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An update on the use of health information technology in newborn screening

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ABSTRACT

Newborn screening (NBS) has high-stakes health implications and requires rapid and effective communication between many people and organizations. Multiple NBS stakeholders worked together to create national guidance for reporting NBS results with HL7 (Health Level 7) messages that contain LOINC (Logical Observation Identifiers Names and Codes) and SNOMED-CT (Systematized Nomenclature of Medicine–Clinical Terms) codes, report quantitative test results, and use standardized computer-readable UCUM units of measure. This guidance (a LOINC panel and an example annotated HL7 message) enables standard HL7 v2.5.1 laboratory messages to carry the information required for reporting NBS results. Other efforts include HL7 implementation guides for reporting point-of-care (POC) NBS results as well as standardizing follow-up of patients diagnosed with conditions identified through NBS. If the guidance is used nationally, regional and national registries can aggregate results from state programs to facilitate research and quality assurance and help ensure continuity of operations following a disaster situation.

Introduction

Newborn screening (NBS) is a complex system of public health laboratories and follow-up programs, hospitals, clinicians, courier services, and families with newborns. The information flow and communication network involved in NBS combined with the growing adoption of electronic health records (EHRs) and electronic exchange of laboratory test results, created an opportunity to develop consensus-based standard vocabularies that would enable NBS health information exchange (HIE) as well as provide a foundation for establishing research and quality measures.[1–3] In 2009, Downing et al.[3] described the potential benefits of using health information technology (HIT) for NBS and the initial

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steps that had been taken to enable the use of HIT. In this update, we summarize the efforts over the last 5 years to make HIT use for NBS a reality.

Background

Members of multiple government agencies, NBS programs, and laboratories worked together to develop standardized guidance for electronic reporting of NBS results[4] using nationally accepted vocabulary[5,6] and electronic messaging standards: (1) LOINC (Logical Observation Identifiers Names and Codes) contains standard codes for identifying laboratory tests and other clinical measures; (2) SNOMED CT (Systematized Nomenclature of Medicine—Clinical Terms) is an international terminology standard for systematically specifying symptoms and diagnoses; (3) UCUM (Unified Code for Units of Measure) specifies the units for a given test or measure in a standard, machine-readable format; and (4) HL7 (Health Level 7) specifies the standards for electronic messaging. The guidance includes a comprehensive LOINC panel and an example annotated HL7 message that states can use as a template to develop their specifications for transmitting electronic NBS result messages.[7] The National Library of Medicine (NLM) has continued to refine and add to this guidance, as new conditions are added to NBS.

Other efforts in the area of NBS HIT include publication of implementation guides for NBS dried blood spot (DBS) orders[8] and results[9] reporting and balloting of HL7 implementation guides for point-of-care (POC) critical congenital heart disease (CCHD)[10] and infant hearing[11] screening. A 2013 survey conducted by the Association of Public Health Laboratories (APHL), which is described in more detail below, assessed different state NBS programs' progress in implementing HIT systems for NBS. This HIT work is also evolving beyond NBS, to short- and long-term follow-up, because the infant NBS is only the first step in a long continuum of care.

HRSA/NLM HIT guidance for NBS

In 2009, the NLM and the Health Resources and Services Administration (HRSA) created a LOINC panel that covered all of the conditions stated on the Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children's (SACHDNC) Recommended Uniform Screening Panel (RUSP)[12] as well as other conditions that were screened by any U.S. NBS program.[13] It included amino acid, acylcarnitine, endocrine, and hemoglobin disorders, as well as infant hearing screening. The NLM/HRSA guidance also included an example HL7 NBS result message with detailed annotations that was meant to be a primer on electronic messaging and data standards. The NLM worked closely with several state NBS programs, reviewed their early test HL7 messages, and refined the LOINC panel and HL7 messaging guidance based on their feedback. This work was reviewed by the SACHDNC Laboratory Standards and Procedures Subcommittee and first published on the NLM NBS website (http://newbornscreeningcodes.nlm.nih.gov) in September 2009.

