HRSA/NLM Guidance for
Sending Electronic Newborn Screening Results with HL7 Messaging
Version 5.2

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Introduction

This implementation guide is the product of a joint effort by the US Health Resources and Services Administration (HRSA) and the US National Library of Medicine (NLM). The purpose of this guidance is to assist those sending and receiving the results of newborn screening (NBS) tests in electronic format to implement systems that facilitate such messaging. It has been harmonized with the Public Health Informatics Institute's (PHII) Newborn Dried Blood Spot (NDBS) Screening Implementation Guide for Laboratory Results, which should be consulted when additional technical details are required.

Here we present an annotated example of a Health Level Seven (HL7) version 2.5.1 NBS result message in a machine-readable (HL7) format. This example includes the clinical questions asked on the dried blood spot (DBS) card, summary information about the result and detailed information about specific tests and other data related to the infant. 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 specification is tightly tied to the template defined in Logical Observations, Identifiers, Numbers and Codes (LOINC®) panel 54089-8 (Newborn screening panel American Health Information Community, available at http://loinc.org/newborn-screening/54089-8/details.pdf, which fully defines the observations that *may* be included in a NBS message, including items such as the specimen quality, the baby's birth weight, the TSH result and much more.

The LOINC panel 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 template lists its data type, and if applicable according to its data type, associated coded answer list (plus SNOMED CT codes), standard units of measure for reporting quantitative information, and other attributes.

See Figure 1 on the next page for a small part of the LOINC NBS panel. To see the whole panel, or to download it in pdf or xls format, go to:
http://newbornscreeningcodes.nlm.nih.gov/HL7.

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	R	1..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	New born screening report - overall interpretation	R	1..1	CE	
57131-5	Newborn conditions with positive markers [Identifier] in Dried blood spot	R	1..n	CE	
57720-5	Newborn conditions with equivocal markers [Identifier] in Dried blood spot	R	1..n	CE	
57724-7	Newborn screening short narrative summary	O	0..1	FT	
57129-9	Full newborn screening summary 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	
57717-1	Newborn screen card data panel				
57716-3	State of origin [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	
57722-1	Birth plurality of Pregnancy	R		CE	
57714-8	Obstetric estimation of gestational age	R		NM	wk

Figure 1- A small portion of the LOINC NBS panel

The LOINC panel includes variables to accommodate every analyte and every analyte ratio that we have identified as part of any jurisdiction's newborn screening program. It also includes variables for reporting most of the dried blood spot (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.

HL7 messages contain some predefined segments (e.g. PID, NK1, ORC) that are intended to carry certain universal data elements (e.g. name, date of birth), as well as more general OBX segments that are used to report observation results. Some of the demographic card variables, such as mother's name and contact information, should be reported in the predefined segments and therefore are not represented in the LOINC template. Other card variables will be reported as result data in OBX segments using their appropriate LOINC codes. Complete details regarding each type of segment and the information it carries may be obtained from the HL7 v2.5.1 messaging specification. The Public Health Informatics Institute's (PHII) Newborn Dried Blood Spot (NDBS) Screening Implementation Guide for Laboratory Results includes a table of the essential fields in each HL7 segment used to send newborn screening results, as well as a table with details about which specific segment should be used to report each card variable. The general approach is to have each NBS laboratory choose the elements it needs to report the specific tests it performs from this comprehensive set of variables.

Emphasis on Structure and Comprehensiveness

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. We discourage the use of NTE (general un-coded notes) segments anywhere in the message so that the data in the message is as clear and structured as possible. We have included variables for comment and discussion within every subpanel, so NTE segments should not be necessary.

Brief overview of the major HL7 segments OBR and OBX

Here we give a very brief overview of HL7 version 2, but implementers and serious users should develop a working knowledge of HL7 v2 in general as well as more specifically, the HL7 Version 2.5.1 Implementation Guide: Orders and Observations; Interoperable Laboratory Result Reporting to EHR, Release 1, which is available free to HL7 members and for purchase by non-members from Health Level Seven (https://www.hl7.org/store/index.cfm?ref=nav)

HL7 version 2.x messages consist of "records" called segments; these are represented as ASCII Text – with data fields and sub-fields separated by delimiters.

- Segments always begin with a 3-character designation (e.g. OBR, OBX, MSH, PID, NK1) that indicate segment type
- Segments always end with a carriage return character, sometimes indicated as <CR>.
- Vertical bars or pipes (|) separate 2 adjacent data fields in a segment and also separate the segment ID from the first data field in a segment.
- Hats (^) separate subfields
- Ampersands (&) separate subfield components
- Tildes (~) separate repeating values within a field

HL7 Payload Segments and Content

- Each kind of segment is distinguished by a leading three-character code. The order/report header segment identifies the panel, carries other information that applies to all of the observations within the panel and is identified by a leading "OBR." The observation segment – that carries answers to questions and the value of measured and computed results – is identified by a leading "OBX."

- 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
```

- The fields in a segment are identified by counting delimiters. The first field in the OBX segment, OBX-1, begins after the first field delimiter (the vertical bar). The second one (OBX-2) follows immediately after the second delimiter, and so on.
- OBX-1 carries the sequence number to distinguish multiple OBX segments. These are simple counts that start at 1 after each OBR segment. Consequently, a message can have multiple OBX segments, each with a different sequence number and each with multiple numbered fields.
- OBX-2 contains the data type of the test result (e.g. ST = string, NM = numeric, CE = coded entry).

- OBX-3 contains the observation ID – including code, print text and code system – it is a CE data type, and it represents the variable or "question." HL7 permits senders to include both the Universal LOINC code and the local code for a given observation. Within the OBX-3 fields in our example (above) we display the universal code in red and the local code in turquoise as follows: LOINC Code^Print Text^LN^Local Code^Print Text^L
- OBX-4 (sub ID) – is used to distinguish multiple OBX's with the same Observation ID (OBX-3). The value of the sub ID should increment by one for each OBX with the same observation ID. OBX-4 also enables grouping of OBXs, but we do not use that capability in NBS reporting.
- OBX-5 contains the observation value or test result/impression. You can think of it as the value of the variable identified in OBX-3 or the "answer" to the question asked in OBX-3. In the example OBX above, we display these values in green to clarify the structure of the message. Depending upon the data type identified in OBX-2 the value will be :
    - Numeric – e.g. TSH results. Most numerically valued measures will have units of measure (listed in OBX-6). These should be represented as Unified Code for Units of Measure (UCUM) units as shown in the LOINC template. Some numerically valued measures – e.g. pure ratios – do not have true units but can be indicated in UCUM with the text string {Ratio} to help users identify computed ratios. The reference range or cutoff for a numeric result is reported in OBX-8.
    - Coded -- e.g. Conditions with positive markers. As indicated above, both a standard code set, such as SNOMED CT or LOINC, and a local code set may be used.
    - Narrative text – e.g. the discussion/ description variables
- OBX-6 Units of Measure. We display them in blue for emphasis within the example OBX above. UCUM is the preferred standard to represent units of measure.
- OBX-7 Reference Ranges. We display them in pink for emphasis in the example OBX above. These are strings and, if they contain units of measure, these units should match those in OBX-6.
- OBX-8 Abnormal Flags: Consistent with HL7 v 2.5.1, we use 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 documented in table 0078 Abnormal Flags in the HL7 specification.
- OBX-11 Observation Result Status. These are derived from HL7 table 0085. For example, F = Final results, 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 is the observation date/time, which is the time when the specimen was collected. OBR-14 is 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 when the results were reported. 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 to the nearest day, which is why the time of birth should also be sent in a separate OBX segment.

Note: It has been proposed that OBX-13 could be used to tailor access to specific results based on user role, e.g. detailed quantitative data would not be displayed to clinicians when all results in a section are normal. However, there are no clear and uniform standards for using OBX-13 to suppress printing of selected OBX segments. Therefore, we suggest that message receivers can build rule-based filters to remove quantitative detail that

some users might not wish to print, thus shortening routine clinical reports while retaining full detail for other purposes. One possible approach is filtering the message using the OBX-8 abnormal flags.

For a comprehensive and official HL7 list of segments and the fields they contain, please see the HL7 version 2.5.1 specification: HL7 Version 2.5.1 Implementation Guide: Orders and Observations; Interoperable Laboratory Result Reporting to EHR, Release 1

Overview of the example message

The focus in this example message is on the "results payload" – the OBR and OBX segments. In this version of the example, we have included all of the segments required for this message.

We invented the result values and normal ranges you see in this example without any attempt to be clinically correct. These formats are similar to what you would see in real messages, but a NBS lab would include its real results and local reference ranges according to its usual practices.

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. This specification recommends including the local laboratory code in addition to the LOINC code for identifying the variable. The string "LN" is used to represent LOINC as the primary coding system, and the local code is identified by an "L" in the alternate coding system field. SNOMED-CT, which is used as an alternate coding system for some of the answer codes, is identified by "SCT."

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.

Annotated Example NBS HL7 Message:

Throughout the annotated example message, notes or comments usually precede the segment(s) they refer to as they often cover several segments and clarify the information that will follow the note. For a complete listing of the LOINC codes for newborn screening, please download the LOINC panel 54089-8 Newborn screening panel American Health Information Community (AHIC), available in pdf or xls format at http://newbornscreeningcodes.nlm.nih.gov/HL7. The LOINC panel specifies whether individual LOINC codes are required, optional or conditional.

NBS Message Section 1:
Administrative Segments of HL7 message –
Message description, patient identification

The administrative segments, sometimes called header segments, appear at the beginning of the message and are summarized in the Public Health Informatics Institute's (PHII) Newborn Dried Blood Spot (NDBS) Screening Implementation Guide for Laboratory Results. They carry essential demographic and message control data used to process the message.

Note: 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 using the HL7 hierarchical data (HD) data type.

