Genetic Disease Screening Program

Electronic Newborn Screening (NBS) Results

NBS Results Onboarding Program

Contents

Introductions: Key Participants from GDSP

California Department of Public Health (CDPH) *Genetic Disease Screening Program (GDSP)*	
Stan Sciortino	Chief of Program Development and Evaluation (PDE)
Tracey Bishop	Chief of Newborn Screening
Jorge Palacios	Health Program Specialist

Genetic Disease Screening Program (GDSP) Program Overview

Mission

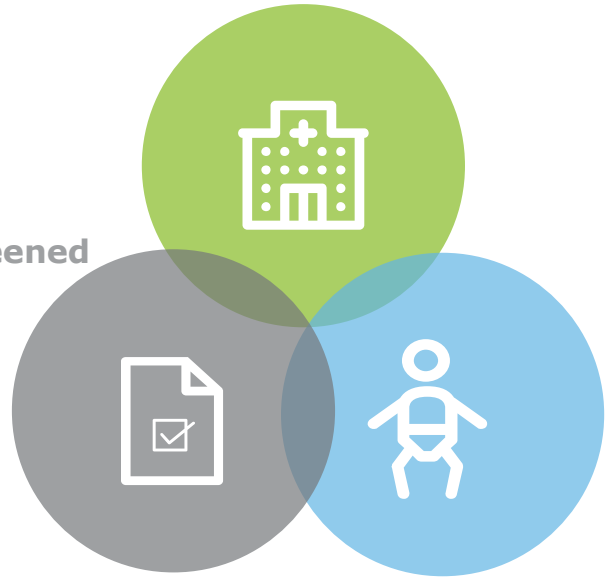
"To serve the people of California by reducing the emotional and financial burden of disability and death caused by genetic and congenital disorders."

Program Overview

- The objective of the Newborn Screening Electronic Results Program is to extend the functionality of the State of California's Screening Information System application to provide Newborn Screening Test Results electronically in HL7 format for healthcare providers.

33 Primary Conditions Screened

- *9 Organic Acid Disorders*
- *5 Fatty Acid Oxidation Disorders*
- *6 Amino Acid Disorders*
- *2 Endocrine Disorders*
- *3 Hemoglobin Disorders*
- *9 Other Disorders*



11 Hospital Organizations Partnering

Hospital Name	# of Births(2020)
Cedar-Sinai Medical Center	6061
Community Hospitals of Monterey Peninsula	1038
Contra Costa Regional Medical Center	1769
Dignity Health	37133
Family Health Centers of San Diego	0
Kern Medical Center	2274
Loma Linda University Health	3582
North Bay Medical Center	1215
Rady Children's Hospital (San Diego)	656
Santa Clara Valley Medical Center	4073
Stanford Medical Center	5863

420,000 Newborns Screened per year

For over 80 genetic and congenital disorders, and over 350,000 pregnant women for down syndrome, trisomy 18 and neural tube defects

Based on 2020 metrics, California represents about **12%** of all births in the United States.

Updated: 12/10/2021

Electronic Results: Project Overview

Key Principles

Offer a more efficient and timely option for obtaining Newborn Screening results

Reduce dependencies associated with delivery via paper mailers

Adhere to new national standards for the secure transmission of health information

Potential Benefits

- Expedited receipt of Newborn Screening results
- Reduction of manual processes of scanning of paper results
- Increased traceability of result received
- Direct integration with provider organization's EHR or LIS
- Enhanced data quality through discrete data transmission
- Improved security through direct electronic integration
- Ability to send HL7 with PDF attachment

Electronic Results: Overall Partner Onboarding Process

The *Electronic Results Onboarding* process will follow the phases outlined below. GDSP will support onboarding provider organizations throughout the implementation process.

	Project Phase	Key Activities	Activity Owner
1	**Initiate Project**	• Obtain necessary project approvals • Conduct Kick-Off discussions & outline the Onboarding Process. Share necessary documentation	➢ **Partner Organization** ➢ **GDSP**
2	**Finalize Design**	• Create interface design and process for consuming HL7 results • Provide Online/Offline Technical and Functional guidance via Meeting Cadence/E-mail	➢ **Partner Organization** ➢ **GDSP**
3	**Perform Development & Internal Testing**	• Utilize onboarding documents, tools for internal testing • Provide Online/Offline Technical and Functional guidance via Meeting Cadence/E-mail	➢ **Partner Organization** ➢ **GDSP**
4	**Perform Interface Testing**	• Build and transmit HL7 results messages for test (sample) cases over a test interface • Read and process sample HL7 messages, return ACK/NACK files	➢ **Partner Organization** ➢ **GDSP**
5	**Go-Live**	• Begin transmitting HL7 results and monitor interface performance • Begin receiving electronic results and monitor interface performance	➢ **Partner Organization** ➢ **GDSP**
6	**Post Go-Live Support**	• Continue monitoring and report any issues observed • Provide support as needed to resolve issues	➢ **Partner Organization** ➢ **GDSP**

Legend **Partner Organization Activity**
GDSP Activity

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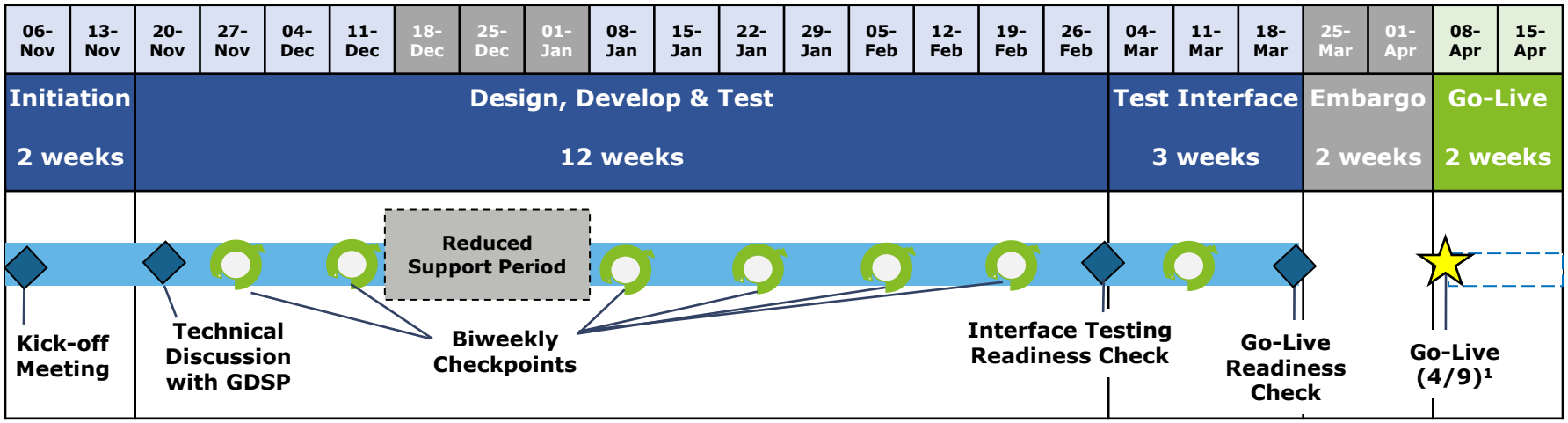
Electronic Results: Implementation Schedule

The *Electronic Results Onboarding* (Wave 4) is planned to follow the schedule outlined below. The durations specified are reflective of a typical onboarding schedule



Initiation

Key Activities:

- Project Planning
- Review HL7 Design
- Submit Consent form

Design, Develop & Test

Key Activities:

- Finalize Design
- Develop and test interface for HL7 results processing
- Biweekly checkpoints
- Partner Organization get access to interface folders (SFTP) – needed for interface testing & go-live

Test Interface

Key Activities:

- GDSP transmits sample HL7 result messages using test orders data
- Partner Organization process HL7 results and send ACK/NACK messages to GDSP

Go-Live

Key Activities:

- Go-Live
- Post Go-Live monitoring for issues by Partner Organization (and GDSP)

Event Milestone Go-Live Meeting Cadence

1 Go-Live is done a day prior (4/8). However, GDSP will begin publishing HL7 results messages the following day (4/9).

Updated: 12/10/2021

Key Meetings during the Onboarding Process (1 of 2)

Meeting Name	Project Phase	Project Timeframe	Description	Frequency	Mandatory?	Duration
Onboarding Kick-Off Discussion	**Initiate Project**	**Week 1**	This meeting is intended to level set on the overall process for onboarding with CDPH for receiving NBS results electronically	Once	Yes	1 Hour
Technical Consultation	**Finalize Design**	**Week 3 - 4**	This meeting is organized on the request of the partner orgranization. The partner team drives the agenda, and it is aimed at helping the partner team implementing the interface with guidance on any techical aspects of the implementation. Conversation topics are focused on the interface and HL7 design	Once	No (scheduled upon Partner's request)	0.5 - 1 Hour
Biweekly Checkpoints	**Finalize Design, Develop and Test, Interface Testing**	**Week 3 - 15**	This standing meeting occurs during the core of implementation and testing. It is aimed at GDSP and Partner onboarding teams to discuss - 1. Any issues, concerns or challenges with the implementation 2. Overall progress against timelines, and risks if any 3. Any opportunities for improvement identified during the implementation (or planning)	Every two weeks	Yes (unless both teams agree to cancel)	0.5 Hours

Apart from the above, for any general questions, issues or feedback during the onboarding process, partner organizations should email nbshie@cdph.ca.gov.

