HRSA/NLM Guidance for
Sending Electronic Newborn Screening Results with HL7 Messaging
Version 6

Contents

Key points

- This document is intended to supplement the HL7 version 2 implementation guide for electronic messaging, available at: http://www.hl7.org.
- This document uses codes from the June 2014 LOINC NBS panel 54089-8. Implementers should use the codes from the latest LOINC release, available at http://loinc.org.
- The example NBS result message covers all of the conditions that are on the Recommended Uniform Screening Panel (RUSP) as recommended by the Advisory Committee on Heritable Disorders in Newborns and Children ("the Committee") and adopted by the Secretary of Health and Human Services (HHS) as of June 2014. The LOINC newborn screening panel 54089-8 also covers additional conditions for which some US newborn screening programs elect to screen, such as lysosomal storage disorders. To get the latest information about the RUSP, please visit http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/.
- You do NOT need to use all of the LOINC codes related to newborn screening in every message, but rather just the subset that is relevant to the results you are sending.
- You do NOT have to follow the exact order of OBR/OBX segments presented in this document. For example, you could report the endocrine screening results followed by the metabolic results or the metabolic results followed by the endocrine results. Another option is to report all of the condition interpretations followed by all of the quantitative results.
- The annotated example HL7 message in this document omits some content to conserve space (as indicated with ellipses and notes). The full example NBS result message is available at http://newbornscreeningcodes.nlm.nih.gov/HL7.

Introduction

This example message was developed by the Lister Hill National Center for Biomedical Communications (LHNCBC), a research division of the U.S. National Library of Medicine, in conjunction with the Health Resources and Services Administration, to assist those implementing electronic messaging for newborn screening (NBS) results. This guide provides an overview of data and messaging standards for NBS as well as an example Health Level Seven (HL7) version 2.5.1 NBS result message in a machine-readable (HL7) format with detailed notes and explanations. It is not meant to be a technical HL7 implementation guide. LHNCBC, in collaboration with other agencies and

organizations, also created and maintains the Newborn Screening Coding and Terminology Guide: http://newbornscreeningcodes.nlm.nih.gov/, which provides mappings to additional coding terminologies, mappings between conditions and analytes, and other resources.

Our example message includes the clinical data collected on the dried blood spot (DBS) card, summary information about the NBS result and detailed information about specific test results. HRSA and NLM based this guidance on the work done by the American Health Information Community (AHIC) Personalized Healthcare Workgroup, discussions with the US Health Information Technology Standards Panel (HITSP) NBS work group and leaders from the NBS community. It was informed by a sample of de-identified NBS messages and reports from many states.

This HL7 guide is tightly tied to Logical Observations, Identifiers, Numbers and Codes (LOINC®) panel 54089-8 Newborn screening panel American Health Information Community, which includes LOINC codes for all observations that could be included in a NBS message, as well as for the nested relationships among the LOINC subpanels. For each LOINC code, the panel lists its data type, and if applicable according to its data type, associated coded answer list with LOINC answer (LA) or SNOMED CT (SCT) codes, standard units of measure for reporting quantitative information, and other attributes (See Figure 1). The panel includes variables to accommodate every analyte and every analyte ratio that we have identified as part of any U.S. jurisdiction's newborn screening program and includes separate subpanels for DBS screening results and point-of-care (POC) screening results. It also includes variables for reporting most of the DBS card variables (data elements recorded directly on the card that is used to collect the DBS specimen) and for reporting an interpretation of and narrative comments/discussions about the results for particular conditions or condition complexes.

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

PANEL HIERARCHY

LOINC#	LOINC Name	R/O/C	Cardinality	Data Type	Ex. UCUM Units
54089-8	Newborn screening panel American Health Information Community (AHIC)				
57128-1	Newborn Screening Report summary panel	C	0..1		
57721-3	Reason for lab test in Dried blood spot	R	1..1	CE	
57718-9	Sample quality of Dried blood spot	R	1..1	CE	
57130-7	Newborn screening report - overall interpretation	C	1..n	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 report for display or printing	O	0..1	FT	
57719-7	Conditions tested for in this newborn screening study [Identifier] in Dried blood spot	R	1..n	CE	
69969-4	Newborn screening report overall laboratory comment	O	0..n	FT	
57717-1	Newborn screen card data panel	C			
57716-3	State printed on filter paper card [Identifier] in NBS card	R		ST	
8339-4	Birthweight	R		NM	g
58229-6	Body weight Measured --when specimen taken	O		NM	g
57715-5	Time of birth	R		TM	{clock_time}
57722-1	Birth plurality of Pregnancy	R		CE	
57714-8	Obstetric estimation of gestational age	R		NM	wk
57713-0	Infant NICU factors that affect newborn screening interpretation	R	1..n	CE	
67703-9	Other infant NICU factors that affect newborn screening interpretation	C	0..n	TX	

Figure 1. Excerpt of LOINC 54089-8 (Newborn screening panel American Health Information Community (AHIC). The panel has a nested hierarchy of panels and subpanels and includes top-level panels for result summary, dried blood spot card data, lab results, and point of care results.

Emphasis on Structured and Comprehensive Data

We encourage NBS laboratories to report all quantitative results (and not just interpretations) to the appropriate NBS program, and to send at least the quantitative results that support positive and equivocal findings to the birth institution and attending clinicians. All of the variables included in the LOINC panel can be reported using HL7 OBR/OBX segments. Each NBS laboratory may choose the elements it needs from the comprehensive set of variables in the LOINC panel to report the specific tests it performs. We discourage the use of NTE segments (general unstructured notes) anywhere in the message so the data in the message is computer-readable.

Brief overview of major HL7 segments and grammar

HL7 version 2.x messages consist of segments that are represented as ASCII text with data fields and subfields separated by specific delimiters.

- Segments always begin with a 3-character designation (e.g., MSH, PID, NK1, OBR, OBX) that indicate segment type (segment ID)
- Vertical bars or pipes (|) separate the segment ID from the first data field and also separate adjacent data fields within each segment
- Hats (^) separate subfields
- Ampersands (&) separate subfield components
- Tildes (~) separate repeating values within a field
- Segments always end with a carriage return character (not a vertical bar), which is sometimes indicated as <CR>

Common HL7 segment types:

- Message header (MSH): sender and intended recipient(s) of the electronic message and associated information, including unique identifier(s)
- Patient identification (PID): patient demographics, including name, address, date of birth, and unique identifiers such as medical record number or Social Security Number
- Next of kin (NK1): information about the patient's next of kin, including name, address and phone number(s). In the context of NBS, we have included the infant's mother as the infant's next of kin.
- Common order (ORC): general order information including order number and ordering hospital or provider along with address(es) and unique identifier(s)
- Observation request (OBR): information about the specific test or panel, including the test name and test code, whose results are being reported in the OBX segment(s) immediately following that OBR. A single OBR segment can be followed by one or more OBX segments. For example, an OBR segment containing a hemoglobin order will be followed by a single OBX segment with the hemoglobin result, and an OBR segment containing a complete blood count order will be followed by multiple OBX segments containing the individual results for hemoglobin, hematocrit, white blood cell count, etc.
- Observation result (OBX): the result for the specific test indicated in the preceding OBR segment and associated information such as reference range, units of measure, out of range flag, and result status (e.g., preliminary, final). See details in the next section of the document.