```
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
```

Note: The PID (Patient Identification) segment refers to the baby, but the data may come from the mother's record. Usually, at the time of the initial screen, an infant will not have a SSN but should have a medical record number.

```
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
```

Note: The NK1 (Next of Kin) segment is used to carry data about the mother, and additional NK1 segments can be added to carry data about the father or another caregiver. In some circumstances when the mother's data is not reported (e.g. adoption), there will be only one NK1 segment with the caregiver's information. The example NK1 includes a mother's Medicaid number that was assigned by the state of Georgia with the identifier type codes as MA for Medicaid and the assigning authority coded as GA for Georgia, using a FIPS 2 letter state code to identify the assigning authority for the Medicaid number.

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

Note: 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 is identified by its NPI number and assigning authority identifier type is identified by the string "NPI" which is a recommended extension to HL7 table 301 for universal identifier types. The ordering provider is also identified by an NPI number, and in that case the string "NPI" is also used with the OID for National Provider Identifiers using the "ISO" universal identifier type. Both methods for identifying the assigning authority for an NPI are valid and are included here to show both options.

```
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
```

NBS Message Section 2: Report Summary

A report summary section is required. At a minimum, this section should include the required OBX (observation/result) 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.

Note: The first OBR (Observation Request) segment marks the beginning of the result data and can contain the optional sub-panel OBR headers or may be followed directly by the OBX segments with result data.

```
OBR|1|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|54089-8^NB
Screen Pnl Patient AHIC^LN|||201010141853|||^VH||||201101040920
||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010
160918|||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
.113883.4.6&ISO^L^^^NPI^^^^^^^^MD||||||201010160918|||F

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

OBX|2|CE|57718-9^Sample quality of Dried blood spot^LN|1|LA12432-3^Acceptable^LN|||N|||F

OBX|3|CE|57130-7^Newborn screening report - overall interpretation^LN|1|LA12431-5^Not normal
requiring immediate non-filter paper follow-up for at least one condition^LN|||A|||F

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|LA14039-4^GBA^LN^190794006^Gaucher's disease^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
```

Note: 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. If the sender wants to embed a PDF file for the printable report within an HL7 message, one method is to send it as binary data.

```
OBX|7|FT|57724-7^Newborn screening short narrative summary^LN||"\.br\SUMMARY: Newborn
Metabolic Screen REQUIRES FOLLOW UP\.br\Sample Quality: Acceptable\.br\Amino Acids,
Normal\.br\Fatty acids, ABNORMAL MCAD SCREEN\.br\Organic acids, Normal\.br\TSH (CH),
Normal\.br\17-OH-Progesterone (CAH), Normal CAH screen\.br\Biotinidase, BORDERLINE BIOT
SCREEN\.br\IRT (Cystic Fibrosis), No evidence of cystic fibrosis.\.br\Hemoglobinopathies,
ABNORMAL HGB SCREEN due to transfusion, pre-transfusion screen was normal\.br\Lysosomal
disorders screen, ABNORMAL GAUCHER SCREEN\.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, Date of birth: 13 Oct 2010, Time
of birth: 06:32 am, Sex: Female, Age at collection: 30 hours, Mother's name: Lois
Lane\.br\Accession number: 200902, Collected: 14 Oct 2010, Received: 15 Oct 2010, Ordering
physician: Dr. Minnie Smiles\.br\SUMMARY: Newborn Metabolic Screen REQUIRES FOLLOW
UP\.br\Sample Quality: Acceptable\.br\Disorder, Screening Result, Analyte (Normal)\.br\Amino
Acids, Normal\.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

. . .

|||A|||F
```

Note: The required information about the conditions tested for in this newborn screening study is reported using separate OBX segments with a unique LOINC answer (LA) code for each test performed. Most of the conditions include a SNOMED CT code as a secondary code to facilitate the addition of these conditions to a problem list in an electronic health record when a diagnosis is confirmed.

The same answer list is used for conditions tested, conditions with positive markers, and conditions with equivocal markers, and the full answer list is included as part of the LOINC panel, available at:

http://loinc.org/newborn-screening/54089-8/details.pdf. Users can obtain the latest version of this list of conditions with mappings to SNOMED CT, and additional information about coding conditions detected through newborn screening at http://newbornscreeningcodes.nlm.nih.gov

```
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|LA12464-6^2M3HBA^LN^444755001^Disorder of isoleucine metabolism^SCT|||N|||F

OBX|11|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|3|LA12465-3^2MBG^LN|||N|||F

OBX|12|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|4|LA12466-1^3-MCC^LN^13144005^Methylcrotonyl-CoA carboxylase
deficiency^SCT|||N|||F

OBX|13|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|5|LA12468-7^3MGA^LN^297235006^Unclassified 3-methylglutaconic
aciduria^SCT|||N|||F

OBX|14|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|6|LA12469-5^5-OXO^LN^39112005^Glutathione synthase deficiency with 5-
oxoprolinuria^SCT|||N|||F

OBX|15|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|7|LA12470-3^ARG^LN^23501004^Arginase deficiency^SCT|||N|||F

OBX|16|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|8|LA12471-1^ASA^LN^41013004^Argininosuccinate lyase deficiency^SCT|||N|||F

OBX|17|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|9|LA12472-9^BIOPT-BS^LN^237914002^6-Pyruvoyl-tetrahydrobiopterin synthase
deficiency^SCT|||N|||F

OBX|18|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|10|LA12473-7^BIOPT-REG^LN^58256000^Dihydropteridine reductase
deficiency^SCT|||N|||F

OBX|19|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|11|LA12474-5^BKT^LN^237953006^Mitochondrial 2-methylacetoacetyl-CoA thiolase
deficiency - potassium stimulated^SCT|||N|||F

