Standardizing Newborn Screening Results for Health Information Exchange

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Abstract

Newborn screening is a complex process that has high-stakes health implications and requires rapid and effective communication between many people and organizations. Currently, each state program has its own method of reporting results, with wide variation in content and format. Pediatric care providers receive reports by mail, email, fax or telephone, depending on whether the results are normal or abnormal. This process is slow and prone to errors, which can lead to delays in treatment. Multiple agencies worked together to create national guidance for reporting newborn screening results with HL7 messages that contain a prescribed set of LOINC and SNOMED CT codes, report quantitative test results, and use standardized units of measure. Several states are already implementing this guidance. If the guidance is used nationally, office practice systems could capture NBS results more efficiently in EHRs, and regional and national registries could better analyze aggregate results to facilitate further research for these rare conditions.

Introduction

Newborn screening (NBS) is a vital process that identifies apparently healthy infants with serious medical conditions so they can be treated before they suffer significant morbidity or mortality. NBS includes both dried blood spot (DBS) and hearing tests. Most NBS conditions are rare and comparing data across states is necessary to optimize screening protocols and assess screening outcomes. Until this project began, there was no standard for reporting NBS results, and therefore no way to efficiently transmit data to pediatric care providers, or to reliably compare or pool data across states. In this report we describe a standard way to deliver newborn screening results in a Health Level Seven (HL7) message.

Background

In the United States, NBS programs are operated by fifty states plus the District of Columbia, certain U.S. territories and the military. Almost all of the programs test for the 29 core conditions defined by the Recommended Uniform Screening Panel of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.[1] Many also screen for various additional conditions. Most programs recommend DBS screening at 24-48 hours after birth, and hearing screening at least 24 hours after birth but before hospital discharge. Nine states require, and others recommend, a repeat DBS screen at one to two weeks of age such that about 25% of US newborns receive two screens. The newborn's blood is obtained by heel stick, and collected on special filter paper attached to a collection card that includes questions ("card variables") about the newborn and mother. Metabolic, hematologic, endocrine and other abnormalities are screened using various tests on the DBS. Congenital hearing loss is screened by physiologic measures.

Programs differ not only in the number of conditions screened, but also in how the results are reported. Each program uses its own local non-standard codes for tests and results. Some programs report only qualitative interpretations of results ("normal"), while others use various combinations of narrative, qualitative and quantitative reporting. Even quantitative results can vary – some programs report numeric values, while others result ranges (e.g. 2.3 vs. <10 or 5-7) or percentiles. Some programs report results for individual conditions or the individual test measure, while others group results based on disorder categories, with some variations in grouping among states (e.g. amino acids, fatty acids and acylcarnitines). Given all of this variability, it is very difficult for office practice system developers to capture NBS results efficiently into electronic health records (EHRs), and for regional or national registries to collect and analyze NBS information.

NBS programs report positive DBS results to pediatric care providers by phone due to the urgent need for follow up and treatment. Currently, most NBS programs use postal mail to send normal NBS reports to the birthing facility or pediatric care provider designated when the baby is screened; however, the provider who sees the baby in the hospital is often not the same one that follows the baby long-term. Some states do not send negative results as timely as they could, which can cause confusion and delay. In one survey of pediatricians, 26% reported they were not routinely notified of the screen-negative results. When asked hypothetically if they would actively track down NBS results for a 2-week-old infant with a normal exam, 28% reported they would not either because they assumed "no news is good news," the state does a good job, or a combination of "the infant is healthy and lack of report implies the test results were negative."[2] Although a few states provide websites or automated voice response systems where physicians can obtain screening results,[3] tracking down newborn screening results can require many attempts per baby, which is a burden on the office staff.

Newborn hearing screening results are hospital-based, not laboratory-based. The mechanism for reporting hearing screening results depends on the jurisdiction and, in some cases, hospital-level policy and procedures. Hospitals can communicate hearing screening results in various formats to stakeholders such as the family, the state Early Hearing Detection and Intervention (EHDI) program, and audiologists. This non-uniformity of communications processes is one barrier to effective hearing screening follow-up.