Anatomy of an OBX segment

Because the focus of this guide is NBS results reported using a series of OBX segments, in this section we included a more detailed description of OBX fields and syntax.

Example OBX segment:

```
OBX|4|ST|53160-8^Propionylcarnitine (C3)^LN^3403^C3^L|1|5.17|umol/L|4.62-
5.50|N|||F|||20090714074205
```

Segment identifier and field numbering:

- The first 3 letters (in this case, "OBX") identify the segment ID.
- The first vertical bar (|) separates the segment ID from the first field; the remaining vertical bars separate adjacent fields. Thus, the first field (OBX-1) begins after the first vertical bar, the second field (OBX-2) follows immediately after the second vertical bar, and so on.
- If a field is not populated, the surrounding vertical bars will serve as placeholders so that the subsequent fields can be counted appropriately. For example, in both of the following fictional OBX segments, the value 'd' is contained in OBX-5:

```
OBX|1|a|b|c|d|e|f
OBX|2|a|||d|e|f
```

Field definitions:

Each field is defined to contain a specific piece of information. For example, in any OBX segment, OBX-5 will always contain the result and OBX-7 the reference range. Computers are able to interpret HL7 messages because they know that a given field is supposed to contain a specific type of information.

Not all of the fields in an OBX segments are relevant for reporting NBS results, such as OBX-9, 10, 12 and 13. We only describe the relevant OBX fields below; in our examples, the fields that are not relevant are not populated.

OBX-1: sequence number, which distinguishes consecutive OBX segments contained under a single OBR segment. These are simple counts that re-start at 1 after each OBR segment.

OBX-2: data type of the test result (e.g., ST = string, NM = numeric, CE = coded entry).

OBX-3: observation ID, including code, variable name and code system (e.g., "LN" for LOINC, "L" for local"), which represents the variable or "question" such as hemoglobin. HL7 permits senders to include two sets of codes and names in this field so that both the standard LOINC code and the local code can be reported for a given observation. Within the OBX-3 fields in our example (above) we display the standard code in **red** and the local code in **turquoise** as follows: `LOINC code^LOINC variable name^LN^Local code^Local variable name^L`.

OBX-4: sub-ID that is used to distinguish multiple OBX's with the same Observation ID in OBX-3. The value of the sub-ID should increment by one for each OBX with the same observation ID.

OBX-5: observation value or test result/impression. This is the value of the variable identified in OBX-3 or the "answer" to the question asked in OBX-3 and is displayed in **green** in the example OBX above. Observation values can have different data types (OBX-2 contains the data type of the result reported in OBX-5):

- Numeric (OBX-2 = NM): such as hemoglobin or phenylalanine level.
- Coded (OBX-2 = CE): coded values from a predefined list of answers such as eye color or NBS conditions with positive markers. Similar to OBX-3, OBX-5 can include two coded answers, one from a standard code set, such as SCT or LOINC, and one from a local code set. When available, the coded answer should be reported using a SCT code. If an SCT code is not available, a LOINC answer (LA) code should be used.

- Narrative text (OBX-2 = ST): such as comments or recommendations for follow-up.

OBX-6: standard units of measure, displayed in **blue** within the example OBX above. These should be represented as Unified Code for Units of Measure (UCUM) units as shown in the LOINC panel. Most numeric results will have true units of measure listed in OBX-6, but some that do not have a true unit of measure, such as ratios, can be indicated in UCUM with the text string {Ratio}.

OBX-7: reference range, displayed in **pink** above. These are strings and, if they contain units of measure, these units should match those in OBX-6.

OBX-8: normal/abnormal flags. N for normal, A for abnormal (when the observation is a code), H for high, L for low, AA for critically abnormal, HH for critically high and LL for critically low. These are derived from HL7 table 0078 Abnormal Flags.

OBX-11: observation result status. These are derived from HL7 table 0085. For example, F = Final result, I = Specimen in lab, results pending, C = Corrected result that replaces a prior final result, and P = Preliminary results.

OBX-14: date/time of the observation. It is not necessary to include the date/time of the observation in each OBX segment since the receiving application will use the value in OBR-7 for all OBX segments included under that test or panel.

Note on Date/Time:

There are three different date/time values that are important when reporting newborn screening results. OBR-7 contains the observation date/time, which is the time when the specimen was collected. OBR-14 contains the specimen received date/time, when the specimen was received in the laboratory. Since newborn screening dried blood spots are sent to a laboratory outside of the hospital, this value is useful. OBR-22 contains the report or status change date/time, which indicates when the results were reported or the status (OBX-11) was updated. All date/time values should be reported to the nearest hour and minute since the baby's age in hours at the time of specimen collection is important for results interpretation. Some hospital information systems only record the patient's date of birth and not the time of birth, which is why the time of birth should be sent in a separate OBX segment.

Annotated Example HL7 message

We invented the result values and normal ranges you see in this example and they are not clinically correct. These formats are similar to what you would see in real NBS messages, but a NBS lab would include its real results and local reference ranges according to its usual practices.

A full example newborn screening HL7 message, with unformatted text, is available at: http://newbornscreeningcodes.nlm.nih.gov/HL7. In the annotated version below, to conserve space, we have excerpted some content as indicated by ellipses (…).

While it is not required, the HL7 specification allows the user to send two codes in every coded value field: a primary code and an alternate code. We recommend including the local laboratory code in addition to the primary code for identifying the variable. Depending on the variable, either LOINC or SNOMED CT, represented by "LN" and "SCT", respectively, will be the primary code system. In cases where both LOINC answer and SCT codes are available for the same result, SCT is the preferred primary code system. The local code is identified by an "L" in the alternate coding system field.

Throughout this annotated example message, explanations usually precede the segment(s) they refer to and often cover several segments that follow. For formatting purposes and to improve readability, we have inserted line breaks in some places before and after the hat (^) and vertical bar (|) symbols in the message, but the real message would not have such line breaks. Also, we have broken the message into the following sections for instructive purposes, but a real message would simply consist of a series of HL7 segments without section breaks:

1. Administrative segments
2. NBS report summary
3. Dried blood spot card data
4. Dried blood spot screening results
5. Point-of-care screening results

This document has LOINC codes that are current as of the June 2014 release. Implementers should use the codes from the latest LOINC release, available from http://loinc.org.

Section 1: Administrative segments

Administrative segments, sometimes called header segments, appear at the beginning of the message. They contain essential demographic and message control data used to process the message.