OBX|20|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|12|LA12475-2^CACT^LN^238003000^Carnitine acylcarnitine translocase
deficiency^SCT|||N|||F

OBX|21|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|13|LA12476-0^CBL A^LN^73843004^Cobalamin A disease^SCT|||N|||F

OBX|22|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|14|LA12477-8^CBL B^LN^82245003^Cobalamin B disease^SCT|||N|||F

OBX|23|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|15|LA12478-6^CBL C^LN^74653006^Cobalamin C disease^SCT|||N|||F

OBX|24|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|16|LA12479-4^CBL D^LN^31220004^Cobalamin D disease^SCT|||N|||F
```

```
OBX|25|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|17|LA12480-2^CBL E^LN^360373000^Homocystinuria vitamin B12-responsive type
III^SCT|||N|||F

OBX|26|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|18|LA12481-0^CBL G^LN^237938003^Cobalamin G (disorder)^SCT|||N|||F

OBX|27|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|19|LA12482-8^CIT-I^LN^398680004^Citrullinaemia^SCT|||N|||F

OBX|28|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|20|LA12483-6^CIT-II^LN^30529005^"Citrullinemia, neonatal type"^SCT|||N|||F

OBX|29|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|21|LA12485-1^CPT-Ia^LN^238001003^Carnitine palmitoyltransferase I
deficiency^SCT|||N|||F

OBX|30|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|22|LA12486-9^CPT-II^LN^238002005^Carnitine palmitoyltransferase II
deficiency^SCT|||N|||F

OBX|31|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|23|LA12487-7^CUD^LN^21764004^Renal carnitine transport defect^SCT|||N|||F

OBX|32|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|24|LA12489-3^De-Red^LN|||N|||F

OBX|33|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|25|LA12490-1^E3^LN^29914000^Dihydrolipoamide dehydrogenase
deficiency^SCT|||N|||F

OBX|34|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|26|LA12491-9^EMA^LN^81308009^^SCT|||N|||F

OBX|35|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|27|LA12492-7^FIGLU^LN^59761008^Glutamate formiminotransferase
deficiency^SCT|||N|||F

OBX|36|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|28|LA12493-5^GA-1^LN^76175005^"Glutaric aciduria, type 1"^SCT|||N|||F

OBX|37|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|29|LA12495-0^GA-2^LN^22886006^"Glutaric aciduria, type 2"^SCT|||N|||F

OBX|38|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|30|LA12497-6^HHH^LN^30287008^Hyperornithinaemia-hyperammonaemia-
homocitrullinuria syndrome^SCT|||N|||F

OBX|39|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|31|LA12498-4^HIS^LN^410058007^Histidinemia^SCT|||N|||F

OBX|40|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|32|LA12499-2^HMG^LN^410059004^Hydroxymethylglutaric aciduria^SCT|||N|||F

OBX|41|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|33|LA12500-7^H-PHE^LN^68528007^Hyperphenylalaninaemia^SCT |||N|||F

OBX|42|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|34|LA12501-5^Hyper LYS^LN^58558003^Hyperlysinemia^SCT|||N|||F
```

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OBX|43|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|35|LA12502-3^Hyper ORN^LN^314467007^Gyrate atrophy^SCT|||N|||F

OBX|44|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|36|LA12503-1^Hyper VAL^LN^47719001^Hypervalinemia^SCT|||N|||F

OBX|45|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|37|LA12504-9^IBG^LN|||N|||F

OBX|46|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|38|LA12505-6^IVA^LN^87827003^Isovaleryl-CoA dehydrogenase
deficiency^SCT|||N|||F

OBX|47|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|39|LA12506-4^LACTIC^LN^190882007^Lactic acidemia^SCT|||N|||F

OBX|48|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|40|LA12508-0^MAL^LN^124594007^Deficiency of malonyl-CoA
decarboxylase^SCT|||N|||F

OBX|49|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|41|LA12509-8^MCAD^LN^128596003^Medium-chain acyl-coenzyme A dehydrogenase
deficiency^SCT|||A|||F

OBX|50|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|42|LA12510-6^MCD^LN^360369003^Holocarboxylase synthase deficiency^SCT|||N|||F

OBX|51|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|43|LA12511-4^MCKAT^LN^124265004^Deficiency of acetyl-CoA
acyltransferase^SCT|||N|||F

OBX|52|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|44|LA12512-2^MET^LN^43123004^Hypermethioninemia (disorder)^SCT|||N|||F

OBX|53|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|45|LA12513-0^MSUD^LN^27718001^Maple syrup urine disease^SCT|||N|||F

OBX|54|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|46|LA12514-8^MTHFR^LN^41797007^"5,10-Methylenetetrahydrofolate reductase
deficiency"^SCT|||N|||F

OBX|55|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|47|LA12515-5^MUT^LN^124680001^Deficiency of methylmalonyl-CoA
mutase^SCT|||N|||F

OBX|56|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|48|LA12516-3^NKHG^LN^237939006^Non-ketotic hyperglycinaemia^SCT|||N|||F

OBX|57|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|49|LA12517-1^OH PRO^LN^25739007^Hyperhydroxyprolinaemia^SCT|||N|||F

OBX|58|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|50|LA12518-9^OTC^LN^80908008^Ornithine carbamoyltransferase
deficiency^SCT|||N|||F

OBX|59|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|51|LA12519-7^PC^LN^87694001^Pyruvate carboxylase deficiency^SCT|||N|||F

OBX|60|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|52|LA12520-5^PKU^LN^7573000^Classical phenylketonuria^SCT|||N|||F
```

```
OBX|61|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|53|LA12521-3^PRO I^LN^61071003^Proline dehydrogenase deficiency^SCT|||N|||F

OBX|62|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|54|LA12522-1^PRO II^LN^124177001^Deficiency of pyrroline-5-carboxylate
reductase^SCT|||N|||F

OBX|63|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|55|LA12523-9^PROP^LN^69080001^Propionic acidemia^SCT|||N|||F

OBX|64|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|56|LA12524-7^SCAD^LN^124166007^Deficiency of butyryl-CoA
dehydrogenase^SCT|||N|||F

OBX|65|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|57|LA12525-4^SCHAD^LN^237998000^Short chain 3-hydroxyacyl-CoA dehydrogenase
deficiency^SCT|||N|||F

OBX|66|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|58|LA12526-2^SUCLA2^LN|||N|||F

OBX|67|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|59|LA12527-0^TFP^LN^237999008^Mitochondrial trifunctional protein
deficiency^SCT|||N|||F

OBX|68|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|60|LA12528-8^TYR-1^LN^410056006^Tyrosinaemia type I^SCT|||N|||F

OBX|69|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|61|LA12529-6^TYR-II^LN^4887000^"Hypertyrosinemia, Richner-Hanhart
type"^SCT|||N|||F

OBX|70|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|62|LA12530-4^TYR-III^LN^415764005^Tyrosinemia type III^SCT|||N|||F

OBX|71|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|63|LA12531-2^VLCAD^LN^237997005^Very long chain acyl-CoA dehydrogenase
deficiency^SCT|||N|||F

OBX|72|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|64|LA12532-0^BIO^LN^8808004^Biotinidase deficiency^SCT|||A|||F

OBX|73|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|65|LA12533-8^CAH^LN^124214007^Deficiency of steroid 11-beta-
monooxygenase^SCT|||N|||F

OBX|74|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|66|LA12537-9^CF^LN^190905008^Cystic fibrosis^SCT|||N|||F

OBX|75|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|67|LA12538-7^CH^LN^190268003^Congenital hypothyroidism^SCT|||N|||F

OBX|76|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|68|LA12539-5^CH2^LN^82598004^Secondary hypothyroidism^SCT|||N|||F

OBX|77|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|69|LA12540-3^G6PD^LN^62403005^Glucose-6-phosphate dehydrogenase deficiency
anemia^SCT|||N|||F
```

```
OBX|78|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|70|LA12541-1^GALE^LN^8849004^UDPglucose-4-epimerase deficiency^SCT|||N|||F

OBX|79|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|71|LA12542-9^GALK^LN^124302001^Deficiency of galactokinase^SCT|||N|||F

OBX|80|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|72|LA12543-7^GALT^LN^398664009^Deficiency of UTP-hexose-1-phosphate
uridylyltransferase^SCT|||N|||F

OBX|81|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|73|LA12602-1^Hb C-carrier^LN^76050008^Hemoglobin C trait^SCT|||N|||F

OBX|82|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|74|LA12603-9^Hb D-carrier^LN^7391009^Hemoglobin D trait^SCT|||N|||F

OBX|83|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|75|LA12604-7^Hb E-carrier^LN^46248003^Hemoglobin E trait^SCT|||N|||F

OBX|84|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|76|LA12605-4^Hb O-Arab carrier^LN^445542007^Hemoglobin O-Arab trait^SCT|||N|||F

OBX|85|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|77|LA12606-2^Hb S (sickle)-carrier^LN^16402000^Sickle cell trait^SCT|||N|||F

OBX|86|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|78|LA12607-0^Hb C-disease^LN^51053007^Hemoglobin C disease^SCT|||N|||F

OBX|87|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|79|LA12608-8^Hb C beta-thalassemia^LN^61777009^Thalassemia-hemoglobin C
disease^SCT|||N|||F

OBX|88|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|80|LA12609-6^Hb D-disease^LN|||N|||F

OBX|89|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|81|LA12610-4^Hb D beta-thalassemia^LN^47047009^Thalassemia with other
hemoglobinopathy^SCT|||N|||F

OBX|90|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|82|LA12611-2^Hb beta zero-
thalassemia^LN^86715000^beta^0^Thalassemia^SCT|||N|||F

OBX|91|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|83|LA12612-0^Hb E-disease^LN^25065001^Hemoglobin E disease^SCT|||N|||F

OBX|92|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|84|LA12613-8^Hb E beta-thalassemia^LN^234392002^Hemoglobin E/beta thalassemia
disease^SCT|||N|||F

OBX|93|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|85|LA12614-6^Hb SS-disease (sickle cell anemia)^LN^127040003^Hereditary
hemoglobinopathy disorder homozygous for hemoglobin S^SCT|||N|||F

OBX|94|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|86|LA12616-1^Hb SC-disease^LN^35434009^Sickle cell-hemoglobin C
disease^SCT|||N|||F
```

```
OBX|95|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|87|LA12617-9^Hb SD-disease^LN^25472008^Sickle cell-hemoglobin D
disease^SCT|||N|||F

OBX|96|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|88|LA12618-7^Hb SE-disease^LN^47024008^Sickle cell-hemoglobin E
disease^SCT|||N|||F

OBX|97|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|89|LA12619-5^Hb S O-Arab disease^LN^127048005^Sickle cell-Hemoglobin O Arab
disease^SCT|||N|||F

OBX|98|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|90|LA12621-1^"Hb disease other than A, C, D, E, H,O-Arab,
S"^LN^80141007^Hemoglobinopathy^SCT|||N|||F

OBX|99|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried
blood spot^LN|91|LA12622-9^"Hb carrier other than C, D, E, S ,O-
Arab"^LN^123773003^Heterozygous hemoglobinopathy^SCT|||N|||F

OBX|100|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|92|LA12565-0^HIV^LN^52079000^Congenital human immunodeficiency virus
infection (disorder)^SCT|||N|||F

OBX|101|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|93|LA12566-8^SCID^LN^31323000^Severe combined immunodeficiency
disease^SCT|||N|||F

OBX|102|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|94|LA12567-6^TBG^LN^237544006^Thyroid-binding globulin
deficiency^SCT|||N|||F

OBX|103|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|95|LA12568-4^TOXO^LN^73893000^Congenital toxoplasmosis^SCT|||N|||F

OBX|104|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|96|LA14036-0^GLA^LN^16652001^Fabry's disease^SCT|||N|||F

OBX|105|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|97|LA14037-8^GAA^LN^237967002^"Glycogen storage disease, type
II"^SCT|||N|||F

OBX|106|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|98|LA14038-6^GALC^LN^192782005^Krabbe disease^SCT|||N|||F

OBX|107|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|99|LA14039-4^GBA^LN^190794006^Gaucher’s disease^SCT|||A|||F

OBX|108|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in
Dried blood spot^LN|100|LA14040-2^ASM^LN^58459009^Sphingomyelin/cholesterol
lipidosis^SCT|||N|||F
```