Updated: 12/10/2021

Key Meetings during the Onboarding Process (2 of 2)

Meeting Name	Project Phase	Project Timeframe	Description	Frequency	Mandatory?	Duration
Interface Testing Readiness Checkpoint	**Develop and Test**	**Week 13 - 14**	This meeting is planned to discuss the readiness from a partner perspective for initiating interface testing. Key agenda items include - 1. Status of development and (system) testing completion 2. Access to test interface folders, required for sending/accessing test HL7s between GDSP and the partner system 3. Test data requirements from partner for building and transmitting sample HL7 messages	Once	Yes (can be done as part of a biweekly checkpoint meeting, or email if needed)	0.5 Hours
Go-Live Readiness Checkpoint	**Interface Testing**	**Week 16 - 17**	This meeting is planned to discuss the overall readiness from a GDSP and partner perspective for the go-live. Key agenda items include - 1. Status of interface testing completion 2. Access to production interface folders, required for sending/accessing test HL7s between GDSP and the partner system 3. Any pending form(s) or administrative actions from either side 4. Discuss the process of go-live and any post go-live monitoring activities	Once	Yes (can be done as part of a biweekly checkpoint meeting, or email if needed)	0.5 Hours

Apart from the above, for any general questions, issues or feedback during the onboarding process, partner organizations should email nbshie@cdph.ca.gov.

Onboarding: Driving to a Successful Implementation

Based on past experience gained from provider partner onboarding, the program recommends the following key items, in order to prevent major issues or challenges and successfully complete the integration effort

1. Defining a plan upfront that aligns with the high-level project timeline shown earlier with clear milestones established for each phase. This will help drive the work more effectively and gauge risks as the project progresses.
    - *The GDSP team will request this plan/information as part of starting the onboarding process.*

2. Having an engaged Project sponsorship & leadership during the course of the onboarding implementation. This will help manage risks and/or competing priorities, and help keep the partner team focused as they design, develop and test the new interface.
    - *This should also be shared with GDSP at the beginning of the onboarding process.*

3. Utilize the biweekly recurring & other meetings during the onboarding process. This will help get any questions answered on time, or technical conversations needed to progress the interface work & keep progress on track against the plan. In addition, these will serve as checkpoints where we collectively assess the project health and any key risks to the timeline.

Key Next Steps (Post Kick-off Meeting)

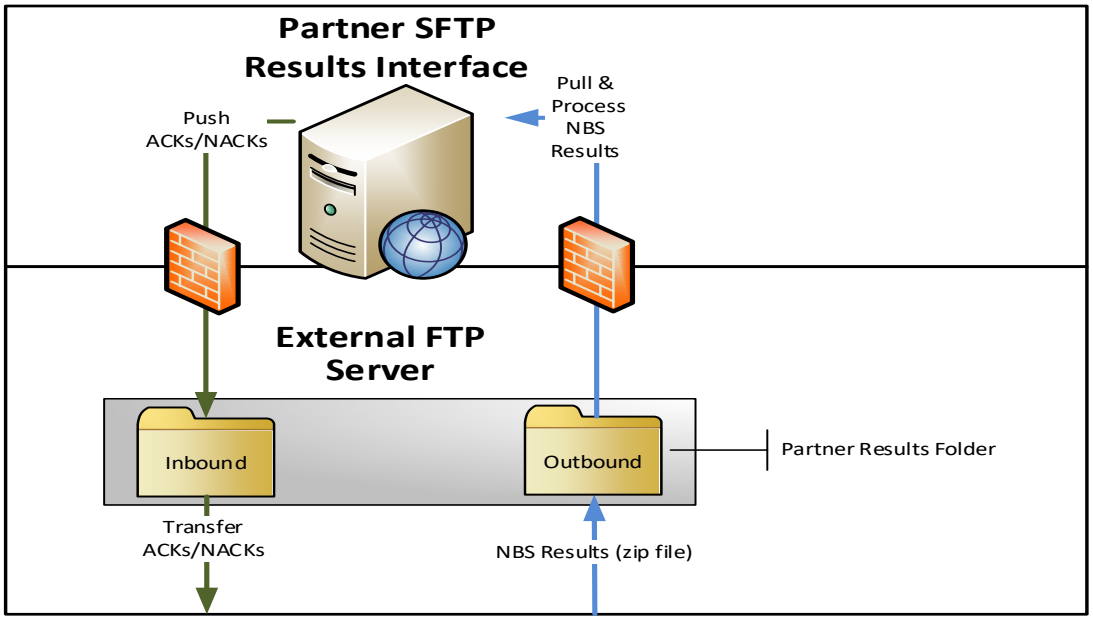
1. **GDSP team to send the following documents via email**: *(ETA within 1-2 days of Kick-off meeting)*
   - Onboarding (Kickoff meeting) overview deck
   - CDPH Website for onboarding support documents
   - Newborn screening Results - HL7 Interface Design Documents
   - Sample Timeline for the onboarding project – *to be completed by partner organization*
   - Hospitals/Health Practice Management (HPM) Firm Consent Form (A form for hospitals/HPM to enroll into GDSP Electronic Results project and receive NBS test results electronically) - *to be completed by partner organization*

2. **Electronic Partner Organization to take the following actions**: (*ETA – Within 2 weeks of Kick-off meeting to prevent onboarding delays)*
   - Complete and return the Hospitals/Health Practice Management (HPM) Firm Consent Form with authorizing signatures. The form can be emailed back to the Onboarding Coordinator at nbshie@gdsp.ca.gov.
   - Complete the high-level timeline view and email back to the Onboarding Coordinator at nbshie@gdsp.ca.gov. This should include preferred dates/times for *ETA – Within 2 weeks of Kick-off meeting*
     - Technical Consultation/Discussion (as needed).
     - Ongoing Biweekly Checkpoint meetings

3. Once received, GDSP will review the documents sent by the Partner Organization and provide approval. In addition, GDSP onboarding team will setup the meetings (technical consultation and biweekly checkpoints) and publish the meeting invites.