The MSH (Message Header) segment defines the message source, purpose and destination. The sending laboratory is identified by a CLIA number and the receiving hospital or practice by an NPI number.

```
MSH|^~\&|PHLIMS^3.11.333.1.333333.1.333^ISO|TNSPHLAB^77D7777777^CLIA|EHRSYSTEM|ST ELSEWHERE
HOSPITAL^9999999999^NPI|20101014210405-0400||ORU^R01^ORU_R01|123|P|2.5.1
```

The PID (Patient Identification) segment refers to the baby, but some of the data, such as address and phone number, may come from the mother's record.

```
PID|1||123456789^^^ST ELSEWHERE HOSPITAL&9999999999&NPI^MR||Lane^Jane^Mary^^^^L~Smith^Baby
Girl^^^^^A|Smith|20101013|F||2106-3^white^HL70005|123 Main Street^Apartment 3-
C^Anytown^TN^55555^USA^^^333|333|^^^^^865^5551212||||||||N^Not Hispanic or
Latino^HL70189||Y|1|||||N
```

The NK1 (Next of Kin) segment typically contains data about the mother, but in some circumstances, such as adoption, it may contain data about a different caregiver. Additional NK1 segments can be added to carry data about the father or other caregivers.

```
NK1|1|Lane^Lois^^^^^L|MTH^Mother^HL70063|123 Main Street^Apartment 3-
C^Anytown^TN^55555^USA^^^333|^^^^^865^5551212||||||||||19850710|||||||||||||||123121234^^^SS
A&2.16.840.1.113883.4.1&ISO^SS~222222222A2^^^TN^MA
```

The ORC (Common Order) segment is used to send information that is universal to all orders, such as the order number, the person entering the order, and the ordering provider. In this example ORC segment, the hospital that created the placer order number and the ordering provider are identified by their NPI numbers, and the laboratory by its CLIA number.

```
ORC|RE|128993^ST ELSEWHERE
HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|||||||||1111111111^Smiles^Minnie^^^Dr^^
^NPI&2.16.840.1.113883.4.6&ISO^L^^^NPI^^^^^^^^MD|||||||||ST ELSEWHERE
HOSPITAL^^^^^NPI&2.16.840.1.113883.4.6&ISO^NPI^^^9999999999|211 Small
Street^^Anytown^TN^55555^USA^^^333|^^^^^865^4442222||||||I
```

Section 2: NBS report summary

The report summary panel is required and at a minimum, should include the required OBX segments for reason for test, specimen quality, conditions tested, conditions with positive markers, and conditions with equivocal markers. The narrative summary segments are optional; however, they are recommended to help generate a clinical display.

The first OBR segment in the message should contain LOINC code 54089-8 for the overarching NBS panel as a way to tie together all of the nested panels (sub-panels) that follow. In general, OBR segments are followed by OBX segments with result data; however, in the case of nested panels such as the NBS panel, the top OBR segment can be followed by another OBR segment containing the secondary panel LOINC code, which is then followed by one or more OBX segments.

```
OBR|1|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54089-
8^Newborn screening panel American Health Information Community
(AHIC)^LN|||201010141853|||^VH||||201010140920||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.
113883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBR|2|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57128-
1^Newborn Screening Report summary
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|57721-3^Reason for lab test in Dried blood spot^LN||LA12421-6^Initial
screen^LN|||N|||F

OBX|2|CE|57718-9^Sample quality of Dried blood spot^LN||LA12432-3^Acceptable^LN|||N|||F

OBX|3|CE|57130-7^Newborn screening report - overall interpretation^LN||LA12431-5^Out of range
requiring immediate second-tier testing for at least one condition^LN|||A|||F
```

The required information about the conditions with positive and equivocal markers in this newborn screening study is reported using separate OBX segments for each positive and/or equivocal condition using the LOINC answer list LL835-0. The examples below use LA and SCT codes as the two coded answers in OBX-5. However, senders can send one primary code (SCT is preferred) and their local code as the alternate code.

```
OBX|4|CE|57131-5^Newborn conditions with positive markers [Identifier] in Dried blood
spot^LN|1|LA12509-8^MCAD^LN^128596003^Medium-chain acyl-coenzyme A dehydrogenase
deficiency^SCT|||A|||F

OBX|5|CE|57131-5^Newborn conditions with positive markers [Identifier] in Dried blood
spot^LN|2|LA16207-5^Hemoglobinopathies^LN^80141007^Hemoglobinopathy^SCT|||A|||F

OBX|6|CE|57720-5^Newborn conditions with equivocal markers [Identifier] in Dried blood
spot^LN||LA12532-0^BIO^LN^8808004^Biotinidase deficiency^SCT|||A|||F
```

The escape sequence "\.br\" indicates a line break in an HL7 formatted text field (data type FT) as specified in the HL7 v2.5.1 specification chapter 2. Other escape sequences can specify indents or ASCII characters. One method for embedding a PDF file for the printable report within an HL7 message is to send it as binary data.

```
OBX|7|FT|57724-7^Newborn screening short narrative summary^LN||SUMMARY: Newborn Metabolic
Screen REQUIRES FOLLOW UP\.br\Sample Quality: Acceptable\.br\Amino Acids, In range\.br\Fatty
acids, ABNORMAL MCAD SCREEN\.br\Organic acids, In range\.br\TSH (CH), In range\.br\17-OH-
Progesterone (CAH), No evidence of CAH\.br\Biotinidase, BORDERLINE BIOT SCREEN\.br\IRT (Cystic
Fibrosis), No evidence of cystic fibrosis\.br\Hemoglobins, ABNORMAL HGB SCREEN due to a type of
hemoglobin that could not be identified\.br\|||N|||F

OBX|8|FT|57129-9^Full newborn screening summary report for display or printing^LN||NEWBORN
METABOLIC SCREEN\.br\Patient's Name: Babygirl Lane Twin A\.br\Date of birth: 13 Oct 2010, Time
of birth: 06:32 am\.br\Sex: Female\.br\Age at collection: 30 hours\.br\Mother's name: Lois
Lane\.br\Accession number: 200902, Collected: 14 Oct 2010, Received: 15 Oct 2010\.br\Ordering
physician: Dr. Minnie Smiles\.br\SUMMARY: Newborn Metabolic Screen REQUIRES FOLLOW
UP\.br\Sample Quality: Acceptable\.br\Amino Acids, In range\.br\Fatty acids, ABNORMAL MCAD
SCREEN\.br\Screen positive for medium chain acyl-CoA dehydrogenase deficiency (MCAD). Immediate
clinical follow-up and contact with metabolic specialist indicated. Result phoned to Dr. Bob
Healthy (865) 444-2222 2010-10-16, 2:34 pm, by Nurse Nancy. C8 = 19.71 umol/L (< 0.25 umol/L),
C6 = 2.81 umol/L (< 0.25 umol/L), C10:1 = 0.71 umol/L (< 0.20 umol/L), C8/C10 = 11.324 (<
4.000), C8/C2 = 0.813 (< 0.050).\.br\...\.br\(full example report not included in this message
for brevity)\.br\|||A|||F
```

The required information about the conditions tested for in this newborn screening study is reported using separate OBX segments for each condition using the LOINC answer list LL841-8. Because each of the OBX segments below includes the same LOINC code in OBX-3 (observation ID), OBX-4 (observation sub-ID) is utilized to help distinguish between them. The value of the sub-ID in OBX-4 should increment by one for each OBX with the same observation ID.