NBS Message Section 3: Newborn Screen Card Data Panel

The Card Variables contain demographics and clinical data from the filter paper order form that is used to send the request to the laboratory. See the LOINC panel for a full list. Each newborn screening program should only use the codes it needs to send the information it is required to send by law, policy or practice. Some information is entered in the administrative segments, and other variables are reported using LOINC or LOINC answer codes

in 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), there are LOINC answer codes that are entered in OBX-5 as result values. The required or optional status of some of variables may vary by state. The full answer lists are included the LOINC Panel, available at: http://newbornscreeningcodes.nlm.nih.gov/HL7.

```
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
```

NBS Message Section 4: Newborn Screening Results

This annotated example message includes screening result data (identified by LOINC codes) for many markers and derived variables (e.g. ratios). The LOINC AHIC newborn screening panel, available at http://loinc.org/newborn-screening/54089-8/details.pdf, includes all of the conditions and variables that could be reported by any state. Think of it as a master template from which each state could select the items it uses.

We propose that state laboratories report all quantitative and qualitative results to the state newborn screening program regardless of whether they are positive or negative indicators for the condition.

We recommend that state laboratories send quantitative results for at least all of the screen positive or equivocal conditions to all report receivers.

OBX-8 contains the normal/abnormal flag for the result reported in that segment. Per HL7 v 2.5.1, we use 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 flags could be used to filter a message for clinical display.

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 may 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. States generally should not send OBX segments without a value and should omit segments with LOINC codes for results that are not measured in their laboratory.

```
OBR|4|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |57794-
0^Newborn screening test results panel in Dried blood
spot^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie
^^^Dr^^^NPI&2.16.840.1.113883.4.6&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 with one segment (with LOINC code (46733-2) for amino acidemias) for a coded interpretation (Normal, Borderline, Abnormal requiring a repeat dried blood spot, Abnormal requiring an immediate other test), a second segment (with LOINC code (57793-2) for amino acidemias) to identify the specific amino acid disorder, a third segment (with LOINC code (57710-6) for amino acidemias) for narrative comment/discussion, and then a series of segments with the appropriate LOINC codes for the quantitative measurement of the individual amino acids included in the amino acid panel for a particular state lab.

For historical reasons, a few states have legislative mandates to report two specific conditions separately and explicitly, instead of using the general purpose approach that includes all amino acid conditions. These two conditions, Phenylketonuria (PKU) and Maple Syrup Urine Disease (MSUD), have their own individual LOINC codes for interpretation and for comment/discussion, which allows states to report Phenylketonuria Normal, Maple Syrup Disease Normal, and Amino Acids Normal, if they are required to do so. Other states can omit these condition specific interpretation and comment/discussion codes, and just report Amino Acids Normal.

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46733-2^Amino acidemias newborn screen interpretation^LN||LA6626-1^Normal^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
```

Note: The answer list for amino acid disorder suspected is a subset of the full condition list that is used to enter conditions with positive markers. The specific amino acid answer list is also specified under LOINC code 57793-2.