The messaging guidance included help for transitioning from local codes to standard codes or, for the systems that need to continue using local codes, for incorporating both into their HL7 messages. For example, the HL7 message can include both the standard LOINC code and test name (in italics for illustrative purposes) and the local code and test name (underlined, also for illustrative purposes) for the third field (observation ID) of the observation (OBX) segment:

OBX|4|NM|*59407-7^Oxygen saturation in Blood Preductal by Pulse oximetry^LN*^Pre-SaO2^Preductal O2 saturation^L||99|%|>95|N|||F

The fifth field of the OBX segment can carry both a standard SNOMED CT code and name (in italics) and either a LOINC answer (LA) code and name or a local code and name (underlined) for the NBS condition:

OBX|4|CE|57131-5^Newborn conditions with positive markers [Identifier] in Dried blood spot^LN|1|**128596003**^*Medium-chain acyl-coenzyme A dehydrogenase deficiency^SCT*^ MCAD^Med-chain acyl-coenzyme A dehydro def^L|||A|||F

Since the original LOINC panel and HL7 example message were published, NLM has been continuously refining the guidance based on feedback from NBS programs and other stakeholders. As an example, the original LOINC panel included a method for reporting hemoglobin screening results that was based on hemoglobin patterns, which was unsustainable, as the number of hemoglobin variants that states could identify continued to grow. The NLM worked with hemoglobin and NBS experts from multiple federal and state agencies and NBS programs to devise a new method for reporting hemoglobin screening results based on reporting the individual hemoglobin variants suspected rather than the pattern or peaks observed. The NLM and the Regenstrief Institute created a LOINC panel containing five LOINC codes for reporting up to five suspected hemoglobins in a specimen in terms of their relative concentrations (Table 1). Depending on the number of hemoglobins suspected in a given sample, one to all the five codes can be used in separate HL7 message segments. For example, the following three HL7 OBX segments together represent the three hemoglobin types suspected (F, A, and S) in the order of decreasing concentration in a single NBS specimen:

OBX|1|CE|64117-5^ Most predominant hemoglobin ^LN^^^ |1| LA16208-3^Hb F^LN ||||||F||| 2009071414 5203
OBX|2|CE|64118-3^Second most predominant hemoglobin^LN^^^ |1|LA11112-2^Hb A^LN ||||||F||| 20090714145203
OBX|3|CE|64119-1^Third most predominant hemoglobin ^LN^^^ |1| LA13007-2^Hb S^LN ||||||F||| 20090714145203

The new method is easy to understand and complies with HL7 messaging standards, while remaining straightforward to implement using LOINC codes.

In May 2011, the SACHDNC Laboratory Standards and Procedures Subcommittee suggested that the NLM review

Table 1 – LOINC panel 64116-7 for NBS suspected hemoglobin observations.

LOINC observation long common name	LOINC code
Hemoglobin observations newborn screening panel	64116-7
Most predominant hemoglobin in dried blood spot	64117-5
Second most predominant hemoglobin in dried blood spot	64118-3
Third most predominant hemoglobin in dried blood spot	64119-1
Fourth most predominant hemoglobin in dried blood spot	64120-9
Fifth most predominant hemoglobin in dried blood spot	64121-7
Hemoglobins that can be presumptively identified based on available controls in dried blood spot	64122-5

Panel of LOINC codes for reporting up to five hemoglobin types suspected to be present in a NBS dried blood spot sample.

the LOINC answer list for "Clinical events that affect newborn screening interpretation." The NLM reviewed the "clinical events" answer list with representatives from NBS programs and the APHL Newborn Screening and Genetics in Public Health Committee. They also conducted an evidence-based literature search to develop a proposal for revising it[14–16] and reached consensus for a revised set of LOINC codes and answer lists. Based on the findings, the NLM split the "Clinical events that affect newborn screening interpretation" into three separate LOINC observations: "Infant NICU Factors that Affect Newborn Screening Interpretation," "Maternal Factors that Affect Newborn Screening Interpretation," and "Feeding Types." Based on the infant's history, clinicians can select one or more responses from the answer list for each question. Each of these three LOINC codes with answer lists includes a definition/description, explaining which NBS assays are affected by particular feeding types, infant NICU, or maternal factors. Each of the three questions also offers an answer option "Other," if selected, narrative (Nar) data type LOINC codes are available to provide details.

New additions to NBS LOINC panel

As new conditions and methods have been added to NBS over the years, both the LOINC panel and the example HL7 message, which are adaptable, have been updated accordingly. When severe combined immunodeficiency (SCID) was added to the RUSP by the SACHDNC in May 2010, the NLM developed new condition and analyte variables for this condition. The SCID panel (LOINC code 62333-0) includes codes for the quantitative T-cell receptor excision circle (TREC) assay, test interpretation, and comment/discussion. Additional variables and codes can be added following addition of a new condition to the RUSP.