Following are example segments for conditions on the RUSP as of June 2014 (note the sub-IDs 1 through 32 in OBX-4):

```
OBX|9|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|1|LA12463-8^HEAR^LN^15188001^Hearing loss^SCT|||N|||F

OBX|10|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|2|LA20349-9^CCHD^LN|||N|||F

OBX|11|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|3|LA12466-1^3-MCC^LN^13144005^Methylcrotonyl-CoA carboxylase
deficiency^SCT|||N|||F

OBX|12|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|4|LA12471-1^ASA^LN^41013004^Argininosuccinate lyase deficiency^SCT|||N|||F
```

```
OBX|13|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|5|LA12474-5^BKT^LN^237953006^Mitochondrial 2-methylacetoacetyl-CoA thiolase
deficiency - potassium stimulated^SCT|||N|||F
```

…

Note: OBX segments with sub-IDs 6-25 omitted here, but included in the Example Message document, available at: http://newbornscreeningcodes.nlm.nih.gov/HL7

…

```
OBX|34|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|26|LA12537-9^CF^LN^190905008^Cystic fibrosis^SCT|||N|||F

OBX|35|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|27|LA12538-7^CH^LN^190268003^Congenital hypothyroidism^SCT|||N|||F

OBX|36|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|28|LA12543-7^GALT^LN^398664009^Deficiency of UTP-hexose-1-phosphate
uridylyltransferase^SCT|||N|||F

OBX|37|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|29|LA12614-6^Hb SS-disease (sickle cell anemia)^LN^127040003^Hereditary
hemoglobinopathy disorder homozygous for hemoglobin S^SCT|||N|||F

OBX|38|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|30|LA12616-1^Hb SC-disease^LN^35434009^Sickle cell-hemoglobin C
disease^SCT|||N|||F

OBX|39|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|31|LA12615-3^Hb S beta-thalassemia^LN^127041004^Sickle cell-beta-
thalassemia^SCT|||N|||F

OBX|40|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|32|LA12566-8^SCID^LN^31323000^Severe combined immunodeficiency
disease^SCT|||N|||F
```

Section 3: Dried blood spot (DBS) card data

The DBS card variables contain demographics and clinical data from the filter paper order form that is used both for collecting the specimen and gathering the data necessary to send the order request to the laboratory. Each NBS program should only use the codes it needs to send the Ask at Order Entry information it is required to send by law, policy or practice. Some information is entered in the administrative segments as described above, and the remaining variables are reported using OBX segments nested under an OBR segment for the card data panel. Many of the variables in both the administrative and OBX segments require selection from a fixed list of choices or answers. For the card variable data reported in administrative segments (e.g. race, ethnicity), some of the answer lists are predefined by HL7, and for the variables reported in OBX segments (e.g. birth plurality, clinical events that affect NBS interpretation), LA code(s) from the appropriate LOINC answer list should be used in OBX-5. The required or optional status of some of variables may vary by state.

```
OBR|3|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57717-
1^Newborn screen card data panel^LN|||201010141853|||^VH||||201101040920||1111111111
^Smiles^Minnie^^^Dr ^^^NPI&2.16.840.1.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|ST|57716-3^State printed on filter paper card [Identifier] in NBS card^LN||TN|||N|||F

OBX|2|ST|57723-9^Unique bar code number of Current sample^LN||97893203|||N|||F
```

```
OBX|3|CE|57721-3^Reason for lab test in Dried blood spot^LN||LA12426-5^Subsequent screen –
required by protocol^LN||||||O

OBX|4|ST|57711-4^Unique bar code number of Initial sample^LN||43554432|||N|||F

OBX|5|CE|57722-1^Birth plurality of Pregnancy^LN||LA12412-5^Twins^LN|||N|||F

OBX|6|TM|57715-5^Birth time^LN||0632-0500|||N|||F

OBX|7|NM|57714-8^Obstetric estimation of gestational age^LN||37|wk^weeks||N|||F

OBX|8|NM|8339-4^Birthweight^LN||2920|g||N|||F

OBX|9|NM|58229-6^Body weight Measured --when specimen taken^LN||2750|g^gram|||||F

OBX|10|TX|62323-1^Post-discharge provider ID [Identifier]^LN||4444444444||||||F

OBX|11|TX|62324-9^Post-discharge provider name in Provider^LN||Dr Bob Healthy||||||F

OBX|12|TX|62325-6^Post-discharge provider practice ID^LN||5555555555||||||F

OBX|13|TX|62326-4^Post-discharge provider practice name^LN||Healthy Clinic||||||F

OBX|14|TX|62327-2^Post-discharge provider practice address^LN||100 Small Street, Suite 3B,
Anytown, Tennessee 55555||||||F

OBX|15|TN|62328-0^Post-discharge provider practice telephone number in Provider^LN||(865) 542-
3333||||||F

OBX|16|TX|62329-8^Birth hospital facility ID [Identifier] in Facility^LN||9999999999||||||F

OBX|17|TX|62330-6^Birth hospital facility name^LN||ST ELSEWHERE HOSPITAL||||||F

OBX|18|TX|62331-4^Birth hospital facility address^LN||211 Small Street, Anytown, Tennessee
55555||||||F

OBX|19|TN|62332-2^Birth hospital facility phone number in Facility^LN||(865) 444-2222|||N|||F

OBX|20|CE|67704-7^Feeding types^LN|1|LA14041-0^Lactose free formula (including soy or
hydrolyzed)^LN||||||F

OBX|21|CE|67704-7^Feeding types^LN|2|LA16914-6^Breast milk^LN||||||F

OBX|22|CE|57713-0^Infant NICU factors that affect newborn screening
interpretation^LN|1|LA12419-0^Infant in ICU at time of specimen collection^LN||||||F

OBX|23|CE|57713-0^Infant NICU factors that affect newborn screening
interpretation^LN|2|LA12417-4^Any blood product transfusion (including ECMO)^LN||||||F

OBX|24|DTM|62317-3^Date of Last Blood Product Transfusion^LN||201010131723||||||F

OBX|25|CE|67706-2^Maternal factors that affect newborn screening interpretation^LN||LA46-
8^Other^LN||||||F

OBX|26|TX|67707-0^Other maternal factors that affect newborn screening
interpretation^LN||Mother has Lupus||||||F
```

Section 4: Newborn screening DBS results

We recommend that state laboratories report quantitative results for at least all of the screen positive or equivocal conditions to all report receivers, and we propose that state laboratories report all quantitative and qualitative results to the state newborn screening program.

Nested OBR segments can be used to identify the various sub-panels within the results. This helps to organize the data on clinical reports and facilitates sending partial results. The use of multiple nested OBR segments is optional, but encouraged.

The values below are for illustration only and are not be clinically valid. In some cases where typical values were not available, the example message contains "99" for numeric values with a reference range of "<999," and "XXXX" for string values. There is no need to send OBX segments without a value and you should omit segments with LOINC codes for results that are not measured in your laboratory.

```
OBR|4|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57794-
0^Newborn screening test results panel - Dried blood
spot^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.11
3883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F
```

Section 4.1: Amino Acid Panel

The amino acid panel is the first of many subpanels that follow a similar pattern: one segment for a general coded interpretation (In range, Borderline, Out of range requiring immediate second tier testing for at least one condition, etc.), a second segment to identify the specific disorder suspected, a third segment for narrative comment/discussion, and then a series of segments for the relevant quantitative measurements.

```
OBR|5|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|53261-4^Amino
acid newborn screen
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46733-2^Amino acidemias newborn screen interpretation^LN||LA18592-8^In
range^LN|||N|||F

OBX|2|TX|57710-6^Amino acidemias newborn screening comment-discussion^LN||Any baby with
clinical features suggestive of a metabolic disorder requires clinical and diagnostic follow-up
regardless of whether the NBS result is normal or abnormal.|||N|||F

OBX|3|CE|57793-2^Amino acidemia disorder suspected [Identifier] in Dried blood spot^LN||LA137-
2^None^LN|||N|||F
```