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

OBX|4|CE|46746-4^Phenylketonuria and variants/Biopterin defects newborn screen
interpretation^LN||LA6626-1^Normal^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
```

Note: The following condition-specific LOINC code should only be used by states that are required to report Maple Syrup Urine Disease separately from all other amino acid disorders. This OBX should be omitted by states that do not have that obligation as it is redundant with the information reported using code 46733-2.

```
OBX|6|CE|46743-1^Maple syrup urine disease newborn screen interpretation^LN|1|LA6626-
1^Normal^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
```

Note: Some of the quantitative result LOINC codes report 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}, which follows UCUM rules, so that all quantitative measurements have units regardless of whether they are computed or measured and to help users identify the computed values. These ratios are helpful to interpret the test results and identify the correct suspected condition.

```
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|< 950
umol/L|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 umol/L|N|||F

OBX|46|NM|53151-7^Valine/Phenylalanine [Molar ratio] in Dried blood spot^LN||1.44|{Ratio}|<
4.00|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 can be split into two separate sub-panels for two classes of disorders -- Fatty acid oxidation disorders and Organic acid disorders -- which are indicated by a common set of quantitative measures, some of which apply to one category of disorder, some to the other, and some to both. States can choose how to report their results under the single Acylcarnitine panel, under the two condition panels, or a combination of both.

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|58088-6^Acylcarnitine newborn screen interpretation^LN||LA12431-5^Not normal
requiring immediate non-filter paper follow-up 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
```

Note: The acylcarnitine panel includes a sub-panel for Fatty acid oxidation disorders.

```
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.
113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F
```

Note: This example uses the disorder MCAD to illustrate how to use the related codes within a subpanel for reporting an abnormal result. This group of codes represents fatty acid oxidation defects interpretation (46736-5), fatty acid oxidation suspected condition (57792-4), and fatty acid oxidation comment/discussion (57709-8), as well as the quantitative results and ratios with the appropriate normal/abnormal flag.

```
OBX|1|CE|46736-5^Fatty acid oxidation defects newborn screen panel^LN|1|LA12431-5^Not normal
requiring immediate non-filter paper follow-up 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|4|67.04|{Ratio}|<999|N|||F

OBX|6|NM|54462-7^Malonylcarnitine (C3-DC) [Moles/volume] in Dried blood
spot^LN||0.13|umol/L|< 1.40 umol/L|N|||F

OBX|7|NM|53234-1^Carnitine free (C0)/Stearoylcarnitine (C18) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|8|NM|53235-8^Carnitine free (C0)/Palmitoylcarnitine (C16)+Stearoylcarnitine (C18) [Molar
ratio] in Dried blood spot^LN||45.87|{Ratio}|<999|N|||F
```

```
OBX|9|NM|53236-6^Carnitine.free (C0)+Acetylcarnitine (C2)+Propionylcarnitine
(C3)+Palmitoylcarnitine (C16)+Oleoylcarnitine (C18:1)+Stearoylcarnitine (C18)/Citrulline
[Molar ratio] in Dried blood spot^LN||0.09|{Ratio}|<999|N|||F

OBX|10|NM|50157-7^Acetylcarnitine (C2) [Moles/volume] in Dried blood
spot^LN||31.78|umol/L|<999|N|||F

OBX|11|NM|53166-5^Butyrylcarnitine+Isobutyrylcarnitine (C4) [Moles/volume] in Dried blood
spot^LN||0.84|umol/L|0.75-1.02|N|||N

OBX|12|NM|53167-3^Butyrylcarnitine+Isobutyrylcarnitine (C4)/Acetylcarnitine (C2) [Molar
ratio] in Dried blood spot^LN||0|{Ratio}|<999|N|||F

OBX|13|NM|53168-1^Butyrylcarnitine+Isobutyrylcarnitine (C4)/Propionylcarnitine (C3) [Molar
ratio] in Dried blood spot^LN||0.26|{Ratio}|<999|N|||F

OBX|14|NM|53169-9^Butyrylcarnitine+Isobutyrylcarnitine (C4)/Octanoylcarnitine (C8) [Molar
ratio] in Dried blood spot^LN||2.04|{Ratio}|< 18.00|N|||F

OBX|15|NM|50102-3^3-Hydroxybutyrylcarnitine (C4-OH) [Moles/volume] in Dried blood
spot^LN||0.59|umol/L|0.43-0.66 |N|||F

OBX|16|NM|45211-0^Hexanoylcarnitine (C6) [Moles/volume] in Dried blood spot^LN||2.81|umol/L|<
0.25|H|||F

OBX|17|NM|53173-1^3-Hydroxyhexanoylcarnitine (C6-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|18|NM|45207-8^Glutarylcarnitine (C5-DC) [Moles/volume] in Dried blood
spot^LN||0.05|umol/L|[A75]|N|||F

OBX|19|NM|53174-9^Octenoylcarnitine (C8:1) [Moles/volume] in Dried blood
spot^LN||0.52|umol/L|0.21-0.7|N|||F

OBX|20|NM|53175-6^Octanoylcarnitine (C8) [Moles/volume] in Dried blood
spot^LN||19.71|umol/L|< 0.25|H|||NF

OBX|21|NM|53176-4^Octanoylcarnitine (C8)/Acetylcarnitine (C2) [Molar ratio] in Dried blood
spot^LN||0.813|{Ratio}|<0.050|H|||F

OBX|22|NM|53177-2^Octanoylcarnitine (C8)/Decanoylcarnitine (C10) [Molar ratio] in Dried blood
spot^LN||11.324|{Ratio}|< 4.000|H|||F

OBX|23|NM|53178-0^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-DC) [Moles/volume]
in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|24|NM|53402-4^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-
DC)/Butyrylcarnitine+Isobutyrylcarnitine (C4) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|25|NM|53179-8^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-
DC)/Decanoylcarnitine (C10) [Molar ratio] in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|26|NM|53180-6^Decadienoylcarnitine (C10:2) [Moles/volume] in Dried blood
spot^LN||0.07|umol/L|<0.12 |N|||F

OBX|27|NM|45198-9^Decenoylcarnitine (C10:1) [Moles/volume] in Dried blood
spot^LN||0.71|umol/L|< 0.20|H|||F
```

```
OBX|28|NM|45197-1^Decanoylcarnitine (C10) [Moles/volume] in Dried blood
spot^LN||0.31|umol/L|0.28-0.40|N|||F

OBX|29|NM|53182-2^3-Hydroxydecenoylcarnitine (C10:1-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|30|NM|53183-0^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)
[Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|31|NM|53403-2^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-
OH)/Butyrylcarnitine+Isobutyrylcarnitine (C4) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|32|NM|53184-8^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)/3-
Hydroxyisovalerylcarnitine (C5-OH) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|33|NM|53185-5^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-
OH)/Octanoylcarnitine (C8) [Molar ratio] in Dried blood spot^LN||3.63|{Ratio}|0.21-0.72|H|||F

OBX|34|NM|53186-3^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-
OH)/Palmitoylcarnitine (C16) [Molar ratio] in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|35|NM|45200-3^Dodecenoylcarnitine (C12:1) [Moles/volume] in Dried blood
spot^LN||0.31|umol/L|0.28-0.50|N|||F

OBX|36|NM|45199-7^Dodecanoylcarnitine (C12) [Moles/volume] in Dried blood
spot^LN||0.77|umol/L|0.44-0.80|N|||F

OBX|37|NM|53188-9^3-Hydroxydodecenoylcarnitine (C12:1-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|38|NM|53189-7^3-Hydroxydodecanoylcarnitine (C12-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|39|NM|53190-5^Tetradecadienoylcarnitine (C14:2) [Moles/volume] in Dried blood
spot^LN||0.12|umol/L|0.09-0.15|N|||F

OBX|40|NM|53191-3^Tetradecenoylcarnitine (C14:1) [Moles/volume] in Dried blood
spot^LN||0.48|umol/L|0.37-0.71|N|||F