APPENDIX

Newborn Screening (NBS): HL7 Results Messaging Flow



Sending Newborn Screening Results to Electronic Results Partner:

1. SIS GDSP will place NBS results in HL7 v.2.5.1 messages in a **zip file**, in a specified folder, on the Outbound directory of the secure FTP server
2. Electronic Results Partner will **pull** the zipped HL7 messages from the secure FTP server and unzip them for HL7 message translation
3. Electronic Results Partner will **translate** the HL7 messages and process into their EHR

Receiving Acknowledgments (ACK) and Error (NACK) messages at SIS GDSP:

1. Electronic Results Partner will **create ACK/NACK HL7 messages** for each NBS result message it processed
2. Electronic Results Partner will **zip the ACK/NACK messages** and **push** the zipped file to a specified folder in the Inbound directory of the secure FTP server
3. SIS GDSP will **translate** the ACK/NACK messages from the secure FTP server for processing

HL7 Message Format

Below are key details regarding the HL7 message format:

- **HL7 Version:** HL7 v2.5.1
- **Standard:** Public Health Informatics Institute (PHII) and U.S. National Library of Medicine (NLM)
  - PHII website: http://www.phii.org/
  - NLM website: http://newbornscreeningcodes.nlm.nih.gov/nb/sc/constructingNBSHL7messages
- **Medium:** Secure File Transfer Protocol through an external server

  Note: Each HIE Receiver will have a dedicated folder, for both the Inbound and the Outbound Directory
- **Frequency:** Sent daily, excluding Saturdays and State Holidays
- **File Format:** HL7 messages to be zipped and place on both Inbound and Outbound Directory of secure FTP server

Below are details for each message type (Newborn Screening results & Acknowledgement/Error message):

- **Newborn Screening Results:**
  - **Message Type:** ORU^R01 (Results)
  - **Timing:** Electronic Test Results will be placed in respective Outbound directories at **9:00 PM PST**
- **Acknowledgments / Error Messages:**
  - **Message Type**: ACK^R01^ACK (Acknowledgments)
  - **Timing:** ACK messages should be placed in respective Inbound directories by **4:00 AM PST**

Electronic Results: Additional Features Available to Partner Organizations

HL7 with PDF 'attachments':

- This is an opt-in functionality in addition to the traditional HL7 results message that allows Partner organizations to receive results in PDF format (encoded) within the HL7 message
- This is activated upon request from the Partner organization to CDPH, and effective the next day once the configurations are made on the CDPH side

HIE Online Tool

The HIE online tool allows partner organizations to generate various 'test' HL7 results messages for different newborn screening test combinations. In addition, it has a feature for validation of ACK/NACK files generated by the partner's system. There are two versions

- Version 1 – This is based on the HL7 design currently in production. https://hiegateway.cdph.ca.gov/GDSPHL7Tools/
- Version 2 – This contains upcoming HL7 design changes that are in flight. This allows partner organizations to generate and test sample HL7s prior to production release. https://hiegatewaystag.cdph.ca.gov/GDSPHL7Tools/

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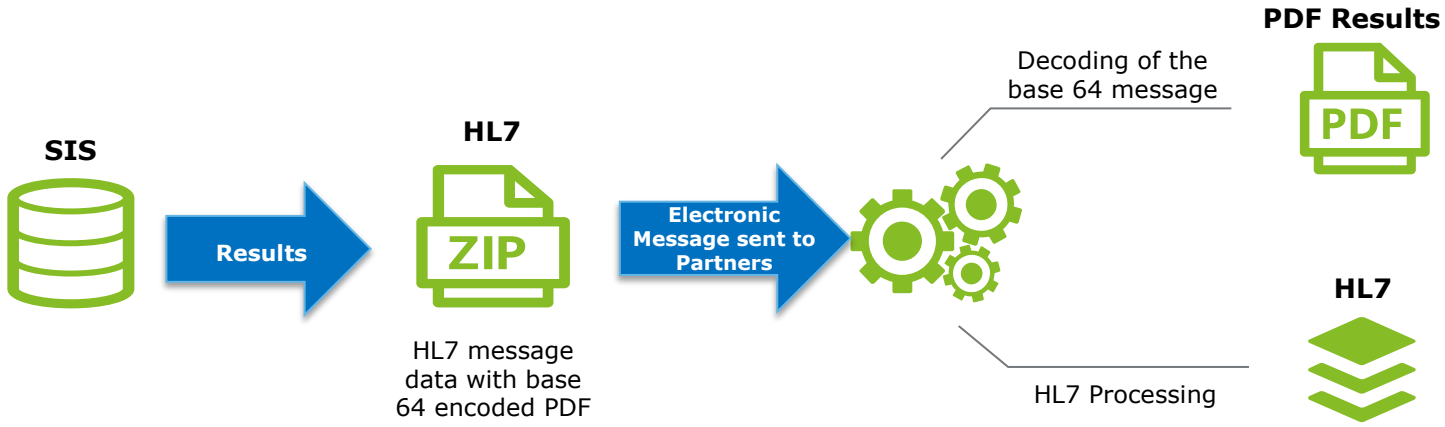
PDF Image Data in HL7 message

An enhancement was developed to attach the PDF version of the results mailer to the HL7. This is functionality in addition to the HL7 electronic message that the results partners can opt in to receive in addition to the electronic message.

PDF Attachment to HL7



The optional PDF Attachment enables partners to receive the data to create the PDF mailer image in addition to the HL7 message. This provides an alternative view of the HL7 results that mimics the paper mailer and is considered more easily readable by users. To enable this functionality, the partners need to decode the base 64 encoded PDF data to display the image.

*To opt into this functionality, please initiate conversation by contacting **nbshie@cdph.ca.gov**.*

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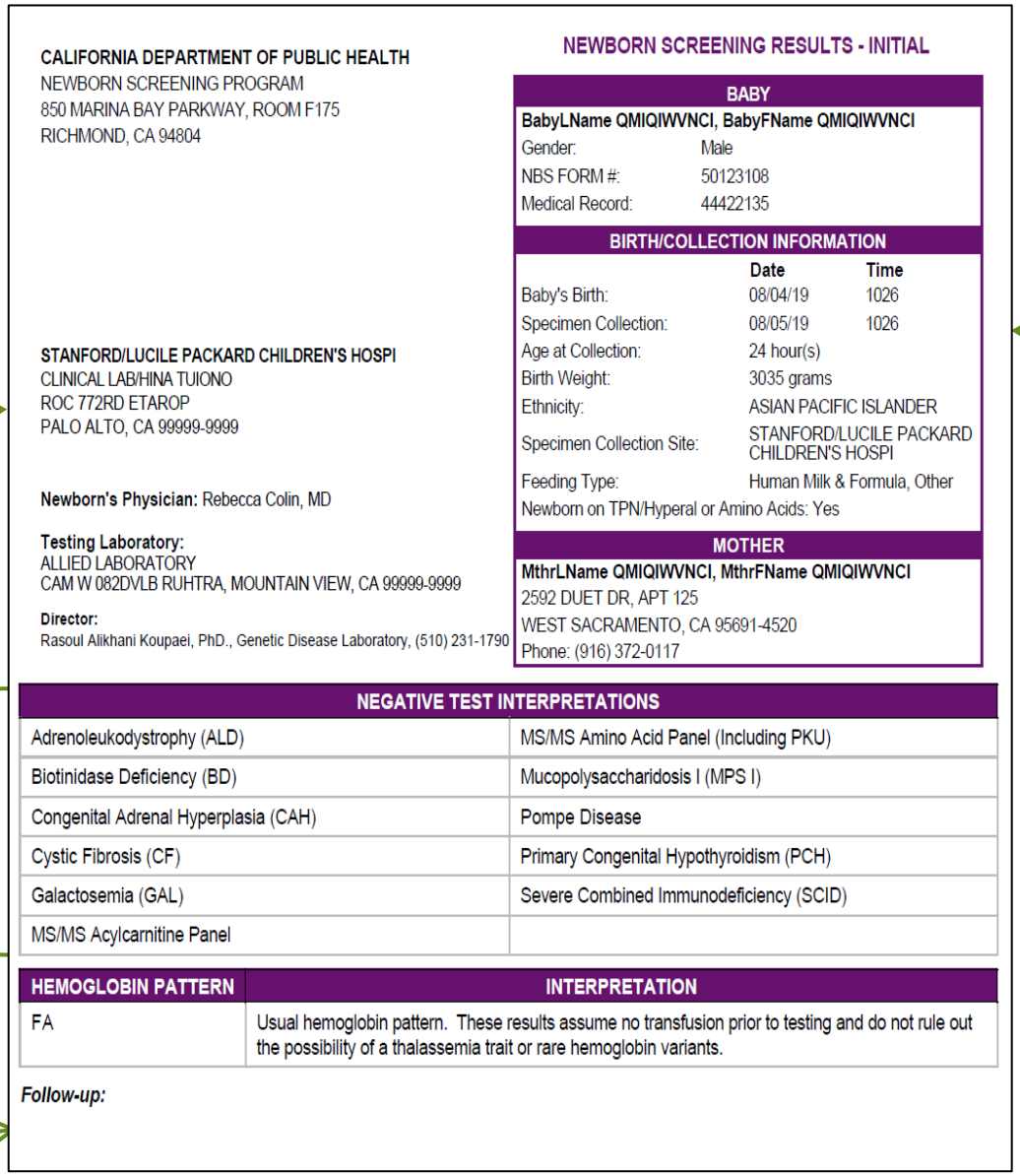
Key Terms and Acronyms