For historical reasons, a few states have legislative mandates to report two specific conditions (Phenylketonuria (PKU) and Maple Syrup Urine Disease (MSUD)), separately and explicitly, instead of using the general purpose approach that includes all amino acid conditions. We have created individual LOINC codes for interpretation and for comment/discussion for these two conditions for the states that need them and illustrate their use below; others can omit these condition specific segments.

```
OBX|4|CE|46746-4^Phenylketonuria and variants/Biopterin defects newborn screen
interpretation^LN||LA18592-8^In range^LN|||N|||F

OBX|5|TX|58231-2^Phenylketonuria and variants/Biopterin defects newborn screening comment-
discussion^LN||Any baby with clinical features suggestive of a metabolic disorder requires
clinical and diagnostic follow-up regardless of whether the NBS result is normal or
abnormal.|||N|||F
```

```
OBX|6|CE|46743-1^Maple syrup urine disease newborn screen interpretation^LN||LA18592-8^In
range^LN|||N|||F

OBX|7|TX|58230-4^Maple syrup urine disease newborn screening comment-discussion^LN||Any baby
with clinical features suggestive of a metabolic disorder requires clinical and diagnostic
follow-up regardless of whether the NBS result is normal or abnormal.|||N|||F

OBX|8|NM|47539-2^3-Methylhistidine [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|9|NM|53232-5^5-Oxoproline+Pipecolate [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F
```

Some of the quantitative results that laboratories report are computed ratios of several amino acids. Because ratios of two measurements with the same units do not have units themselves, we recommend using the string {Ratio} per Uniform Code for Units of Measure (UCUM) guidelines so that all quantitative measurements have units regardless of whether they are computed or measured and to help users identify the computed values.

```
OBX|10|NM|53394-3^5-Oxoproline+Pipecolate/Phenylalanine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|11|NM|53150-9^Alanine+Beta Alanine+Sarcosine [Moles/volume] in Dried blood
spot^LN||1236.06|umol/L|<1500|N|||F

OBX|12|NM|53393-
5^Alloisoleucine+Isoleucine+Leucine+Hydroxyproline+Valine/Phenylalanine+Tyrosine [Molar ratio]
in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|13|NM|53152-5^Alloisoleucine+Isoleucine+Leucine+Hydroxyproline [Moles/volume] in Dried
blood spot^LN||99|umol/L|<999|N|||F

OBX|14|NM|53153-3^Alloisoleucine+Isoleucine+Leucine+Hydroxyproline/Phenylalanine [Molar ratio]
in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|15|NM|53154-1^Alloisoleucine+Isoleucine+Leucine+Hydroxyproline/Alanine [Molar ratio] in
Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|16|NM|47562-4^Arginine [Moles/volume] in Dried blood spot^LN||5.89|umol/L|<90|N|||F

OBX|17|NM|53398-4^Arginine/Phenylalanine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|18|NM|53062-6^Argininosuccinate [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|19|NM|53200-2^Argininosuccinate/Arginine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|20|NM|53155-8^Asparagine+Ornithine [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|21|NM|53395-0^Asparagine+Ornithine/Serine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|22|NM|53396-8^Asparagine+Ornithine/Phenylalanine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|23|NM|47573-1^Aspartate [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|24|NM|42892-0^Citrulline [Moles/volume] in Dried blood spot^LN||19.4|umol/L|<55|N|||F
```

```
OBX|25|NM|54092-2^Citrulline/Arginine [Molar ratio] in Dried blood spot^LN||5.63|{Ratio}|5.1-
6.0|N|||F

OBX|26|NM|53157-4^Citrulline/Phenylalanine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|27|NM|53399-2^Citrulline/Tyrosine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|28|NM|47623-4^Glutamate [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|29|NM|47633-3^Glycine [Moles/volume] in Dried blood spot^LN||528|umol/L|<999|N|||N

OBX|30|NM|47643-2^Histidine [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|31|NM|53158-2^Homocitrulline [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|32|NM|47689-5^Lysine [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|33|NM|47700-0^Methionine [Moles/volume] in Dried blood spot^LN||45.97|umol/L|44-49|N|||F

OBX|34|NM|53397-6^Methionine/Alloisoleucine+Isoleucine+Leucine+Hydroxyproline [Molar ratio] in
Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|35|NM|53156-6^Methionine/Phenylalanine [Molar ratio] in Dried blood
spot^LN||0.82|{Ratio}|0.76-1.0|N|||F

OBX|36|NM|29573-3^Phenylalanine [Moles/volume] in Dried blood spot^LN||104.61|umol/L|99-
135|N|||F

OBX|37|NM|35572-7^Phenylalanine/Tyrosine [Molar ratio] in Dried blood
spot^LN||2.46|{Ratio}|1.64-2.50|N|||F

OBX|38|NM|47732-3^Proline [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|39|NM|53392-7^Proline/Phenylalanine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|40|NM|47742-2^Serine [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|41|NM|53231-7^Succinylacetone [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|42|NM|47784-4^Threonine [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|43|NM|53159-0^Tryptophan [Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|44|NM|35571-9^Tyrosine [Moles/volume] in Dried blood spot^LN||281.53|umol/L|205-223|H|||F

OBX|45|NM|47799-2^Valine [Moles/volume] in Dried blood spot^LN||76|umol/L|<250|N|||F

OBX|46|NM|53151-7^Valine/Phenylalanine [Molar ratio] in Dried blood
spot^LN||1.44|{Ratio}|<4.0|N|||F
```

Section 4.2: Acylcarnitine Panel

The Acylcarnitine panel follows a very similar pattern to the amino acid panel and includes many ratios and computed values as well as a long list of qualitative measures generated by tandem mass spectrometry.

The Acylcarnitine panel is different from other panels because it contains two separate subpanels for two groups of disorders (fatty acid oxidation and organic acid) that have overlapping analytes. Each subpanel includes all of the relevant analytes for that disorder group, so there is some duplication of analytes in the two subpanels. States can choose whether to report their results under the single Acylcarnitine panel or use the two separate fatty acid oxidation and organic acid panels with some duplication of results. We illustrate use of the two subpanels below.

```
OBR|6|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|58092-
8^Acylcarnitine newborn screen
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|58088-6^Acylcarnitine newborn screen interpretation^LN||LA12431-5^Out of range
requiring immediate second-tier testing for at least one condition^LN|||A|||F

OBX|2|TX|58093-6^Acylcarnitine newborn screening comment-discussion^LN||ABNORMAL MCAD SCREEN.
Screen positive for medium chain acyl-CoA dehydrogenase deficiency (MCAD). Immediate clinical
follow-up and contact with metabolic specialist indicated. Result phoned to (XXX) XXX-XXXX;
YYYY-MM-DD, HHMMh, by NAME.|||A|||F

OBR|7|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57084-6^Fatty
acid oxidation newborn screen
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F
```