Critical congenital heart disease (CCHD) was added to the RUSP in 2011 as a second POC screening, joining early hearing loss. Many variables are captured and reported, and out-of-range results require immediate follow-up. While there are variations on CCHD screening protocols, most begin by considering a preductal and postductal oxygen saturation measurement and the difference between the two. Abnormal screens may be followed by diagnostic testing or subsequent measurements at specified intervals depending upon the initial result. To accommodate the intricacies of point-of-care screening for CCHD, the LOINC panel (code 73805-4) includes terms for the actual screening results (preductal and postductal oxygen saturation measurements), several variables that are related to the screening process (e.g., type of sensor and sensor wrap), and related physiologic measurements such as heart rate (Table 2). Each program can use the appropriate combination of the available CCHD codes to reflect its protocol.

Several NBS programs have started pilot studies or implemented screening for five of the lysosomal storage disorders (LSDs): Fabry disease, Pompe disease, Gaucher disease, Niemann–Pick disease A/B, and Krabbe disease. A workgroup of LSD experts analyzed variations in naming LSDs and the tests used for screening as part of a larger evidence review and

Table 2 – LOINC panel 73805-4 for newborn critical congenital heart disease (CCHD) screening.

LOINC observation long common name	LOINC code	UCUM units
CCHD newborn screening panel	73805-4	
CCHD newborn screening interpretation	73700-7	
Oxygen saturation.preductal-oxygen saturation.postductal (mass fraction difference) in Bld. preductal and Bld.postductal	73696-7	%
Newborn age in hours	73806-2	h
Number of prior CCHD screens (#) Qualitative	73699-1	
Oxygen saturation sensor name	73804-7	
Oxygen saturation sensor type	73803-9	
Oxygen saturation sensor wrap name	73802-1	
Oxygen saturation sensor wrap type	73801-3	
Oxygen saturation sensor wrap size	73800-5	
CCHD newborn screening protocol used (type)	73697-5	
Reason CCHD oxygen saturation screening not performed	73698-3	
Oxygen saturation in blood preductal by pulse oximetry	59407-7	%
Heart rate blood preductal pulse oximetry	73799-9	/min
Perfusion index blood preductal pulse oximetry	73798-1	%
Signal-quality blood preductal pulse oximetry	73797-3	
Infant activity during preductal oxygen saturation measurement	73796-5	
Oxygen saturation in blood postductal by pulse oximetry	59418-4	%
Heart rate blood postductal pulse oximetry	73795-7	/min
Perfusion index blood postductal pulse oximetry	73794-0	%
Signal-quality blood postductal pulse oximetry	73793-2	
Infant activity during postductal oxygen saturation measurement	73792-4	

Panel of LOINC codes to report newborn CCHD screening results and associated information.

guideline development process on the diagnosis and management of the presymptomatic LSD patient.[17] Informed by this work, the NLM and the HRSA collaborated with the workgroup plus other LSD and NBS experts, selected standard names, assigned standard LOINC and SNOMED CT codes to LSD tests and conditions, and updated the example electronic HL7 message to illustrate how to use these codes to report NBS LSD results.

The NLM and the Regenstrief Institute also worked with Oregon's NBS program to develop new LOINC codes for analytes measured using the non-derivatized tandem mass spectrometry (MS/MS) test kit method. The methods for derivatized and non-derivatized MS/MS are similar and nearly all reports of analyte values and ratios will be identical; however, by consulting with experts, we identified exceptions for which we obtained new LOINC codes.

HL7 messaging guides

In 2011, the Public Health Informatics Institute (PHII), funded by a HRSA grant, published implementation guides for NBS dried blood spot (DBS) orders[8] and results[9] based on HL7 messaging. PHII worked with representatives from public health laboratories, public health agencies, and NLM. NewSTEPs (Newborn Screening Technical assistance and Evaluation program), a HRSA-funded cooperative agreement that is a collaboration between APHL and the Colorado School of Public Health, is building on the PHII work by finalizing the HL7 implementation guide for NBS DBS results and submitting it for formal HL7 balloting.

In 2012, implementation guides for reporting CCHD and EHDI results were balloted through HL7. The NLM helped align the HL7 guides with the existing LOINC codes and updated the existing codes as necessary. In conjunction with this work, the NLM revised the NBS LOINC panel to create a separate subpanel for POC results.