Methods

Our goal was to develop a template that could carry the DBS screening results and accommodate state variations in hearing screening and reporting styles. We used a hierarchy of nested Logical Observation Identifiers Names and Codes (LOINC®)[4] panels to create this template, following an approach that has been successful for many other complex LOINC data capture processes.[‡] This approach provides a way to organize variables in a nested structure with their associated attributes: data type, cardinality,[§] UCUM©[5] units of measure (for numeric variables), answer lists (for categorical variables), descriptions and help messages. The contents of this structure can be mapped to an HL7 message with each LOINC code and its corresponding test value carried in one OBX (observation result) segment within the HL7 message. Nesting can be reflected in the message by incorporating an OBR (observation request) segment for each node in the hierarchy.

The information in a LOINC panel can be represented by three relational database tables. One table carries a record for each LOINC term used in the panel with all of that term's attributes. The second describes the relationship of a nested LOINC panel to its observation codes as a hierarchy. Each record in the second table contains a link to a parent LOINC term in the first table and other attributes that vary for a given LOINC term across panels. The third table contains answer lists for all of the categorical questions in the panel.

We designed an all-inclusive LOINC NBS panel – called the American Health Information Community (AHIC) Newborn screening panel – based on the above structure so that a given NBS program can select the variables it needs for reporting conditions screened. Therefore, different states can include different subsets of tests in their test reports, but any result for a test that is the same across more than one state will be reported using the same LOINC code in the same format.

The U.S. Department of Health and Human Services Office of the National Coordinator for Health IT (ONC) obtained and analyzed DBS cards from all U.S. NBS DBS programs, and we developed a condensed set of questions and answer lists that covered the content represented in these cards. This standardized content included demographic information (such as baby and mother's name and contact information – which go into the HL7 Patient Identification (PID) and Next of Kin (NK1) segments respectively), as well as birth history information that laboratories and clinicians may use to interpret and analyze screening results (such as history of blood transfusion or antibiotic administration prior to specimen collection). We worked with many organizations and individuals to develop and refine the answer lists for card variables, overall screening impressions, hearing loss risk indicators, hemoglobin disorders and more.

The Centers for Disease Control and Prevention's (CDC) EHDI Program helped develop a single set of LOINC answer codes for hearing screening methodology, results, and hearing loss risk indicators. A LOINC answer list includes all of the hearing loss

‡ Including HL7 clinical genomic reporting, many large and complex Medicare forms (OASIS, MDS, CARE), HEDIS quality measures, laboratory test panels and standardized research measurements (PhenX and PROMIS).

§ Cardinality specifies whether the field is required, and whether you can have multiple repeated values.

risk indicators identified by the Joint Committee on Infant Hearing (JCIH) 2007 Recommendations.[6] In an HL7 message, a single LOINC code for "hearing loss risk indicators" can repeat as necessary across many HL7 OBX segments to carry information about multiple risk factors. When no risk factors are identified, a single OBX segment should be used with the answer code for "None."

The National Library of Medicine (NLM) also worked with the CDC National Center for Health Statistics Division of Vital Statistics to create LOINC codes and corresponding answer lists for several card variables that reflect information contained in the 2003 revisions of the U.S. Standard Certificate of Live Birth. These variables include date of birth, time of birth, obstetric estimation of gestational age and mother's education. Everything we did was reviewed and refined via feedback from many NBS experts and agencies as well as input during a formal Health Information Technology Standards Panel (HITSP) public comment period.

Regenstrief Institute assigned to all of the variables a unique LOINC code, units of measure, and cardinality as appropriate. For categorical variables, we defined formal answer lists and assigned each answer a placeholder LOINC answer code. We also included SNOMED CT codes (with permission from the International Health Terminology Standards Development Organisation) where available for the answers that represent the conditions, and, as they become available, we will add new SNOMED CT codes to other answer lists to facilitate universal interpretation.