- **SIS:** State of CA's Screening Information System application
- **HL7:** Health Level 7 messaging standard that enables clinical applications to exchange data; "book of rules" detailing interfacing information that sets forth a framework for negotiation in interfacing
- Coding Systems
  - **LOINC –** Logical Observation Identifiers Names and Codes (LOINC®): provides standardized codes for the questions in lab results messages; used for ordering and resulting of lab tests; preferred code set for HL7 messages
  - **SNOMED –** Standardized terminology for clinical data for diseases, clinical findings and procedures
- **Health Information Technology Standards Panel (HITSP)** Newborn Screening Interoperability Specification: messaging standards specific to NBS

HL7 Message Structure: *PDF Results to HL7 Results Sample Mapping (1 of 2)*



CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
NEWBORN SCREENING PROGRAM
850 MARINA BAY PARKWAY, ROOM F175
RICHMOND, CA 94804

NEWBORN SCREENING RESULTS - INITIAL

BABY

BabyLName QMIQIWVNCI, BabyFName QMIQIWVNCI

Gender:	Male
NBS FORM #:	50123108
Medical Record:	44422135

BIRTH/COLLECTION INFORMATION

	Date	Time
Baby's Birth:	08/04/19	1026
Specimen Collection:	08/05/19	1026
Age at Collection:	24 hour(s)	
Birth Weight:	3035 grams	
Ethnicity:	ASIAN PACIFIC ISLANDER	
Specimen Collection Site:	STANFORD/LUCILE PACKARD CHILDREN'S HOSPI	
Feeding Type:	Human Milk & Formula, Other	

Newborn on TPN/Hyperal or Amino Acids: Yes

MOTHER

MthrLName QMIQIWVNCI, MthrFName QMIQIWVNCI
2592 DUET DR, APT 125
WEST SACRAMENTO, CA 95691-4520
Phone: (916) 372-0117

STANFORD/LUCILE PACKARD CHILDREN'S HOSPI
CLINICAL LAB/HINA TUIONO
ROC 772RD ETAROP
PALO ALTO, CA 99999-9999

Newborn's Physician: Rebecca Colin, MD

Testing Laboratory:
ALLIED LABORATORY
CAM W 082DVLB RUHTRA, MOUNTAIN VIEW, CA 99999-9999

Director:
Rasoul Alikhani Koupaei, PhD., Genetic Disease Laboratory, (510) 231-1790

NEGATIVE TEST INTERPRETATIONS	
Adrenoleukodystrophy (ALD)	MS/MS Amino Acid Panel (Including PKU)
Biotinidase Deficiency (BD)	Mucopolysaccharidosis I (MPS I)
Congenital Adrenal Hyperplasia (CAH)	Pompe Disease
Cystic Fibrosis (CF)	Primary Congenital Hypothyroidism (PCH)
Galactosemia (GAL)	Severe Combined Immunodeficiency (SCID)
MS/MS Acylcarnitine Panel	

HEMOGLOBIN PATTERN	INTERPRETATION
FA	Usual hemoglobin pattern. These results assume no transfusion prior to testing and do not rule out the possibility of a thalassemia trait or rare hemoglobin variants.

Follow-up:

Patient Information
(PID)

NBS Screen Card
(OBR| 3)

Next of Kin
(NK1)

Residing Clinician
(MSH.6)

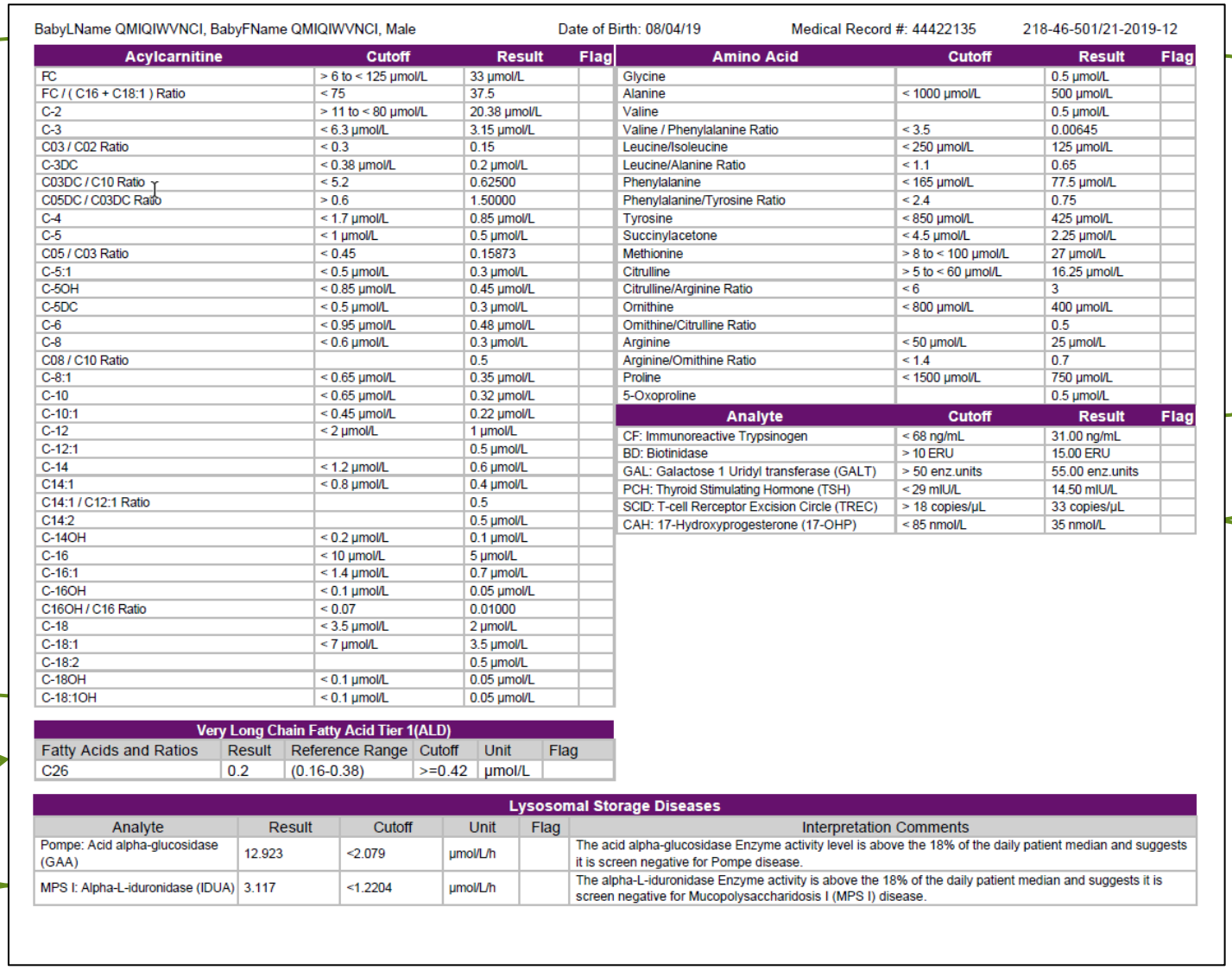
Conditions Tested & Positive Markers
(OBR|2)

Hemoglobinopathies Panel
(OBR|13)

Newborn Screening Short Summary
(OBR|2)

Updated: 12/10/2021

HL7 Message Structure: *PDF Mailer to HL7 Sample Mapping (2 of 2)*



BabyLName QMIQIWVNCI, BabyFName QMIQIWVNCI, Male Date of Birth: 08/04/19 Medical Record #: 44422135 218-46-501/21-2019-12