State progress in implementing HIT for NBS

The NLM has worked closely with several state NBS programs to help map their local codes to LOINC and review their preliminary HL7 messages. At least 15 states are mapping to standard LOINC and SNOMED CT codes and building infrastructure to exchange NBS orders and results with hospitals and providers, public health, or health information exchanges (HIEs) via HL7 messages based on the HRSA/NLM guidance. To further enrich and better utilize the NBS data, several states are working to integrate their NBS systems with related systems, including long-term follow-up, vital records (birth certificates), and immunization registries. "Please contact the corresponding author for more information if you have questions about specific state NBS programs."

APHL conducted a survey of U.S. NBS programs in 2013, in order to understand the current landscape of health HIT implementation for NBS. One representative from each NBS program in the country received the survey electronically, with respondents encouraged to seek information from other individuals (including HIT personnel) when completing the survey. Survey participation was voluntary. In accordance with APHL's Data Access and Sharing policy, the reports and findings related to this survey were released only in aggregate data form without individual identifiers.[18] Overall, 33 NBS programs participated in the survey; however, some answers were completed by a subset of the total. Internal data integration between laboratory and follow-up programs was reported in 27/32 programs, with 23/27 reported to be fully automated internal data sharing. Data exchange with entities outside the NBS program was reported by 15/32 (47%) of the programs. The programs exchange NBS data with birthing hospitals ($n = 9$), physician offices ($n = 6$), vital records ($n = 7$), immunization registries ($n = 1$), early hearing detection intervention data systems ($n = 6$), CCHD data systems ($n = 1$), and birth defects registries ($n = 11$). Of programs exchanging data, the most common type of information exchanged was NBS results, exchanged in 13 programs (HL7 = 10 programs, CSV file = 3 programs), while NBS orders are exchanged by 6 programs (HL7 = 6/6 programs).

Further, NBS screening program data sharing happens through web portals where users can access NBS results through a secure website (13/30, 43%), allowing access to NBS results to authorized users 24 hours a day. In addition to results on individual newborns, program performance metrics are available through web portals in some programs, reporting information such as the timing of specimen receipt by lab and subsequent result turnaround time ($n = 5$), number of rejected samples due to inadequate collection ($n = 7$), and other *ad hoc* reports that are unique to an individual program ($n = 3$). The web portals can be accessed by physicians, midwives, hospitals, and other state NBS programs when services are shared through regional laboratories.

The biggest challenges for state NBS programs in implementing NBS HIT were competing priorities (22/33, 67%), lack of HIT expertise (14/33, 42%), and lack of funding (14/33, 42%).

Beyond NBS

Because NBS is only a first step in a longer continuum of care, the NLM's collaboration with NBS programs has also contributed to the development of other codes such as diet monitoring codes for children with phenylketonuria (PKU) and other conditions diagnosed after NBS. This work began with a request from the Oregon NBS program for certain codes because the Oregon NBS lab processes samples from patients with metabolic disorders in Alaska whose parents periodically draw DBS samples at home and send to Oregon for processing. The NLM gathered information from state as well as international NBS programs on the full scope of conditions monitored by DBS and the analytes used for monitoring and worked with Regenstrief Institute to create a LOINC panel with codes for dietary monitoring of conditions diagnosed on NBS (Table 3).

Discussion

NBS is on the forefront of public health adoption of health information technology. Using standard codes and names will

Table 3 – LOINC panel 74874-9 for monitoring metabolic disorders using dried blood spots (DBS).