OBX|41|NM|53192-1^Tetradecanoylcarnitine (C14) [Moles/volume] in Dried blood
spot^LN||0.61|umol/L|0.50-0.80|N|||F

OBX|42|NM|53193-9^Tetradecenoylcarnitine (C14:1)/Acetylcarnitine (C2) [Molar ratio] in Dried
blood spot^LN||0.51|{Ratio}|0.37-.070|N|||F

OBX|43|NM|53194-7^Tetradecenoylcarnitine (C14:1)/Dodecenoylcarnitine (C12:1) [Molar ratio] in
Dried blood spot^LN||1.53|{Ratio}|<999|N|||F

OBX|44|NM|53195-4^Tetradecenoylcarnitine (C14:1)/Palmitoylcarnitine (C16) [Molar ratio] in
Dried blood spot^LN||0.47|{Ratio}|0.37–0.70|N|||F

OBX|45|NM|53196-2^3-Hydroxytetradecadienoylcarnitine (C14:2-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F

OBX|46|NM|53197-0^3-Hydroxytetradecenoylcarnitine (C14:1-OH) [Moles/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F
```

```
OBX|47|NM|50281-5^3-Hydroxytetradecanoylcarnitine (C14-OH) [Moles/volume] in Dried blood
spot^LN||0.09|umol/L|<999|N|||F

OBX|48|NM|53198-8^Palmitoleylcarnitine (C16:1) [Moles/volume] in Dried blood
spot^LN||0.09|umol/L|<999|N|||F

OBX|49|NM|53199-6^Palmitoylcarnitine (C16) [Moles/volume] in Dried blood
spot^LN||6.13|umol/L|5.86-7.16|N|||F

OBX|50|NM|50121-3^3-Hydroxypalmitoleylcarnitine (C16:1-OH) [Moles/volume] in Dried blood
spot^LN||0.13|umol/L|0.10-0.15|N|||F

OBX|51|NM|50125-4^3-Hydroxypalmitoylcarnitine (C16-OH) [Moles/volume] in Dried blood
spot^LN||0.17|umol/L|0.09-0.19|N|||F

OBX|52|NM|53201-0^3-Hydroxypalmitoylcarnitine (C16-OH)/Palmitoylcarnitine (C16) [Molar ratio]
in Dried blood spot^LN||0.03|{Ratio}|< 0.20|N|||F

OBX|53|NM|45217-7^Linoleoylcarnitine (C18:2) [Moles/volume] in Dried blood
spot^LN||0.63|umol/L|0.62-0.65|N|||F

OBX|54|NM|53202-8^Oleoylcarnitine (C18:1) [Moles/volume] in Dried blood
spot^LN||2.42|umol/L|2.39-2.50|N|||F

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
```

Note: The acylcarnitine panel also includes a sub-panel for Organic Acid disorders.

```
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.
113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46744-9^Organic acidemias newborn screen interpretation^LN||LA6626-
1^Normal^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

OBX|6|NM|53160-8^Propionylcarnitine (C3) [Moles/volume] in Dried blood
spot^LN||5.17|umol/L|4.62-5.50|N|||F

OBX|7|NM|53161-6^Propionylcarnitine (C3)/Methionine [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|8|NM|53162-4^Propionylcarnitine (C3)/Carnitine.free (C0) [Molar ratio] in Dried blood
spot^LN||0.03|{Ratio}|<999|N|||F

OBX|9|NM|53163-2^Propionylcarnitine (C3)/Acetylcarnitine (C2) [Molar ratio] in Dried blood
spot^LN||0.15|{Ratio}|<999|N|||F

OBX|10|NM|54462-7^Malonylcarnitine (C3-DC) [Moles/volume] in Dried blood
spot^LN||0.13|umol/L|< 1.40 umol/L|N|||F

OBX|11|NM|53164-0^Propionylcarnitine (C3)/Palmitoylcarnitine (C16) [Molar ratio] in Dried
blood spot^LN||0.69|{Ratio}|< 2.0|N|||F

OBX|12|NM|53166-5^Butyrylcarnitine+Isobutyrylcarnitine (C4) [Moles/volume] in Dried blood
spot^LN||0.84|umol/L|0.75-1.02|N|||F

OBX|13|NM|53167-3^Butyrylcarnitine+Isobutyrylcarnitine (C4)/Acetylcarnitine (C2) [Molar
ratio] in Dried blood spot^LN||0|{Ratio}|<999|N|||F

OBX|14|NM|53168-1^Butyrylcarnitine+Isobutyrylcarnitine (C4)/Propionylcarnitine (C3) [Molar
ratio] in Dried blood spot^LN||0.26|{Ratio}|<999|N|||F

OBX|15|NM|53169-9^Butyrylcarnitine+Isobutyrylcarnitine (C4)/Octanoylcarnitine (C8) [Molar
ratio] in Dried blood spot^LN||2.04|{Ratio}|< 18.00|N|||F

OBX|16|NM|53170-7^Tiglylcarnitine (C5:1) [Moles/volume] in Dried blood
spot^LN||0.1|umol/L|0.09-0.24|N|||F

OBX|17|NM|45207-8^Glutarylcarnitine (C5-DC) [Moles/volume] in Dried blood
spot^LN||0.05|umol/L|<999|N|||F

OBX|18|NM|45216-9^Isovalerylcarnitine+Methylbutyrylcarnitine (C5) [Moles/volume] in Dried
blood spot^LN||0.43|umol/L|0.39-0.48|N|||F

OBX|19|NM|53238-2^Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Carnitine.free (C0) [Molar
ratio] in Dried blood spot^LN||0.00|{Ratio}|< 0.05|N|||F

OBX|20|NM|53239-0^Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Acetylcarnitine (C2) [Molar
ratio] in Dried blood spot^LN||0.00|{Ratio}|< 0.04|N|||F

OBX|21|NM|53240-8^Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Propionylcarnitine (C3)
[Molar ratio] in Dried blood spot^LN||0.31|{Ratio}|<999|N|||F

OBX|22|NM|53401-6^Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Octanoylcarnitine (C8)
[Molar ratio] in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|23|NM|50106-4^3-Hydroxyisovalerylcarnitine (C5-OH) [Moles/volume] in Dried blood
spot^LN||0.26|umol/L|<999|N|||F

OBX|24|NM|53171-5^3-Hydroxyisovalerylcarnitine (C5-OH)/Carnitine.free (C0) [Molar ratio] in
Dried blood spot^LN||99|{Ratio}|<999|N|||F
```

```
OBX|25|NM|53172-3^3-Hydroxyisovalerylcarnitine (C5-OH)/Octanoylcarnitine (C8) [Molar ratio]
in Dried blood spot^LN||0.436|{Ratio}|0.35-0.70|N|||F

OBX|26|NM|53178-0^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-DC) [Moles/volume]
in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|27|NM|53402-4^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-
DC)/Butyrylcarnitine+Isobutyrylcarnitine (C4) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|28|NM|53179-8^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-
DC)/Decanoylcarnitine (C10) [Molar ratio] in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|29|NM|45222-7^Methylmalonylcarnitine (C4-DC) [Moles/volume] in Dried blood
spot^LN||3.16|umol/L|<999|N|||F

OBX|30|NM|53181-4^Methylmalonylcarnitine (C4-DC)/3-Hydroxyisovalerylcarnitine (C5-OH) [Molar
ratio] in Dried blood spot^LN||99|{Ratio}|<999|N|||F

OBX|31|NM|53183-0^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)
[Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|32|NM|53403-2^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-
OH)/Butyrylcarnitine+Isobutyrylcarnitine (C4) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|33|NM|53184-8^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)/3-
Hydroxyisovalerylcarnitine (C5-OH) [Molar ratio] in Dried blood
spot^LN||99|{Ratio}|<999|N|||F