Acylcarnitine	Cutoff	Result	Flag
FC	> 6 to < 125 µmol/L	33 µmol/L	
FC / (C16 + C18:1) Ratio	< 75	37.5	
C-2	> 11 to < 80 µmol/L	20.38 µmol/L	
C-3	< 6.3 µmol/L	3.15 µmol/L	
C03 / C02 Ratio	< 0.3	0.15	
C-3DC	< 0.38 µmol/L	0.2 µmol/L	
C03DC / C10 Ratio	< 5.2	0.62500	
C05DC / C03DC Ratio	> 0.6	1.50000	
C-4	< 1.7 µmol/L	0.85 µmol/L	
C-5	< 1 µmol/L	0.5 µmol/L	
C05 / C03 Ratio	< 0.45	0.15873	
C-5:1	< 0.5 µmol/L	0.3 µmol/L	
C-5OH	< 0.85 µmol/L	0.45 µmol/L	
C-5DC	< 0.5 µmol/L	0.3 µmol/L	
C-6	< 0.95 µmol/L	0.48 µmol/L	
C-8	< 0.6 µmol/L	0.3 µmol/L	
C08 / C10 Ratio		0.5	
C-8:1	< 0.65 µmol/L	0.35 µmol/L	
C-10	< 0.65 µmol/L	0.32 µmol/L	
C-10:1	< 0.45 µmol/L	0.22 µmol/L	
C-12	< 2 µmol/L	1 µmol/L	
C-12:1		0.5 µmol/L	
C-14	< 1.2 µmol/L	0.6 µmol/L	
C14:1	< 0.8 µmol/L	0.4 µmol/L	
C14:1 / C12:1 Ratio		0.5	
C14:2		0.5 µmol/L	
C-14OH	< 0.2 µmol/L	0.1 µmol/L	
C-16	< 10 µmol/L	5 µmol/L	
C-16:1	< 1.4 µmol/L	0.7 µmol/L	
C-16OH	< 0.1 µmol/L	0.05 µmol/L	
C16OH / C16 Ratio	< 0.07	0.01000	
C-18	< 3.5 µmol/L	2 µmol/L	
C-18:1	< 7 µmol/L	3.5 µmol/L	
C-18:2		0.5 µmol/L	
C-18OH	< 0.1 µmol/L	0.05 µmol/L	
C-18:1OH	< 0.1 µmol/L	0.05 µmol/L	

Amino Acid	Cutoff	Result	Flag
Glycine		0.5 µmol/L	
Alanine	< 1000 µmol/L	500 µmol/L	
Valine		0.5 µmol/L	
Valine / Phenylalanine Ratio	< 3.5	0.00645	
Leucine/Isoleucine	< 250 µmol/L	125 µmol/L	
Leucine/Alanine Ratio	< 1.1	0.65	
Phenylalanine	< 165 µmol/L	77.5 µmol/L	
Phenylalanine/Tyrosine Ratio	< 2.4	0.75	
Tyrosine	< 850 µmol/L	425 µmol/L	
Succinylacetone	< 4.5 µmol/L	2.25 µmol/L	
Methionine	> 8 to < 100 µmol/L	27 µmol/L	
Citrulline	> 5 to < 60 µmol/L	16.25 µmol/L	
Citrulline/Arginine Ratio	< 6	3	
Ornithine	< 800 µmol/L	400 µmol/L	
Ornithine/Citrulline Ratio		0.5	
Arginine	< 50 µmol/L	25 µmol/L	
Arginine/Ornithine Ratio	< 1.4	0.7	
Proline	< 1500 µmol/L	750 µmol/L	
5-Oxoproline		0.5 µmol/L	

Analyte	Cutoff	Result	Flag
CF: Immunoreactive Trypsinogen	< 68 ng/mL	31.00 ng/mL	
BD: Biotinidase	> 10 ERU	15.00 ERU	
GAL: Galactose 1 Uridyl transferase (GALT)	> 50 enz.units	55.00 enz.units	
PCH: Thyroid Stimulating Hormone (TSH)	< 29 mIU/L	14.50 mIU/L	
SCID: T-cell Rerceptor Excision Circle (TREC)	> 18 copies/µL	33 copies/µL	
CAH: 17-Hydroxyprogesterone (17-OHP)	< 85 nmol/L	35 nmol/L	

Very Long Chain Fatty Acid Tier 1(ALD)

Fatty Acids and Ratios	Result	Reference Range	Cutoff	Unit	Flag
C26	0.2	(0.16-0.38)	>=0.42	µmol/L	

Lysosomal Storage Diseases

Analyte	Result	Cutoff	Unit	Flag	Interpretation Comments
Pompe: Acid alpha-glucosidase (GAA)	12.923	<2.079	µmol/L/h		The acid alpha-glucosidase Enzyme activity level is above the 18% of the daily patient median and suggests it is screen negative for Pompe disease.
MPS I: Alpha-L-iduronidase (IDUA)	3.117	<1.2204	µmol/L/h		The alpha-L-iduronidase Enzyme activity is above the 18% of the daily patient median and suggests it is screen negative for Mucopolysaccharidosis I (MPS I) disease.

Acylcarnitine Panel (OBR|6)
Fatty Acid Panel (OBR|7)
Organic Acid Panel (OBR|8)

ALD Tier-1 Panel (OBR|19)

Pompe Panel (OBR|16)

MPS I Panel (OBR|17)

Amino Acid Panel (OBR|5)

Cystic Fibrosis Panel (OBR|9)

Biotinidase Panel (OBR|14)

Galactosemia Panel (OBR|12)

PCH Panel (OBR|11)

SCID Panel (OBR|15)

CAH Panel (OBR|10)