LOINC observation long common name	LOINC code	UCUM units
Metabolic disorder therapy monitoring panel—dried blood spot	74874-9	
Sample quality of dried blood spot	57718-9	
Reason for unsatisfactory specimen not related to sample quality of dried blood spot	74482-1	
Metabolic disorder being monitored (Identifier) in dried blood spot	74873-1	
Alloisoleucine (moles/volume) in dried blood spot—posttherapeutic diet	74875-6	μmol/L
Arginine (moles/volume) in dried blood spot—posttherapeutic diet	74876-4	μmol/L
Argininosuccinate (moles/volume) in dried blood spot—posttherapeutic diet	74877-2	μmol/L
Carnitine free (C0) (moles/volume) in dried blood spot—posttherapeutic diet	74878-0	μmol/L
Galactose (mass/volume) in dried blood spot—posttherapeutic diet	75093-5	mg/dL
Galactose 1 phosphate (mass/volume) in dried blood spot—posttherapeutic diet	74879-8	mg/dL
Galactose 1 phosphate uridyl transferase (enzymatic activity/volume) in dried blood spot—posttherapeutic diet	75094-3	U/g (Hb)
Hydroxyproline (moles/volume) in dried blood spot—posttherapeutic diet	74880-6	μmol/L
Isoleucine (moles/volume) in dried blood spot—posttherapeutic diet	74881-4	μmol/L
Leucine (moles/volume) in dried blood spot—posttherapeutic diet	74882-2	μmol/L
Ornithine (moles/volume) in dried blood spot—posttherapeutic diet	74883-0	μmol/L
Phenylalanine/tyrosine (molar ratio) in dried blood spot—posttherapeutic diet	74300-5	(ratio)
Phenylalanine (moles/volume) in dried blood spot—posttherapeutic diet	74303-9	μmol/L
Succinylacetone (moles/volume) in dried blood spot—posttherapeutic diet	74301-3	μmol/L
Tyrosine/phenylalanine (molar ratio) in dried blood spot—posttherapeutic diet	74299-9	(Ratio)
Tyrosine (moles/volume) in dried blood spot—posttherapeutic diet	74302-1	μmol/L
Valine (moles/volume) in dried blood spot—posttherapeutic diet	74884-8	μmol/L

Panel of LOINC codes to report DBS monitoring results for metabolic disorders diagnosed by NBS.

enable clinicians, public health surveillance efforts, and researchers to exchange and aggregate NBS results from all of the states, which is critical for quality assurance, quality improvement, and research to identify better methods in NBS. Full implementation of health information exchange within NBS will facilitate a more efficient system, resulting in a more timely turnaround for NBS results and a decreased workload in NBS programs. While there are still some challenges ahead to reach full implementation, there is a momentum toward integrating data exchange capabilities in NBS programs. NBS laboratory instrument and information systems vendors including PerkinElmer, OZ Systems, and Natus Neometrics have incorporated NBS LOINC codes and HL7 messaging for NBS based on the HRSA/NLM guidance into their systems and are working with state NBS programs to facilitate adoption of HIT for NBS. As NBS programs adopt data exchange models and new state-of-the art data systems, they are adapting reporting capabilities for each hospital and provider served, a process that is unique to each hospital and provider.

NBS programs that have legacy electronic systems or paper-based systems that use local codes can send both local and standard codes in their HL7 messages as a way to preserve backwards compatibility during the transition from legacy local coding systems to national standard codes. Electronic messaging allows NBS programs to send data to multiple recipients at the same time, including the birth hospital, post-discharge provider and practice, metabolic specialist, health information exchange, and state registry. Because some states may not want to send all quantitative results to all message recipients, they can utilize HL7 features such as normal/abnormal flags or filtering on specific LOINC codes to send specific results or types of results to the selected categories of message recipients.

The authors believe that when results are out of range or indeterminate, laboratories should send the quantitative results to the birth institution and attending clinicians, particularly when fixed cutoffs are used.[3] Pediatric subspecialists who work with state NBS programs anecdotally state that they would prefer to have interpretations rather than quantitative results reported to the birth hospital and pediatric providers because they want to avoid misinterpretation of results and unnecessary phone calls and referrals. However, quantitative data can be valuable for interpreting the results and providing concrete information to the family until the infant sees the appropriate specialist or has follow-up testing done. In cases where dried blood spot result ranges differ from the usual serum results familiar to primary care physicians, minimal educational information delivered with the results should assist them in result interpretation. Furthermore, we encourage NBS laboratories to report all quantitative results (not just interpretations) and appropriate accompanying explanatory information to the NBS follow-up programs so information can be maintained for comparison over time.[3]

Considering that many states share a regional laboratory or use the same laboratory information management systems (LIMS) software, there is an opportunity for states to share their HIT adoption methods with other programs to reduce costs. In addition, open source MIRTH software[19] and commercial BizTalk software can facilitate development of messaging by adapting existing functions to local needs through use of mapping tables, and implementation protocols could be shared between early implementers and other programs. Further collaborations can be considered between LIMS and large hospital systems that may share a common electronic health record system.

Conclusion

While standard codes and electronic messaging guidance are important, NBS program resources and infrastructure for implementing HIT are as important. Much progress has been made over the last 5 years in both standards development and NBS program implementation. We hope adoption of HIT in NBS will continue to grow, because using standard codes and names as well as electronic messaging has the potential to allow faster results reporting, improve patient follow-up and continuity of care, and facilitate aggregation of NBS results across programs.

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