The Interim Final Rule on Health Information Technology specified that electronic laboratory reports be transmitted as HL7 2.5.1 messages.[7] The AHIC Personalized Healthcare Workgroup's NBS Subgroup, with special help from the American College of Medical Genetics (ACMG), HRSA, the CDC EHDI Program, and the National Newborn Screening and Genetics Resource Center (NNSGRC), developed initial tables of NBS conditions screened in any state, associated measurements, and condition details.[8]

Finally, NLM worked with the Health Resources and Services Administration (HRSA) to develop guidance specifying how to construct HL7 newborn screening messages using the codes in the LOINC NBS panel, and developed an annotated example HL7 message as an embodiment of that guidance. This guidance harmonizes with the Public Health Informatics Institute Implementation Guide,[9] which focuses more on the administrative HL7 segments (e.g. MSH, PID, NK1), whereas this project focuses on detailed codes for the results "payload."

Beyond the organizations mentioned above, the effort to produce a standard NBS message also required the expertise and guidance of the HITSP Population Perspective Technical Committee, lab system vendors, and state NBS programs (hearing and DBS).

Results

The LOINC NBS panel includes a total of 219 LOINC codes including 18 panel codes (used to group LOINC codes), 153 codes representing measured results or calculations and 30 codes for reporting interpretations of, or comments/discussion about, NBS results. In addition to individual analyte measurements and interpretations, the LOINC NBS panel contains summary interpretations (Figure 1) and card variables. The specification provides coded OBX segments for transmitting comments, instead of note (NTE) segments. Three of the summary variables (overall interpretation, reason for lab test in dried blood spot, and sample quality of dried blood spot) have specific answer lists, which are based on recommended practices and federal reporting standards, and each answer has its own LOINC answer code (Figure 2).

LOINC code	LOINC Name of variable (question)	R/O	Card'y	Data type
57128-1	Newborn Screening Report summary panel	R	1..1	
57721-3	Reason for lab test in Dried blood spot	R	1..1	CE
57718-9	Sample quality in Dried blood spot	R	1..1	CE
57130-7	Newborn screening report – overall Interpretation	R	1..1	CE
57131-5	Newborn conditions with positive markers [Identifier] in Dried blood spot	R	1..n	CE

Figure 1. Excerpt of the LOINC hierarchy showing codes and attributes (required/optional, cardinality and data type) for four of the eight variables in the Newborn Screening Report summary panel.

SEQ #	Answer	SNOMED CT Code	LA code
31	GA-1	76175005	LA12493-5
32	GA-1 (mat)	76175005	LA12494-3
33	GA-2	22886006	LA12495-0
34	HCY	24308003	LA12496-8
35	HHH	30287008	LA12497-6
36	HIS	124628005	LA12498-4
37	HMG	124611007	LA12499-2
38	H-PHE	68528007	LA12500-7

Figure 2. Answer list excerpt for "Conditions tested for in this newborn screening study," with sequence numbers, SNOMED CT and LOINC answer codes.

The LOINC NBS panel also includes 12 card variables (e.g. state of origin, date of birth, time of birth, birthweight, and unique serial number of current sample), with individual answer lists for the

categorical variables (birth plurality, clinical events that affect newborn screen interpretation, hearing loss risk indicators, and mother's education). The full LOINC NBS panel is most easily reviewed by downloading the PDF from http://newbornscreening codes.nlm.nih.gov/nb/sc/constructingNBSHL7messa ges/. You can also obtain a spreadsheet version from the same web site.

The LOINC NBS panel can accommodate NBS results from all of the U.S. NBS programs. It specifies the codes for an NBS HL7 message. To show how these codes load into such a message, we created an annotated example HL7 v2.5.1 NBS message.[10] The example message includes segments for reporting NBS data including all of the card variables and summary reports, and some of the condition impressions and quantitative results. There are at least four potential destinations for newborn screening result messages: 1) the birth hospital, 2) the physician responsible for the infant's on-going care, 3) the state NBS and state EHDI programs and/or public health department, and 4) national and/or regional registries of NBS data. The message was designed to be used to send data to all such recipients with tailoring where needed, e.g. removing identifying data before sending to central registries.

The 50-plus NBS DBS programs are served by some 36 NBS laboratories, and there are only a few main commercial information system vendors, plus some internal state computer information departments. Because the numbers of involved organizations are limited, relative to other health information exchange contexts, rapid adoption of this standardized HRSA/NLM approach to NBS results messaging is possible. Indeed 15 months after the AHIC report to the HHS Secretary,[8] three major NBS lab system vendors (Natus/Neometrics, PerkinElmer and Oz Systems) can demonstrate early versions of HL7 messages that meet this specification, and at least one laboratory is already sending NBS HL7 messages (Figure 3).