Updated: 12/10/2021

HL7 Results Sample Message *(1 of 3)*

Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results

```
MSH|^~\&|SISGDSP|SISGDSP|SISHIERECEIVER^9414049^L,M,N|^^L,M,N|20190802053800||ORU^R01^ORU_R01|235805324|T|2.5.1
PID|1||44422135^^^&NPI^MR||BabyLName GSWERUXYOO^BabyFName GSWERUXYOO^^^^^B||201907221026|M||2131-1^Other Race||||||||||2186-5^Not Hispanic or Latino||N|1
NK1|1|MthrLName GSWERUXYOO^MthrFName GSWERUXYOO|MTH^Mother|2592 DUET DR, APT 125^^WEST SACRAMENTO^CA^95691-4520^USA|^^^^^916^3720117||||||||||19920724
ORC|RE|4012149713^FormNumber|||||||||^Colin^Rebecca|||||||STANFORD/LUCILE PACKARD CHILDREN'S HOSPI^^^^^^^^^R777|ROC 772RD ETAROP^^PALO ALTO^CA^99999-9999
OBR|1|4012149713^FormNumber||54089-8^NB Screen Panel Patient AHIC|||201907231026|||||||||||||20190802053800|||F
OBR|2|4012149713^FormNumber||57128-1^Newborn Screening Report summary panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|CE|57721-3^Reason for lab test in Dried blood spot^LN|1|LA12421-6\S\Initial screen\S\LN|||N|||F|||20190802053800
OBX|2|CE|57718-9^Sample quality of Dried blood spot^LN|1|LA12432-3\S\Acceptable\S\LN|||N|||F|||20190802053800
OBX|3|CE|57130-7^Newborn screening report - overall interpretation^LN|1|LA12428-1\S\All screening is in range for the conditions tested\S\LN|||N|||F|||20190802053800
OBX|4|CE|57131-5^Newborn conditions with positive markers [Identifier] in Dried blood spot^LN|1|LA137-2\S\None\S\LN|||N|||F|||20190802053800
OBX|5|CE|57720-5^Newborn conditions with equivocal markers [Identifier] in Dried blood spot^LN|1|LA137-2\S\None\S\LN|||N|||F|||20190802053800
OBX|6|TX|57724-7^Newborn screening short narrative summary^LN|1|NBS Testing Lab - ALLIED LABORATORY \R\CAM W 082DVLB RUHTRA, MOUNTAIN VIEW, CA 99999-9999\.br\\.br\Genetic Disease Laboratory - GDL B W 9042TS YAWDAOR, RICHMOND,
CA 99999-9999\.br\\.br\Lab Director - Rasoul Alikhani Koupaei, PhD., Genetic Disease Laboratory, (510) 231-1790\.br\\.br\If you have questions regarding these results, please contact the Newborn Screening staff at Stanford University
Medical Center, (510) 412-1562. \.br\\.br\Disclaimer:\.br\\.br\Testing for ALD Tier-1,ALD Tier-2, Pompe Tier-1 and MPS I Tier-1 was developed and its performance characteristics determined by the Genetic Disease Laboratory.
It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded
as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity testing. The ALD Tier-1, ALD Tier-2, Pompe Tier-1
and MPS I Tier-1 testing was run at the California Department of Public Health Genetic Disease Screening Laboratory. \.br\Due to biological variability of newborns and differences in detection rates for the various disorders
in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative
screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. A newborn screening result should not be considered
diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider. \.br\\.br\|||N|||F|||20190802053800
OBX|7|TX|57129-9^Full newborn screening summary report for display or printing^LN|1|||N|||F|||20190802053800
OBX|8|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|1|LA25796-6\S\X-ALD\S\LN|||N|||F|||20190802053800
OBX|9|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|2|LA14037-8\S\GAA\S\LN|||N|||F|||20190802053800
OBX|10|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|3|LA25797-4\S\MPS-I\S\LN|||N|||F|||20190802053800
OBX|11|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|4|LA12466-1\S\3-MCC\S\LN|||N|||F|||20190802053800
OBX|12|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|5|LA12468-7\S\3MGA\S\LN|||N|||F|||20190802053800
OBX|13|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|6|LA12469-5\S\5-OXO\S\LN|||N|||F|||20190802053800
OBX|14|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|7|LA12470-3\S\ARG\S\LN|||N|||F|||20190802053800
OBX|15|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|8|LA12482-8\S\CIT-I\S\LN|||N|||F|||20190802053800
OBX|16|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|9|LA12483-6\S\CIT-II\S\LN|||N|||F|||20190802053800
OBX|17|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|10|LA12485-1\S\CPT-Ia\S\LN|||N|||F|||20190802053800
OBX|18|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|11|LA12486-9\S\CPT-II\S\LN|||N|||F|||20190802053800
OBX|19|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|12|LA12493-5\S\GA-1\S\LN|||N|||F|||20190802053800
OBX|20|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|13|LA12495-0\S\GA-2\S\LN|||N|||F|||20190802053800
OBX|21|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|14|LA12497-6\S\HHH\S\LN|||N|||F|||20190802053800
OBX|22|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|15|LA12499-2\S\HMG\S\LN|||N|||F|||20190802053800
OBX|23|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|16|LA12505-6\S\IVA\S\LN|||N|||F|||20190802053800
OBX|24|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|17|LA12507-2\S\LCHAD\S\LN|||N|||F|||20190802053800
OBX|25|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|18|LA12508-0\S\MAL\S\LN|||N|||F|||20190802053800
OBX|26|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|19|LA12509-8\S\MCAD\S\LN|||N|||F|||20190802053800
OBX|27|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|20|LA12510-6\S\MCD\S\LN|||N|||F|||20190802053800
OBX|28|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|21|LA12512-2\S\MET\S\LN|||N|||F|||20190802053800
OBX|29|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|22|LA12513-0\S\MSUD\S\LN|||N|||F|||20190802053800
OBX|30|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|23|LA12516-3\S\NKHG\S\LN|||N|||F|||20190802053800
OBX|31|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|24|LA12520-5\S\PKU\S\LN|||N|||F|||20190802053800
OBX|32|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|25|LA12521-3\S\PRO I\S\LN|||N|||F|||20190802053800
OBX|33|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|26|LA12528-8\S\TYR-1\S\LN|||N|||F|||20190802053800
OBX|34|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|27|LA12529-6\S\TYR-II\S\LN|||N|||F|||20190802053800
OBX|35|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|28|LA12531-2\S\VLCAD\S\LN|||N|||F|||20190802053800
OBX|36|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|29|LA12532-0\S\BIO\S\LN|||N|||F|||20190802053800
OBX|37|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|30|LA12533-8\S\CAH\S\LN|||N|||F|||20190802053800
OBX|38|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|31|LA12537-9\S\CF\S\LN|||N|||F|||20190802053800
OBX|39|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|32|LA12543-7\S\GALT\S\LN|||N|||F|||20190802053800
OBX|40|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|33|LA12566-8\S\SCID\S\LN|||N|||F|||20190802053800
OBX|41|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|34|LA12576-7\S\SCAD or EMA or IBG or GA-2 (MADD)\S\LN|||N|||F|||20190802053800
OBX|42|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|35|LA16207-5\S\Hemoglobinopathies\S\LN|||N|||F|||20190802053800
OBX|43|CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood spot^LN|36|99717-3\S\Hypothyroidism\S\L|||N|||F|||20190802053800
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HL7 Results Sample Message *(2 of 3)*

Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results

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OBR|3|4012149713^FormNumber||57717-1^Newborn screen card data panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|ST|57716-3^State printed on filter paper card [Identifier] in NBS card^LN|1|CA|||N|||F|||20190802053800
OBX|2|NM|8339-4^Birthweight^LN|1|3035|grams||N|||F|||20190802053800
OBX|3|TM|57715-5^Time of birth^LN|1|1026|||N|||F|||20190802053800
OBX|4|CE|57722-1^Birth plurality of Pregnancy^LN|1|LA12411-7\S\Singleton\S\LN|||N|||F|||20190802053800
OBX|5|NM|73806-2^Newborn age in hours^LN|1|24|hour(s)||N|||F|||20190802053800
OBX|6|CE|57713-0^Infant NICU factors that affect newborn screening interpretation^LN|1|LA137-2\S\None\S\LN|||N|||F|||20190802053800
OBX|7|CE|67704-7^Feeding types^LN|1|LA16914-6\S\Breast milk\S\LN|||N|||F|||20190802053800
OBX|8|CE|67704-7^Feeding types^LN|2|LA16915-3\S\Lactose formula\S\LN|||N|||F|||20190802053800
OBX|9|CE|67704-7^Feeding types^LN|3|LA46-8\S\Other\S\LN|||N|||F|||20190802053800
OBX|10|CE|67704-7^Feeding types^LN|4|LA12418-2\S\TPN\S\LN|||N|||F|||20190802053800
OBX|11|TX|^^^99717-5^Accession Number^L|1|205-63-401/21-2019-12|||N|||F|||20190802053800
OBX|12|TX|62324-9^Post-discharge provider name^LN|1|Rebecca Colin|||N|||F|||20190802053800
OBX|13|TX|62327-2^Post-discharge provider practice address^LN|1|ROC 772RD ETAROP  PALO ALTO CA 99999-9999 USA|||N|||F|||20190802053800
OBR|4|4012149713^FormNumber||57794-0^Newborn screening test results panel in Dried blood spot|||201907231026|||||||||||||20190802053800|||F
OBR|5|4012149713^FormNumber||53261-4^Amino acid newborn screen panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|47633-3^Glycine [Moles/volume] in Dried blood spot^LN|1|0.5|µmol/L||N|||F|||20190802053800
OBX|2|NM|53150-9^Alanine+Beta Alanine+Sarcosine [Moles/volume] in Dried blood spot^LN|1|500|µmol/L|<1000|N|||F|||20190802053800
OBX|3|NM|47799-2^Valine [Moles/volume] in Dried blood spot^LN|1|0.5|µmol/L||N|||F|||20190802053800
OBX|4|NM|53151-7^Valine/Phenylalanine [Molar ratio] in Dried blood spot^LN|1|0.00645|{Ratio}|<3.5|N|||F|||20190802053800
OBX|5|NM|53152-5^Alloisoleucine+Isoleucine+Leucine+Hydroxyproline^LN|1|125|µmol/L|<250|N|||F|||20190802053800
OBX|6|NM|53154-1^Alloisoleucine+Isoleucine+Leucine+Hydroxyproline/Alanine [Molar ratio] in Dried blood spot^LN|1|0.65|{Ratio}|<1.1|N|||F|||20190802053800
OBX|7|NM|29573-3^Phenylalanine [Moles/volume] in Dried blood spot^LN|1|77.5|µmol/L|<165|N|||F|||20190802053800
OBX|8|NM|35572-7^Phenylalanine/Tyrosine [Molar ratio] in Dried blood spot^LN|1|0.75|{Ratio}|<2.4|N|||F|||20190802053800
OBX|9|NM|35571-9^Tyrosine [Moles/volume] in Dried blood spot^LN|1|425|µmol/L|<850|N|||F|||20190802053800
OBX|10|NM|53231-7^Succinylacetone [Moles/volume] in Dried blood spot^LN|1|2.25|µmol/L|<4.5|N|||F|||20190802053800
OBX|11|NM|47700-0^Methionine [Moles/volume] in Dried blood spot^LN|1|27|µmol/L|8-100|N|||F|||20190802053800
OBX|12|NM|42892-0^Citrulline [Moles/volume] in Dried blood spot^LN|1|16.25|µmol/L|5-60|N|||F|||20190802053800
OBX|13|NM|54092-2^Citrulline/Arginine [Molar ratio] in Dried blood spot^LN|1|3|{Ratio}|<6|N|||F|||20190802053800
OBX|14|NM|53155-8^Asparagine+Ornithine [Moles/volume] in Dried blood spot^LN|1|400|µmol/L|<800|N|||F|||20190802053800