The following three segments illustrate how to use the related codes within a subpanel for reporting an out of range result using the disorder medium chain acyl-CoA dehydrogenase deficiency (MCAD) as an example.

```
OBX|1|CE|46736-5^Fatty acid oxidation defects newborn screen interpretation^LN|1|LA12431-5^Out
of range requiring immediate second-tier testing for at least one condition^LN|||A|||F

OBX|2|CE|57792-4^Fatty acid oxidation conditions suspected [Identifier] in Dried blood
spot^LN||LA12509-8^MCAD^LN^128596003^Medium-chain acyl-coenzyme A dehydrogenase
deficiency^SCT|||A|||F

OBX|3|TX|57709-8^Fatty acid oxidation defects newborn screening comment-discussion^LN||ABNORMAL
MCAD SCREEN. Screen positive for medium chain acyl-CoA dehydrogenase deficiency (MCAD).
Immediate clinical follow-up and contact with metabolic specialist indicated. Result phoned to
(XXX) XXX-XXXX; YYYY-MM-DD, HHMMh, by NAME.|||A|||F

OBX|4|NM|38481-8^Carnitine free (C0) [Moles/volume] in Dried blood spot^LN||11.88|umol/L|7.50-
12.00|N|||F

OBX|5|NM|53233-3^Carnitine free (C0)/Palmitoylcarnitine (C16) [Molar ratio] in Dried blood
spot^LN||67.04|{Ratio}|<999|N|||F
```

…

Note: OBX segments with IDs 6-54 omitted here to conserve space, but included in the Example Message document, available at: http://newbornscreeningcodes.nlm.nih.gov/HL7

…

```
OBX|55|NM|53241-6^Stearoylcarnitine (C18) [Moles/volume] in Dried blood
spot^LN||0.26|umol/L|<0.31|N|||F

OBX|56|NM|53400-8^Stearoylcarnitine (C18)/Propionylcarnitine (C3) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F
```

```
OBX|57|NM|50109-8^3-Hydroxylinoleoylcarnitine (C18:2-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|58|NM|50113-0^3-Hydroxyoleoylcarnitine (C18:1-OH) [Moles/volume] in Dried blood
spot^LN||0.09|umol/L|0.08-0.10|N|||F

OBX|59|NM|50132-0^3-Hydroxystearoylcarnitine (C18-OH) [Moles/volume] in Dried blood
spot^LN||0.08|umol/L|0.07-0.10|N|||F

OBR|8|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57085-
3^Organic acid newborn screen
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46744-9^Organic acidemias newborn screen interpretation^LN||LA18592-8^In
range^LN|||N|||F

OBX|2|CE|57791-6^Organic acidemia conditions suspected [Identifier] in Dried blood
spot^LN||LA137-2^None^LN|||N|||F

OBX|3|TX|57708-0^Organic acidemias defects newborn screening comment-discussion^LN||Any baby
with clinical features suggestive of a metabolic disorder requires clinical and diagnostic
follow-up regardless of whether the NBS result is normal or abnormal.|||N|||F

OBX|4|NM|50157-7^Acetylcarnitine (C2) [Moles/volume] in Dried blood
spot^LN||31.78|umol/L|<999|N|||F

OBX|5|NM|53237-4^Acrylylcarnitine (C3:1) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||N
```

…

Note: OBX segments with IDs 6-35 omitted here to conserve space, but included in the Example Message document, available at: http://newbornscreeningcodes.nlm.nih.gov/HL7

…

```
OBX|36|NM|53187-1^Methylglutarylcarnitine (C6-DC) [Moles/volume] in Dried blood
spot^LN||0.11|umol/L|0.10-0.12|N|||F

OBX|37|NM|53165-7^Formiminoglutamate [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|38|NM|67708-8^Malonylcarnitine (C3-DC)+3-Hydroxybutyrylcarnitine (C4-OH) [Moles/volume] in
Dried blood spot^LN||0.26|umol/L|<999|N|||F

OBX|39|NM|67709-6^Methylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH)
[Moles/volume] in Dried blood spot^LN||3.16|umol/L|<999|N|||F

OBX|40|NM|67710-4^Glutarylcarnitine (C5-DC)+3-Hydroxyhexanoylcarnitine (C6-OH) [Moles/volume]
in Dried blood spot^LN||99|umol/L|<999|N|||F
```

Section 4.3: Cystic fibrosis panel

The cystic fibrosis panel contains the usual coded interpretation and comment/discussion codes. There is no need for a conditions suspected code as there is only one condition in this panel. The cystic fibrosis panel is different from other panels in that it typically uses second tier genetic testing for CFTR gene mutations as part of the initial screen when the trypsinogen result is abnormal, which reduces false positives. For purposes of newborn screening, it is not typical to report the details of the gene testing (e.g. the specific mutation) and hence the code 54083-1 for

CFTR gene mutations is a string data type. Further discussions are underway as to whether and how to report the full confirmatory gene testing results as part of a report that normally conveys screening results. At this time, there are no established standards or answer codes for the data that is reported using code 54083-1.

```
OBR|9|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54078-
1^Cystic fibrosis newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46769-6^Cystic fibrosis newborn screen interpretation^LN||LA18592-8^In
range^LN|||N|||F

OBX|2|TX|57707-2^Cystic fibrosis newborn screening comment-discussion^LN||No evidence of cystic
fibrosis. CF mutation analysis not performed. Further testing is only required if there is
clinical suspicion of cystic fibrosis. Symptoms include poor growth, loose stools or evidence
of malabsorption, persistent cough, or respiratory concerns.|||N|||F

OBX|3|NM|2077-6^Chloride [Moles/volume] in Sweat^LN||99|mmol/L|<999|N|||F

OBX|4|NM|48633-2^Trypsinogen I Free [Mass/volume] in Dried blood spot^LN||99|ug/L|<999|N|||F

OBX|5|TX|54083-1^CFTR gene mutations found [Identifier] in Dried blood spot
Nominal^LN||None|||N|||F
```

Section 4.4: Endocrine panel

The endocrine panel is currently used to report the results of two conditions, congenital adrenal hyperplasia (CAH) and congenital hypothyroidism (CH). States may choose to report them together under the endocrine panel or separately in their own subpanels.

```
OBR|10|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54076-
5^Endocrine newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBR|11|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57086-
1^Congenital adrenal hyperplasia (CAH) newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46758-9^Congenital adrenal hyperplasia newborn screen interpretation^LN||LA18592-8^In
range^LN|||N|||F

OBX|2|NM|53347-1^11-Deoxycorticosterone [Mass/volume] in Dried blood
spot^LN||99|ng/dL|<999|N|||F

OBX|3|NM|53338-0^11-Deoxycortisol [Mass/volume] in Dried blood spot^LN||99|ug/dL|<999|N|||F

OBX|4|NM|38473-5^17-Hydroxyprogesterone [Mass/volume] in Dried blood
spot^LN||182|ng/mL|<190|N|||F

OBX|5|NM|53336-4^17-Hydroxyprogesterone+Androstenedione/Cortisol [Mass Ratio] in Dried blood
spot^LN||99|{ratio}|<999|N|||F

OBX|6|NM|53341-4^21-Deoxycortisol [Mass/volume] in Dried blood spot^LN||99|ug/dL|<999|N|||F

OBX|7|NM|53343-0^Androstenedione [Mass/volume] in Dried blood spot^LN||99|ng/dL|<999|N|||F
```

```
OBX|8|NM|53345-5^Cortisol [Mass/volume] in Dried blood spot^LN||99|ug/dL|<999|N|||F

OBR|12|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54090-
6^Thyroid newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46762-1^Congenital hypothyroidism newborn screen interpretation^LN||LA18592-8^In
range^LN|||N|||F

OBX|2|TX|57705-6^Congenital hypothyroidism newborn screening comment-discussion^LN||Any baby
with clinical features suggestive of a metabolic disorder requires clinical and diagnostic
follow-up regardless of whether the NBS result is normal or abnormal.|||N|||F

OBX|3|NM|31144-9^Thyroxine (T4) [Mass/volume] in Dried blood spot^LN||10.36|ug/dL|<25|N|||F

OBX|4|NM|29575-8^Thyrotropin [Units/volume] in Dried blood spot^LN||1.2|m[IU]/L|<8|N|||F
```