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OBX|26|SN|31144-9^Thyroxine^LN||^21.3^^|ug/dL^
microgram/deciliter^UCUM|>5.0 ug/dL|||F |||200703311010

OBX|27|SN|54084-9^Galactose^LN||^1.9^^|mg/dL^
milligram/deciliter^UCUM|<14 mg/dL|||F |||200703310904

OBX|28|SN|33288-2^Galactose 1 phosphate uridyl transferase^LN||
^N^^|U/g{Hb}^units/gram Hb^UCUM|Enzyme Present|||F
|||200703310904

OBX|29|CE|54105-2^Hemoglobin Pattern^LN||LA11974-5^Hb F,A
(normal)^LN|||||F |||200704031215
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Figure 3. Excerpt from prototype Pennsylvania HL7 message, being developed by PerkinElmer and Oz Systems.

Discussion

The HRSA/NLM HL7 message guidance provides a uniform way to communicate newborn screening results in a computer-understandable form. As hospitals and office practice EHR systems adopt this guidance, they will solve some of the current problems with reporting NBS results. Most commercial EHRs already come equipped with HL7 inbound interfaces, and the Standards and Certification Criteria Interim Final Rule requires the support of LOINC and encourages the use of SNOMED CT in laboratory messages to meet the definition of meaningful use.[7] The Centers for Medicare and Medicaid Services is also considering expanding the Medicaid EHR incentive programs to include NBS documentation as a pediatric clinical quality measure.[11] If all U.S. NBS laboratories adopted the standard described here, EHRs could be designed to accept these messages out of the box, with no need to individually map and customize what would otherwise be large differences in NBS reporting formats, by state.

Some regional health information exchanges provide web-based report delivery systems that accept lab results messages from many sources (e.g. hospital laboratory, stand-alone radiology services) and deliver them in a uniform format to physician offices. Such systems, which provide another vehicle to deliver NBS results to care providers, already operate in Indiana, parts of Ohio, and Ontario, Canada (eCHN). Kentucky is developing a statewide health information exchange that will use HL7 messages to provide NBS results as its inaugural effort.

Having a standard message will also make it easier to collect regional and national data. Many of the conditions are extremely rare, with incidences of 1 in 100,000 births. Therefore, pooled data for all newborns screened are needed to study the effects of NBS follow-up programs and potential health interventions. These collections should contain quantitative results for negatives as well as positives. With such collections of quantitative data, researchers can improve screening methods and reduce false positive rates.

There have been differences of opinion in the NBS community about reporting numerical results as well as interpretations to pediatric care providers when screening tests are positive for a given condition. We support reporting numerical screening results whenever they can be reliably reported as the result of a standardized process. "Less than" or "greater than" results should only be reported when specific

results are outside of the analytical range of the measurement. In the case of tests that produce non-numeric results, such as hemoglobinopathy screening and DNA mutation analysis, the specific hemoglobin or mutation observed should be reported as opposed to a qualitative interpretation such as "positive." If cut-offs are obtained by evaluating percentiles rather than averages of analyte concentrations the limitations of that approach should be explained

Discussions with local pediatricians suggest that they tend to prefer qualitative reporting for negative NBS results because they are quick to read and digest. On the other hand, they prefer to get numerical values for the positives derived from quantitative measures, because the numerical values cue them to the likelihood that the positive is a true positive, needing close follow-up. Having the numerical results also makes it easier to discuss the results with the family.

Though challenges remain – including the unavailability of the follow-up physician's name at the time of initial screening and a lack of electronic and automated linkages to vital records (and other systems that could help assure that all infants are tested and receive appropriate follow up) – we are encouraged that standardized NBS messaging is being embraced so rapidly. This early success is testimony to the great cooperation among many organizations in the NBS community and their keen interest in the health of newborns.

Disclaimer

The findings and conclusions in this paper are those of the authors and do not necessarily represent the official position of the CDC, HRSA, NIH, NLM, or the Department of Health and Human Services.

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