OBX|15|NM|75215-4^Ornithine/Citrulline [Molar ratio] in Dried blood spot^LN|1|0.5|{Ratio}|N|||F|||20190802053800
OBX|16|NM|47562-4^Arginine [Moles/volume] in Dried blood spot^LN|1|25|µmol/L|<50|N|||F|||20190802053800
OBX|17|NM|75214-7^Arginine/Ornithine [Molar ratio] in Dried blood spot^LN|1|0.7|{Ratio}|<1.4|N|||F|||20190802053800
OBX|18|NM|47732-3^Proline [Moles/volume] in Dried blood spot^LN|1|750|µmol/L|<1500|N|||F|||20190802053800
OBX|19|NM|53232-5^5-Oxoproline+Pipecolate [Moles/volume] in Dried blood spot^LN|1|0.5|µmol/L||N|||F|||20190802053800
OBX|20|TX|57710-6^Amino acidemias newborn screening comment/discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|6|4012149713^FormNumber||58092-8^Acylcarnitine newborn screen panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|CE|58088-6^Acylcarnitine newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|2|TX|58093-6^Acylcarnitine newborn screening comment/discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|7|4012149713^FormNumber||57084-6^Fatty acid oxidation newborn screen panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|38481-8^Carnitine.free (C0)^LN|1|33|µmol/L|6-125|N|||F|||20190802053800
OBX|2|NM|53235-8^Carnitine.free (C0)/Palmitoylcarnitine (C16)+Stearoylcarnitine (C18)^LN|1|37.5|{Ratio}|<75|N|||F|||20190802053800
OBX|3|NM|50157-7^Acetylcarnitine (C2)^LN|1|20.38|µmol/L|11-80|N|||F|||20190802053800
OBX|4|NM|75212-1^Malonylcarnitine (C3-DC)/Decanoylcarnitine (C10) [Molar ratio] in Dried blood spot^LN|1|0.62500|{Ratio}|<5.2|N|||F|||20190802053800
OBX|5|NM|45211-0^Hexanoylcarnitine (C6)^LN|1|0.48|µmol/L|<0.95|N|||F|||20190802053800
OBX|6|NM|53175-6^Octanoylcarnitine (C8)^LN|1|0.3|µmol/L|<0.6|N|||F|||20190802053800
OBX|7|NM|53177-2^Octanoylcarnitine (C8)/Decanoylcarnitine (C10)^LN|1|0.5|{Ratio}||N|||F|||20190802053800
OBX|8|NM|53174-9^Octenoylcarnitine (C8:1)^LN|1|0.35|µmol/L|<0.65|N|||F|||20190802053800
OBX|9|NM|45197-1^Decanoylcarnitine (C10)^LN|1|0.32|µmol/L|<0.65|N|||F|||20190802053800
OBX|10|NM|45198-9^Decenoylcarnitine (C10:1)^LN|1|0.22|µmol/L|<0.45|N|||F|||20190802053800
OBX|11|NM|45199-7^Dodecanoylcarnitine (C12)^LN|1|1|µmol/L|<2|N|||F|||20190802053800
OBX|12|NM|45200-3^Dodecenoylcarnitine (C12:1)^LN|1|0.5|µmol/L||N|||F|||20190802053800
OBX|13|NM|53192-1^Tetradecanoylcarnitine (C14)^LN|1|0.6|µmol/L|<1.2|N|||F|||20190802053800
OBX|14|NM|53191-3^Tetradecenoylcarnitine (C14:1)^LN|1|0.4|µmol/L|<0.8|N|||F|||20190802053800
OBX|15|NM|53194-7^Tetradecenoylcarnitine (C14:1)/Dodecenoylcarnitine (C12:1)^LN|1|0.5|{Ratio}||N|||F|||20190802053800
OBX|16|NM|53190-5^Tetradecadienoylcarnitine (C14:2)^LN|1|0.5|µmol/L||N|||F|||20190802053800
OBX|17|NM|50281-5^3-Hydroxytetradecanoylcarnitine (C14-OH)^LN|1|0.1|µmol/L|<0.2|N|||F|||20190802053800
OBX|18|NM|53199-6^Palmitoylcarnitine (C16)^LN|1|5|µmol/L|<10|N|||F|||20190802053800
OBX|19|NM|53198-8^Palmitoleylcarnitine (C16:1)^LN|1|0.7|µmol/L|<1.4|N|||F|||20190802053800
OBX|20|NM|50125-4^3-Hydroxypalmitoylcarnitine (C16-OH)^LN|1|0.05|µmol/L|<0.1|N|||F|||20190802053800
OBX|21|NM|53201-0^3-Hydroxypalmitoylcarnitine (C16-OH)/Palmitoylcarnitine (C16)^LN|1|0.01000|{Ratio}|<0.07|N|||F|||20190802053800
OBX|22|NM|53241-6^Stearoylcarnitine (C18)^LN|1|2|µmol/L|<3.5|N|||F|||20190802053800
OBX|23|NM|53202-8^Oleoylcarnitine (C18:1)^LN|1|3.5|µmol/L|<7|N|||F|||20190802053800
OBX|24|NM|45217-7^Linoleoylcarnitine (C18:2)^LN|1|0.5|µmol/L||N|||F|||20190802053800
OBX|25|NM|50132-0^3-Hydroxystearoylcarnitine (C18-OH)^LN|1|0.05|µmol/L|<0.1|N|||F|||20190802053800
OBX|26|NM|50113-0^3-Hydroxyoleoylcarnitine (C18:1-OH)^LN|1|0.05|µmol/L|<0.1|N|||F|||20190802053800
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HL7 Results Sample Message *(3 of 3)*

Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results

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OBR|8|4012149713^FormNumber||57085-3^Organic acid newborn screen panel|||20190723102600||||||||||||20190802053800|||F
OBX|1|NM|53160-8^Propionylcarnitine (C3)^LN|1|3.15|µmol/L|<6.3|N|||F|||20190802053800
OBX|2|NM|53163-2^Propionylcarnitine (C3)/Acetylcarnitine (C2)^LN|1|0.15|{Ratio}|<0.3|N|||F|||20190802053800
OBX|3|NM|67708-8^Malonylcarnitine (C3-DC)+3-Hydroxybutyrylcarnitine (C4-OH)^LN|1|0.2|µmol/L|<0.38|N|||F|||20190802053800
OBX|4|NM|53166-5^Butyrylcarnitine+Isobutyrylcarnitine (C4)^LN|1|0.85|µmol/L|<1.7|N|||F|||20190802053800
OBX|5|NM|45216-9^Isovalerylcarnitine+Methylbutyrylcarnitine (C5)^LN|1|0.5|µmol/L|<1|N|||F|||20190802053800
OBX|6|NM|53240-8^Isovalerylcarnitine+Methylbutyrylcarnitine (C5)/Propionylcarnitine (C3)^LN|1|0.15873|{Ratio}|<0.45|N|||F|||20190802053800
OBX|7|NM|53170-7^Tiglylcarnitine (C5:1)^LN|1|0.3|µmol/L|<0.5|N|||F|||20190802053800
OBX|8|NM|50106-4^3-Hydroxyisovalerylcarnitine (C5-OH)^LN|1|0.45|µmol/L|<0.85|N|||F|||20190802053800
OBX|9|NM|67710-4^Glutarylcarnitine (C5-DC)+3-Hydroxyhexanoylcarnitine (C6-OH)^LN|1|0.3|µmol/L|<0.5|N|||F|||20190802053800
OBX|10|NM|75216-2^Glutarylcarnitine (C5-DC)/Malonylcarnitine (C3-DC) [Molar ratio] in Dried blood spot^LN|1|1.50000|{Ratio}|>0.6|N|||F|||20190802053800
OBR|9|4012149713^FormNumber||54078-1^Cystic fibrosis newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|48633-2^Trypsinogen I.free^LN|1|31.00|ng/mL|<68|N|||F|||20190802053800
OBX|2|CE|46769-6^Cystic fibrosis newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|57707-2^Cystic fibrosis newborn screening comment/discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|10|4012149713^FormNumber||57086-1^Congenital adrenal hyperplasia newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|38473-5^17-Hydroxyprogesterone^LN|1|35|nmol/L|<85|N|||F|||20190802053800
OBX|2|CE|46758-9^Congenital adrenal hyperplasia newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|57706-4^Congenital adrenal hyperplasia newborn screening comment-discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|11|4012149713^FormNumber||54090-6^Thyroid newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|29575-8^Thyrotropin^LN|1|14.50|mIU/L|<29|N|||F|||20190802053800
OBX|2|CE|46762-1^Congenital hypothyroidism newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|57705-6^Congenital hypothyroidism newborn screening comment-discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|12|4012149713^FormNumber||54079-9^Galactosemia newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|42906-8^Galactose 1 phosphate uridyl transferase^LN|1|55.00|enzyme units|>50|N|||F|||20190802053800
OBX|2|CE|46737-3^Galactosemias newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|57704-9^Galactosemias newborn screening comment-discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|13|4012149713^FormNumber||54081-5^Hemoglobinopathies newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|TX|54104-5^Hemoglobin pattern^LN|1|FA|||N|||F|||20190802053800
OBX|2|TX|57703-1^Hemoglobin disorders newborn screening comment/discussion^LN|1|Usual hemoglobin pattern. These results assume no transfusion prior to testing and do not rule out the possibility of a thalassemia trait or rare hemoglobin
variants.|||N|||F|||20190802053800
OBR|14|4012149713^FormNumber||57087-9^Biotinidase newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|75217-0^Biotinidase [Enzymatic activity/volume] in Dried blood spot^LN|1|15.00|ERU|>10|N|||F|||20190802053800
OBX|2|CE|46761-3^Biotinidase deficiency newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|57699-1^Biotinidase deficiency newborn screening comment-discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|15|4012149713^FormNumber||62333-0^Severe combined immunodeficiency (SCID) newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|62320-7^T-cell receptor excision circle [#/volume] in Dried blood spot by Probe and target amplification method^LN|1|33|copies/µL|>18|N|||F|||20190802053800
OBX|2|CE|62321-5^Severe combined immunodeficiency newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|62322-3^Severe combined immunodeficiency newborn screening comment-discussion^LN|1|Negative|||N|||F|||20190802053800
OBR|16|4012149713^FormNumber||63414-7^Pompe Disease newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|55827-0^Acid alpha glucosidase [Enzymatic activity/volume] in DBS^LN|1|12.923|µmol/L/h|11.55|N|||F|||20190802053800
NTE|1||Cutoff: 18%
OBX|2|CE|63415-4^Pompe Disease deficiency newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|63416-2^Pompe Disease deficiency newborn screening comments-discussion^LN|1|Negative|||N|||F|||20190802053800
OBX|4|TX|63416-2^Pompe Disease deficiency newborn screening comments-discussion^LN|2|Interpretation Comments: The acid alpha-glucosidase Enzyme activity level is above the 18% of the daily patient median and suggests it is screen negative for
Pompe disease.|||N|||F|||20190802053800
OBR|17|4012149713^FormNumber||79563-3^Mucopolysaccharidosis type I newborn screening panel|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|55909-6^Alpha-L-iduronidase [Enzymatic activity/volume] in DBS^LN|1|3.117|µmol/L/h|6.78|N|||F|||20190802053800
NTE|1||Cutoff: 18%
OBX|2|CE|79564-1^Mucopolysaccharidosis type I newborn screen interpretation^LN|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|3|TX|79565-8^Mucopolysaccharidosis type I newborn screening comment-discussion^LN|1|Negative|||N|||F|||20190802053800
OBX|4|TX|79565-8^Mucopolysaccharidosis type I newborn screening comment-discussion^LN|2|Interpretation Comments: The alpha-L-iduronidase Enzyme activity is above the 18% of the daily patient median and suggests it is screen negative for
Mucopolysaccharidosis I (MPS I) disease.|||N|||F|||20190802053800
OBR|18|4012149713^FormNumber||^^^99717-28^Adrenoleukodystrophy newborn screening panel^L|||201907231026|||||||||||||20190802053800|||F
OBX|1|CE|^^^99717-32^Adrenoleukodystrophy deficiency newborn screening interpretation^L|1|LA18592-8\S\In range\S\LN|||N|||F|||20190802053800
OBX|2|TX|^^^99717-33^Adrenoleukodystrophy deficiency newborn screening comments-discussion^L|1|Negative|||N|||F|||20190802053800
OBR|19|4012149713^FormNumber||^^^99717-29^Adrenoleukodystrophy Tier-1 newborn screening panel^L|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|79321-6^Lysophosphatidylcholine(26:0) [Moles/volume] in Dried blood spot^LN|1|0.6|µmol/L|0.16-0.38|H|||F|||20190802053800
NTE|1||Cutoff: >=0.42
OBR|20|4012149713^FormNumber||^^^99717-30^Adrenoleukodystrophy Tier-2 newborn screening panel^L|||201907231026|||||||||||||20190802053800|||F
OBX|1|NM|79567-4^Lysophosphatidylcholine(26:0) [Moles/volume] in Dried blood spot by LC/MS/MS^LN|1|0.1|µmol/L|0.04-0.09|N|||F|||20190802053800
NTE|1||Cutoff: >=0.22
```