OBX|34|NM|53185-5^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-
OH)/Octanoylcarnitine (C8) [Molar ratio] in Dried blood spot^LN||3.63|{Ratio}|0.21-0.72|H|||F

OBX|35|NM|53186-3^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-
OH)/Palmitoylcarnitine (C16) [Molar ratio] in Dried blood spot^LN||99|{Ratio}|<999|N|||F

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
```

Section 4.2.1 Acylcarnitine Panel for Derivatized MSMS Method

Analysis of newborn screening dried blood spots by tandem mass spectrometry (MSMS) involves processing the specimen with reagents using one of two different methods referred to as derivatized and non-derivatized (or underivatized) method. Depending on which method a laboratory is using, a few of the analytes in the Fatty Acid Oxidation panel will be different and the appropriate LOINC codes should be selected based on the analytes and ratios that a laboratory measures and reports. Depending on the method a laboratory is using, some analytes cannot by distinguished on MSMS because they are isobaric or have a similar molecular weight. The example above is based on use of the derivatized MSMS method

When the derivatized MSMS methods is used, the main analytes measured include the following and their associated ratios:

C3-DC + C8-OH

C4-DC
C5-OH
C5-DC + C10-OH

Example OBX segments based on use of the derivatized MSMS method:

```
OBX|23|NM|53178-0^3-Hydroxyoctanoylcarnitine (C8-OH)+Malonylcarnitine (C3-DC) [Moles/volume]
in Dried blood spot^LN||99|umol/L|<999|N|||F

OBX|23|NM|50106-4^3-Hydroxyisovalerylcarnitine (C5-OH) [Moles/volume] in Dried blood
spot^LN||0.26|umol/L|<999|N|||F

OBX|29|NM|45222-7^Methylmalonylcarnitine (C4-DC) [Moles/volume] in Dried blood
spot^LN||3.16|umol/L|<999|N|||F

OBX|30|NM|53183-0^Glutarylcarnitine (C5-DC)+3-Hydroxydecanoylcarnitine (C10-OH)
[Moles/volume] in Dried blood spot^LN||99|umol/L|<999|N|||F
```

Section 4.2.2 Acylcarnitine Panel for Non-derivatized MSMS Method

When the non-derivatized MSMS methods is used, the main analytes measured include the following and their associated ratios:

C3-DC + C4-OH
C4-DC + C5-OH
C5-DC + C6-OH

Examples of OBX segments based on use of the non-derivatized MSMS method are shown below for illustration; however, a single HL7 message would not include OBX segments and LOINC codes from both the derivatized and non-derivatized methods.

```
OBX|23|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|29|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|30|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 offers the usual coded interpretation (such as Normal) and comment/discussion. 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. There are no established standards or answer codes for the data that is reported using code 54083-1 at this time.

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46769-6^Cystic fibrosis newborn screen interpretation^LN||LA6626-1^Normal^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|TX|54083-1^CFTR gene mutations found [Identifier] in Dried blood spot
Nominal^LN||None|||N|||F

OBX|4|NM|2077-6^Chloride [Moles/volume] in Sweat^LN||99|mmol/L|<999|N|||F

OBX|5|NM|48633-2^Trypsinogen I Free [Mass/volume] in Dried blood
spot^LN||99|umol/L|<999|N|||F
```

Section 4.4 Endocrine Panel

The Endocrine panel is 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 each separately in their own panel.

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F
```

Section 4.4.1 CAH Panel

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46758-9^Congenital adrenal hyperplasia newborn screen interpretation^LN||LA6626-
1^Normal^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
nmol/L |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
```

Section 4.4.2 CH Panel

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46762-1^Congenital hypothyroidism newborn screen
interpretation^LN||LA6626-1^Normal^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. There are certain feeding types that may interfere with interpretation, and those should be reported in the Newborn screen card data panel using the coded answer list for LOINC code 67704-7 Feeding Types.

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|46737-3^Galactosemias newborn screen interpretation^LN||LA6626-1^Normal^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

A new LOINC 64116-7 “Hemoglobin observations newborn screening panel” has been introduced to allow more complete and accurate reporting of the Hemoglobin observations than is possible using the fixed answer list for

LOINC 54104-5 Hemoglobin pattern that had been used in the past. 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 to fifth most predominant using LOINC codes 64117-5, 64118-3, 64119-1, 64120-9, and 64121-7. 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. 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. The older LOINC code with a fixed answer list for hemoglobin patterns will be retained for backwards compatibility, but use of the new sub-panel is the preferred method for reporting the hemoglobin screening result.

The "Hemoglobin observations" panel can accommodate the results from all three screening methods: electrophoresis, IEF isoelectric focusing, and HPLC high pressure liquid chromatography. 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. All states will report some uncommon or special findings as variants, but states differ in what they include in the definition of variants. While there are many LOINC codes for reporting hemoglobin included in the NBS Panel, states should only use the ones that are relevant to their laboratory practices and the findings of an individual patient. Transfusions will interfere with test interpretation, particularly when the transfusion introduces adult hemoglobin into the infant. Some conditions cannot be clarified until the infant is older and an adult hemoglobin pattern is established. Similar to cystic fibrosis, some states are beginning to use second tier genetic testing that allows precise diagnosis of certain conditions.

Example of reporting an identified hemoglobin panel and the hemoglobin disorders interpretation for a normal sample:

```
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
.113883.4.6&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
.113883.4.6&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 an identified hemoglobin panel and the hemoglobin disorders interpretation for a post-transfusion sample:

Note: In this clinical scenario, the sample is a second specimen obtained post- transfusion and the pre-transfusion sample was normal, hence additional samples will not be required for hemoglobinopathy diagnosis when the infant is older. The order of the most prominent hemoglobins is reversed because of the transfusion with adult hemoglobin. Reporting of quantitative measurement of hemoglobin percentage is also reported.

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|TX|57703-1^Hemoglobin disorders newborn screen comment/discussion^LN|1|The sample
appears to be a post-transfusion sample with adult hemoglobins. In this case, a pre-
transfusion sample was obtained and was normal. A repeat sample is not required when this
infant is older.|||N|||F

OBX|2|NM|54072-4^Hemoglobin A/Hemoglobin.total in Dried blood spot^LN|1|60|||N|||F

OBX|3|NM|54074-0^Hemoglobin F/Hemoglobin.total in Dried blood spot^LN|1|40|||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.
113883.4.6&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 if unidentifiable hemoglobin detected:

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||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
.113883.4.6&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
```

Example of reporting hemoglobin pattern (not encouraged) and/or percentages:

```
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
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|54104-5^Hemoglobin pattern in Dried blood spot by HPLC^LN||LA11974-5^Hb F,A
(normal)^LN|||N|||F

OBX|2|NM|54072-4^Hemoglobin A/Hemoglobin.total in Dried blood spot^LN||20|%|<100|N|||F

OBX|3|NM|54074-0^Hemoglobin F/Hemoglobin.total in Dried blood spot^LN||80|%|<100|N|||F

OBX|4|CE|46740-7^Hemoglobin disorders newborn screen interpretation^LN||LA11995-0^Normal
hemoglobins^LN|||N|||F
```

Section 4.7 Infectious Disease Panel

Some states have mandatory testing for HIV or other congenital infections. These are usually serologic tests with coded or string value results as well as an interpretation.