Section 4.5: Galactosemia panel

The tests for galactosemia are quantitative enzyme activity measures. Some feeding types may interfere with interpretation and should be reported under the DBS card data panel using LOINC code 67704-7 (Feeding types) with its coded answer list.

```
OBR|13|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54079-
9^Galactosemia newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46737-3^Galactosemias newborn screen interpretation^LN||LA18592-8^In range^LN|||N|||F

OBX|2|TX|57704-9^Galactosemias newborn screening comment-discussion^LN||Any baby with clinical
features suggestive of a metabolic disorder requires clinical and diagnostic follow-up
regardless of whether the NBS result is normal or abnormal.|||N|||F

OBX|3|NM|54084-9^Galactose [Mass/volume] in Dried blood spot^LN||1.6|mg/dL|<11|N|||F

OBX|4|NM|42906-8^Galactose 1 phosphate uridyl transferase [Enzymatic activity/volume] in Dried
blood spot^LN||99|U/g{Hb}|<999|N|||F

OBX|5|NM|40842-7^Galactose 1 phosphate [Mass/volume] in Dried blood
spot^LN||99|mg/dL|<999|N|||F
```

Section 4.6: Hemoglobinopathies panel

The hemoglobinopathies panel provides a flexible way to report the hemoglobins found in a particular sample without having to create and maintain answer lists for every possible hemoglobin combination. Separate OBX segments are used to represent up to five hemoglobin types that are found in the sample in the order of predominance from most predominant to fifth most predominant. The hemoglobin types are specified in a coded LOINC answer list that includes a code for an unidentified hemoglobin. Only when an unidentified hemoglobin is found, additional OBX segments with LOINC 64122-5 should be added to indicate which hemoglobins a lab is able to identify, so that the receiver will know that the unidentified hemoglobin is not one of those. This is similar to the use of multiple OBX segments with LOINC 57719-7 for Conditions tested in this newborn screening. The hemoglobin interpretation may be omitted if no specific hemoglobin condition is suspected based on the pattern.

Note, the older LOINC code with a fixed answer list for hemoglobin patterns is retained for backwards compatibility, but the approach described above and illustrated below is the preferred method for reporting the hemoglobin screening result.

Some states using HPLC report quantitative percentages of the hemoglobin bands that are detected, and they can still do so using the LOINC codes for hemoglobin percentages. Similar to cystic fibrosis, some states are beginning to use second tier genetic testing that allows precise diagnosis of certain conditions, but Regenstrief has not yet developed LOINC codes or methods for reporting genetic testing results for hemoglobin disorders.

Example of reporting an in-range hemoglobin result:

```
OBR|14|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54081-
5^Hemoglobinopathies newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46740-7^Hemoglobin disorders newborn screen interpretation^LN|1|LA11995-0^Normal
hemoglobins^LN|||N|||F

OBR|15|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|64116-
7^Hemoglobin observations newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|64117-5^Most predominant hemoglobin in Dried blood spot^LN|1|LA16208-3^Hb F^LN|||N|||F

OBX|2|CE|64118-3^Second most predominant hemoglobin Dried blood spot^LN|1|LA16209-1^Hb
A^LN|||N|||F
```

Example of reporting a result for a sample that was likely post-transfusion:

```
OBR|14|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54081-
5^Hemoglobinopathies newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46740-7^Hemoglobin disorders newborn screen interpretation^LN|1|LA18593-6^Out of
range^LN|||N|||F

OBX|2|CE|71592-0^ Hemoglobinopathies conditions suspected [Identifier] in Dried blood
spot^LN|1|10593005^Likely transfusion^SCT|||N|||F

OBX|3|TX|57703-1^Hemoglobin disorders newborn screen comment/discussion^LN|1|The sample appears
to be a post-transfusion sample due to the predominance of adult hemoglobin. A repeat sample
should be submitted 90 days after the last transfusion.|||N|||F

OBR|15|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|64116-
7^Hemoglobin observations newborn screening
panel^LN|||201010141853|||VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.11
3883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|64117-5^Most predominant hemoglobin in Dried blood spot^LN|1|LA16209-1^Hb A^LN|||N|||F

OBX|2|CE|64118-3^Second most predominant hemoglobin Dried blood spot^LN|1|LA16208-3^Hb
F^LN|||N|||F
```

Example of reporting an unidentifiable hemoglobin:

```
OBR|14|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54081-
5^Hemoglobinopathies newborn screening
```

```
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46740-7^Hemoglobin disorders newborn screen interpretation^LN|1|LA18593-6^Out of
range^LN|||N|||F

OBX|1|TX|57703-1^Hemoglobin disorders newborn screening comment/discussion^LN|2|An unidentified
hemoglobin was detected that cannot be interpreted by newborn screening. Suggest hematology
referral and diagnostic testing at an appropriate age.|||N|||F

OBR|15|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|64116-
7^Hemoglobin observations newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|64117-5^Most predominant hemoglobin in Dried blood spot^LN||LA16208-3^Hb F^LN|||N|||F

OBX|2|CE|64118-3^Second most predominant hemoglobin Dried blood spot^LN||LA16223-2^Hb
unidentified^LN|||N|||F

OBX|3|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|1|LA16208-3^Hb F^LN|||N|||F

OBX|4|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|2|LA16209-1^Hb A^LN|||N|||F

OBX|5|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|3|LA13002-3^Hb C^LN|||N|||F

OBX|6|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|4|LA13003-1^Hb D^LN|||N|||F

OBX|7|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|5|LA13005-6^Hb E^LN|||N|||F

OBX|8|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|6|LA16218-2^Hb G^LN|||N|||F

OBX|9|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|7|LA16220-8^Hb H^LN|||N|||F

OBX|10|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|8|LA16222-4^Hb O-Arab^LN|||N|||F

OBX|11|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|9|LA13007-2^Hb S^LN|||N|||F

OBX|12|CE|64122-5^Hemoglobins that can be presumptively identified based on available controls
in Dried blood spot^LN|10|LA16223-2^Hb unidentified^LN|||N|||F
```

Section 4.7: Biotinidase panel

The test for biotinidase deficiency gives a qualitative result and is a good illustration of how to report an out of range qualitative test.

```
OBR|16|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|57087-
9^Biotinidase newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.1
13883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F
```

```
OBX|1|CE|46761-3^Biotinidase deficiency newborn screen interpretation^LN||LA4259-
3^Borderline^LN|||A|||F

OBX|2|TX|57699-1^Biotinidase deficiency newborn screening comment/discussion^LN||"Borderline
abnormal screen for biotinidase deficiency (BIOT). Slightly decreased biotinidase activity,
unlikely to be significant. Suggest clinical follow-up and repeat newborn metabolic
screen."|||A|||F

OBX|3|ST|38478-4^Biotinidase [Presence] in Dried blood spot^LN||reduced enzyme activity|||A|||F
```

