine(LEU)^ LOINC^3505^LEU^LOCAL|1|162.52|umol/L  
|<250|N|||F|||20090714074206

OBX|7|ST|47700-0^Methionine^LOINC^3506^MET^LOCAL|1|45.97|umol/L| 44-  
49|N|||F|||20090714074206

OBX|8|ST|29573-3^Phenylalanine^LOINC^3508^PHE^LOCAL |1|104.61|  
umol/L|99-135|N|||F|||20090714074206

OBX|9|ST|35571-9^Tyrosine^LOINC^3509^TYR^LOCAL|1| 281.53| umol/L|  
205-223|N|||F|||20090714074206

OBX|10|ST|47799-2^Valine^LOINC^3510^VAL^LOCAL|148.56| umol/L|140-  
110|N|||F|||20090714074206

# Complete Panel Listing

54089-8\_NewBornScreen\_20100106.pdf - Adobe Acrobat Pro

## 54089-8 Newborn screening panel American Health Information Community (AHIC)

### PANEL HIERARCHY

LOINC#	LOINC Name	R/O/C	Cardinality	Data Type	UCUM	Units
54089-8	Newborn screening panel American Health Information Community (AHIC)					
57128-1	Newborn Screening Report summary panel	R	0..1			
57721-3	Reason for lab test in Dried blood spot	R	1..1	CE		
57718-9	Sample quality of Dried blood spot	O	1..1	CE		
57130-7	New born screening report - overall interpretation	R	1..1	CE		
57131-5	Newborn conditions with positive markers [Identifier] in Dried blood spot	R	1..n	CE		
57720-5	Newborn conditions with equivocal markers [Identifier] in Dried blood spot	R	1..n	CE		
57724-7	Newborn screening short narrative summary	O	0..1	FT		
57129-9	Full newborn screening summary	O	0..1			

# Example of Answer Lists

57720-5 Newborn conditions with equivocal markers [Identifier] in Dried blood spot

**NORMATIVE ANSWER LIST:**

SEQ#	Answer	Global ID	Global ID Code System	Code	Answer ID
0	None				LA137-2
1	HEAR	15188001	SN		LA12463-8
2	2M3HBA				LA12464-6
3	2MBG				LA12465-3
4	3-MCC	13144005	SN		LA12466-1
5	3-MCC (mat)	13144005	SN		LA12467-9
6	3MGA	237950009	SN		LA12468-7
7	5-OXO	39112005	SN		LA12469-5
8	ARG	23501004	SN		LA12470-3
9	ASA	41013004	SN		LA12471-1
10	BIOPT-BS	237914002	SN		LA12472-9
11	BIOPT-REG	58256000	SN		LA12473-7
12	BKT	237953006	SN		LA12474-5
13	CACT	238003000	SN		LA12475-2
14	CBL A	73843004	SN		LA12476-0
15	CBL B	82245003	SN		LA12477-8
16	CBL C	74653006	SN		LA12478-6
17	CBL D	31220004	SN		LA12479-4
18	CBL E	4409006	SN		LA12480-2
19	CBL G	360373000	SN		LA12481-0
20	CIT I	238680004	SN		LA12482-8

# Coordination with Activities at IHE and HIMSS

- Integrating the Healthcare Enterprise (IHE) is a voluntary organization that develops Integration Profiles to demonstrate interoperability and use of standards
- They hold a Connect-a-thon in Jan to demonstrate and certify compliance and an Interoperability Showcase at HIMSS (the largest Health IT meeting)
- IHE has selected Newborn Screening and a Newborn Discharge Summary for their next cycle which will culminate in the Connect-a-thon in Jan, 2011
- Work at IHE will engage vendors in implementing newborn screening interoperability showcase prototype products and speed adoption
- Adding newborn screening results to a Hospital Newborn Discharge Summary will assist hospitals in making this data available automatically after newborns leave the hospital



# Next Steps and Potential Action Items

- While Public Comments on the HITSP Interoperability are closed, inspection testing will continue and NLM will continue to accept comments and make necessary changes to messages and codes
- Vendor participation in the IHE Connect-a-thon in Jan 2011 will demonstrate interoperability of NBS
- Coordination of all NBS datasets and codes through NLM and ACHDNC is where the real use case work will continue to take place and close coordination with HRSA is needed
- Exploration of new approaches to the NHIN is underway at a HIT Policy Committee Workgroup and should assure that the NHIN will include newborn screening. This work is beginning with exploring directories to assist delivery of medical information
- Encouraging CMS to include newborn screening in future regulations on Meaningful Use of EHR will speed adoption

# Roles for the ACHDNC to Drive Implementation at the State Level

- The NBS Interoperability Specification should become the foundation for gathering follow-up information the follow-up and data committees should coordinate their needs with maintenance of the messages and codes. The ACHDNC will be an important user of interoperable newborn screening data
- The advisory committee could encourage all states to continue preparation for electronic messaging by inspection testing of the standards and developing a plan for how each state will use the standards. ACHDNC should consider a letter to the states
- Opportunities to assist states with implementation should be explored and collaboration encouraged
- Comments to ONC and CMS on Meaningful Use Regulations could provide incentives in phase 2

# State Planning for Implementation of the Interoperability Specification

- The specification is an important opportunity for all states to review their data reporting practices and to select the codes and methods they will use when opportunities for system change become available
- Vendors need the states to specify their requirements in terms of the codes and messages of the specification
- Every state needs a plan that identifies the data they will process and how it will be coded

# Comments to ONC and CMS on ARRA Meaningful Use Regulations

- We are now in the middle of a 60 day window of opportunity to comment on regulations released by ONC and CMS on December 30, 2009 as required by the American Recovery and Reinvestment Act (ARRA)
- While newborn screening is not included in phase 1, the framework has been created to include newborn screening as a measure in phase 2
- Incentives will clearly drive adoption and improve the flow of data for newborn screening, but it is essential to move quickly now to make electronic newborn screening widely available in the next 1-2 years