```
OBR|16|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |54082-
3^Infectious diseases newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|57702-3^Infectious diseases newborn screen interpretation^LN||LA6626-
1^Normal^LN|||N|||F

OBX|2|TX|57701-5^Infectious diseases newborn screening comment/discussion^LN||Any baby with
clinical features suggestive of an infectious disease requires clinical and diagnostic
follow-up regardless of whether the NBS result is normal or abnormal. |||N|||F

OBX|3|CE|54086-4^HIV 1+2 IgG Ab [Presence] in Dried blood spot^LN||LA6626-1^Normal^LN|||N|||F

OBX|4|CE|54087-2^Toxoplasma gondii IgG Ab [Presence] in Dried blood spot^LN||LA6626-
1^Normal^LN|||N|||F

OBX|5|CE|54088-0^Toxoplasma gondii IgM Ab [Presence] in Dried blood spot^LN||LA6626-
1^Normal^LN|||N|||F
```

Section 4.8 Hearing Loss Panel

The hearing loss panel is different from other panels because it is 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 report along with dried blood spot (DBS) results to create a single newborn screening report for the convenience of clinicians. The left and right ear are tested separately and reported as Pass or Refer. There are various methods 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|17|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |54111-
0^Newborn hearing loss panel^LN
|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.113883.
4.6&ISO^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|54109-4^Newborn hearing screen - right^LN||LA10392-1^Pass^LN|||N|||F

OBX|3|CE|54108-6^Newborn hearing screen - left^LN||LA10392-1^Pass^LN|||N|||F

OBX|4|CE|54106-0^Newborn hearing screen method^LN||LA10388-9^Auditory brain stem response^LN
|||N|||F
```

Section 4.9 Biotinidase Panel

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

```
OBR|18|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
.113883.4.6&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 4.10 G6PD Panel

A very small number of states test for G6PD so this panel is infrequently used. In addition, the testing methods are changing to specific genetic testing rather than enzyme assays, and oftentimes only an interpretation is given.

```
OBR|19|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |58091-
0^Glucose-6-Phosphate dehydrogenase (G6PD) newborn screen
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|58089-4^Glucose-6-Phosphate dehydrogenase newborn screen interpretation^LN||LA6626-
1^Normal^LN|||N|||F

OBX|2|TX|58090-2^Glucose-6-Phosphate dehydrogenase newborn screening
comment/discussion^LN||DNA analysis was performed for 5 mutations known to cause Glucose-6-
Phosphate Dehydrogenase deficiency. Approximately 11% of all G6PD deficiency cases are caused
by factors other than these five mutations. Results should be interpreted in the context of
clinical presentation|||N|||F
```

Section 4.11 Lysosomal Storage Disorders Panel

The Lysosomal Storage Disorders (LSD) panel is a new panel for a group of five different disorders that are undergoing pilot testing in some states and that have not yet been added to the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended uniform screening panel. The names for these codes are evolving, and codes and names for additional lysosomal storage disorders are expected to be added. Note that in addition to the overarching LSD panel, each disorder is also in a separate panel because states often test for only one of the five conditions, and it is not clear which ones may be included in the uniform panel in the future. Similar to the amino acid panel, all five conditions can either be reported as a group in one panel with one interpretation code, or as individual conditions under separate panels.

```
OBR|20|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |62300-
9^Lysosomal storage disorders newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201010160918|||F

OBX|1|CE|62301-7^Lysosomal storage disorders newborn screen interpretation^LN||LA12431-5^Not
normal requiring immediate non-filter paper follow-up for at least one condition^LN|||A|||F

OBX|2|CE|62302-5^Lysosomal storage disorders suspected [Identifier] in Dried blood
spot^LN||LA14039-4^GBA^LN^190794006^Gaucher's disease^SCT|||A|||F

OBX|3|TX|62303-3^Lysosomal storage disorders newborn screening comment-
discussion^LN||Abnormal result indicates possible Gaucher Disease and immediate referral to a
Metabolic Geneticist is indicated to confirm the diagnosis and begin treatment|||A|||F
```

Note: This is the panel for Fabry Disease:

```
OBR|21|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |62304-
1^Fabry disease newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1
.113883.4.6&ISO ^L^^^NPI^^^^^^^^MD||||||201101051142|||F

OBX|1|TX|62306-6^Fabry 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|2|CE|62305-8^Fabry disease newborn screen interpretation^LN||LA6626-1^Normal^LN|||N|||F

OBX|3|NM|55908-8^Alpha galactosidase A [Enzymatic activity/volume] in Dried blood
spot^LN||5.7|umol/h/L|>2.0|N|||F
```

Note: This is the panel for Krabbe Disease:

```
OBR|22|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA |62307-
4^Krabbe disease newborn screening
panel^LN|||201010141853|||^VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1
.113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201101051142|||F

OBX|1|CE|62308-2^Krabbe disease newborn screen interpretation^LN||LA6626-1^Normal^LN|||N|||F

OBX|2|TX|62309-0^Krabbe 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|3|NM|62310-8^Galactocerebrosidase [Enzymatic activity/volume] in Dried blood
spot^LN||2.4|umol/L/h|>0.5|N|||F
```

Note: This is the panel for Gaucher Disease:

```
OBR|23|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|62311-
6^Gaucher disease newborn screening
panel^LN|||201010141853|||VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.
113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201101051142|||F

OBX|1|CE|62312-4^Gaucher disease newborn screen interpretation^LN||LA12431-5^Not normal
requiring immediate non-filter paper follow-up for at least one condition^LN|||A|||F

OBX|2|TX|62313-2^Gaucher disease newborn screening comment-discussion^LN||Abnormal result
indicates possible Gaucher Disease and immediate referral to a Metabolic Geneticist is
indicated to confirm the diagnosis and begin treatment|||A|||F

OBX|3|NM|55917-9^Acid beta glucosidase [Enzymatic activity/volume] in Dried blood
spot^LN||1.3|umol/L/h|>4.1|L|||F
```

Note: This is the panel for Niemann-Pick disease:

```
OBR|24|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|62315-
7^Nieman Pick disease A/B newborn screening
panel^LN|||201010141853|||VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.
113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201101051142|||F

OBX|1|CE|62318-1^Nieman Pick disease A/B  newborn screen interpretation^LN||LA6626-
1^Normal^LN|||N|||F

OBX|2|TX|62319-9^Nieman Pick disease A/B 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|62316-5^Acid sphingomyelinase [Enzymatic activity/volume] in Dried blood
spot^LN||3.3|umol/L/h|>1.0|N|||F
```

Note: This is the panel for Pompe disease:

```
OBR|25|128993^ST ELSEWHERE HOSPITAL^9999999999^NPI|999555^TNSPHLAB^77D7777777^CLIA|63414-
7^Pompe disease newborn screening
panel^LN|||201010141853|||VH||||201010151121||1111111111^Smiles^Minnie^^^Dr^^^NPI&2.16.840.1.
113883.4.6&ISO^L ^^^NPI^^^^^^^^MD||||||201101051142|||F
```

```
OBX|1|CE|63415-4^Pompe disease newborn screening interpretation^LN||LA6626-
1^Normal^LN|||N|||F

OBX|2|TX|63416-2^Pompe 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|3|NM|55827-0^Acid alpha glucosidase [Enzymatic activity/volume] in Dried blood
spot^LN||6.6|umol/L/h|>4.0|N|||F
```

Section 4.12 SCID Panel

The severe combined immunodeficiency (SCID) panel is a new addition to the LOINC AHIC panel. SCID is the newest condition to be added to the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended uniform screening panel, and several states are currently piloting the screening assay. The SCID panel includes codes for the quantitative TREC assay, test interpretation and comment/discussion.

```
OBR|26|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.
113883.4.6&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
```

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, namesand 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/.

Link For Updates and Additional Information:

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. LHNCBC, in collaboration with other agencies and organizations, also created and maintains the Newborn Screening Coding and Terminology Guide: http://newbornscreeningcodes.nlm.nih.gov/. Please visit this Web site to obtain updates to this example HL7 message, a catalog of NBS-related LOINC and SNOMED CT codes and answer lists, and other guidance and resources. If you have comments or questions, please contact us at: NewbornScreeningCodes@nlm.nih.gov