Section 4.8: Severe combined immunodeficiency (SCID) panel

The SCID panel includes codes for the quantitative TREC assay, interpretation and comment/discussion.

```
OBR|17|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|62333-
0^Severe combined newborn screening immunodeficiency (SCID) panel in Dried blood
spot^LN|||201010141853|||^VH||||201101040920||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.11
3883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201101051142|||F

OBX|1|CE|62321-5^Severe combined immunodeficiency newborn screen interpretation^LN||LA6626-
1^Normal^LN|||N|||F

OBX|2|TX|62322-3^Severe combined immunodeficiency newborn screening comment-discussion^LN||Any
baby with clinical features suggestive of an immune system disorder requires clinical and
diagnostic follow-up regardless of whether the NBS result is normal or abnormal.|||N|||F

OBX|3|NM|62320-7^T-cell receptor excision circle [#/volume] in Dried blood spot by Probe &
target amplification method^LN||100|{copies}|>60|N|||F
```

Section 5: Point-of-care (POC) screening results

The POC screening result panel includes subpanels for early hearing loss detection and critical congenital heart disease (CCHD). These panels are different from other panels because they are reporting the result of a point of service test performed in the hospital, not a result measured in the laboratory. However, the result may be recorded on the filter paper card, and some labs will include the hearing and/or CCHD result t along with dried blood spot (DBS) results to create a single newborn screening report for the convenience of clinicians. Because these two screening tests will likely be performed by different personnel at different points in time, we recommend reporting the results using the subpanels as shown below.

```
OBR|18|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|73738-
7^Newborn screening test results panel - Point of
Care^LN|||201010141853|||^VH||||||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.4.6&ISO
^L^^^NPI^^^^^^^^MD||||||201010160918|||F
```

Section 5.1: Hearing screening panel

The hearing screening panel includes separate subpanels for the left and right ear because they are tested independently. Hearing screening results are reported as “Pass” or “Refer.” Various methods are used for hearing screening, and the specific method used should be recorded using the coded answer list for LOINC code 54106-0 Newborn hearing screen method.

```
OBR|19|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54111-
0^Newborn hearing loss
panel^LN|||201010141853|||^VH||||||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.4.6&IS
O^L^^^NPI^^^^^^^^MD||||||201010160918|||F
```

```
OBX|1|TX|57700-7^Hearing loss newborn screening comment/discussion^LN||Any baby with clinical
features suggestive of hearing loss requires clinical and diagnostic follow-up regardless of
whether the NMS result is normal or abnormal.|||N|||F

OBX|2|CE|54106-0^Newborn hearing screen method^LN||LA10388-9^Auditory brain stem
response^LN|||N|||F

OBR|20|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|73744-
5^Newborn hearing screen panel of Ear -
right^LN|||201010141853|||^VH||||||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.4.6&IS
O^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|54109-4^Newborn hearing screen of Ear - right^LN||LA10392-1^Pass^LN|||N|||F

OBR|21|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|73741-
1^Newborn hearing screen panel of Ear -
left^LN|||201010141853|||^VH||||||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.4.6&ISO
^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|54108-6^Newborn hearing screen of Ear - left^LN||LA10392-1^Pass^LN|||N|||F
```

Section 5.2: Critical congenital heart disease (CCHD) panel

The CCHD panel is unique in that it 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. Different states have different screening protocols, most of which begin by considering a preductal and postductal measurement and the difference between the two, followed by, depending on the initial results, subsequent measurements at specified intervals. Each program can use the appropriate combination of the available CCHD codes to reflect its protocol.

```
OBR|22|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|73805-4^CCHD
newborn screening
panel^LN|||201010141853|||^VH||||||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.4.6&IS
O^L^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|73700-7^CCHD newborn screening interpretation^LN||LA18592-8^In range^LN|||N|||F

OBX|2|NM|73806-2^Newborn age in hours^LN||36|h||N|||F

OBX|3|CE|73699-1^Number of prior CCHD screens [#] Qualitative^LN||LA6111-4^0^LN|||N|||F

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

OBX|5|CE|73796-5^Infant activity during preductal oxygen saturation measurement^LN||LA19830-
1^Awake and quiet^LN|||N|||F

OBX|6|NM|59418-4^Oxygen saturation in Blood Postductal by Pulse oximetry^LN||97|%|>95|N|||F

OBX|7|CE|73792-4^Infant activity during postductal oxygen saturation measurement^LN||LA11864-
8^Sleeping^LN|||N|||F

OBX|8|NM|73696-7^Oxygen saturation.preductal-oxygen saturation.postductal [Mass fraction
difference] in Bld.preductal and Bld.postductal^LN||2|%|<3|N|||F
```

About LOINC, SNOMED CT and UCUM Coding Standards

A coding and terminology framework is essential to standardizing laboratory reporting and enabling interoperability of information exchange across Electronic Health Record (EHR) platforms. Coding standards used in this example message include LOINC, SNOMED CT and UCUM.

Logical Observation Identifiers Names and Codes **(LOINC®)** is a terminology standard for identification of laboratory tests and other measurements. It is available free of charge in a database that carries universal codes, names and other attributes for laboratory and other kinds of tests, clinical reports, measurements, survey instruments and other observations. It was developed to enable the exchange and pooling of clinical results for clinical care, outcomes management, and research. The LOINC terminology was developed by the LOINC Committee and Regenstrief Institute and is maintained by the Regenstrief Institute, Inc., a non-profit medical research organization associated with Indiana University. You can download the database and a browser program (also no cost) from http://loinc.org/downloads. The LOINC and Regenstrief LOINC Mapping Assistant (RELMA®) Terms of Use are available at http://loinc.org/terms-of-use.

Systematized Nomenclature of Medicine — Clinical Terms **(SNOMED CT®)** is a comprehensive, multilingual clinical health care terminology designed for use in electronic health record systems and in health data exchange. SNOMED CT aims to facilitate communication and interoperability in electronic health data exchange. Originally created by the College of American Pathologists (CAP) in cooperation with the UK National Health Service, SNOMED CT is now owned, maintained and distributed by the International Health Terminology Standards Development Organisation (IHTSDO), a not-for-profit association in Denmark, with contract assistance from the CAP. It is available free of charge in IHTSDO member countries, including the US, in low-income countries as defined by the World Bank, and for qualified research projects in any country. NLM is the US Member of the IHTSDO. Information about obtaining SNOMED CT (in multiple formats) is available at http://www.nlm.nih.gov/research/umls/licensedcontent/snomedctfiles.html. A free Unified Medical Language System® (UMLS®) Metathesaurus license (which includes the IHTSDO Affiliate license) is required. It can be obtained via the same site.

Unified Code for Units of Measure **(UCUM©)** units are the preferred units for reporting quantitative NBS results. Using UCUM units creates interoperability by allowing comparison of results from different labs that use different units for the same test. The standard includes a tool for transforming local units into UCUM units. UCUM was developed and is maintained by the Regenstrief Institute. It has been adopted nationally as well as internationally by such standards organizations as HL7 and DICOM. More information and a link to the UCUM specification is available at http://unitsofmeasure.org/.