HL7 Results Sample Message PDF Image Data

Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results with PDF attachment. The segments below are added to the typical HL7 message for electronic results

OBR|20|^FormNumber|||ClinicalPDFReport^Clinical PDF Report 226-95-827/21-2018-21^^ClinicalPDFReport^Clinical PDF Report 226-95-827/21-2018-
21^|||20190119000000|||||||||^ORDGLSTNAME^ORDGFRSTNAME^^^^^^|||||20190117154532|||F
OBX|1|ED|ClinicalPDFReport^Clinical PDF Report 226-95-827/21-2018-21^^ClinicalPDFReport^Clinical PDF Report 226-95-827/21-2018-
21^|||SIS^Image^PDF^Base64^JVBERi0xLjQNCiWys7S1DQolR2VuZXJhdGVkIGJ5IEV4cGVydFBkZiB2OS40LjANCjEgMCBvYmoNCjw8DQovUGFnZXMgMiAwIFINCi9QYWdlTW9kZSAvVXNlTm9uZQ0KL1Bh
Z2VMYXlvdXQgL09uZUNvbHVtbg0KL1R5cGUgL0NhdGFsb2cNCj4+DQoNCmVuZG9iag0KPDwNCi9Db3VudCAyDQovS2lkcyBbMyAwIFIgNCAwIFJdDQovVHlwZSAvUGFnZXMNCj4+DQoNC
mVuZG9iag0KMyAwIG9iag0KPDwNCi9SZXNvdXJjZXMgPDwNCi9YT2JqZWN0IDw8DQovRXhwZXJ0UGRmX2IjZW1IZmhnY2dsaGtvZGVwamlsbWhmYnBuZmhuZXBiIDUgMCBSDQo+Pg0KDQovUHJvY1NI
dCBbL1BERiAvVGV4dCAvSW1hZ2VDXQ0KPj4NCg0KL1BhcmVudCAyIDAgUg0KL01IZGlhQm94IFswIDAgMTM5DQo+Pg0KDQovUGFnZXMgMCBvYmoNCjw8DQovUmVzb3VyY2VzID
NvjzCODQpIbmRzdHJlYW0NCg0KZW5kb2JqDQo3IDAgb2JqDQo8PA0KL0xlbmd0aCAwDQo+Pg0Kc3RyZWFtDQoNCmVuZHN0cmVhbQ0KDQpIbmRvYmoNCjw8DQovUmVzb3VyY2VzID
w8DQovWE9iamVjdCA8PA0KL0V4cGVydFBkZl9pY2VtZWZoZ2NnbGhrb2RlcGppbG1oZmJwbmZobmVwYiA1IDAgUg0KPj4NCg0KL1Byb2NTZXQgWy9QREYgL1RleHQgL0ltYWdlQ10NCj4+DQoNCi9QYXJl
bnQgMiAwIFINCi9NZWRpYUJveCBbMC4wMDAwMCAwLjAwMDAwIDYxMi4wMDAwMCA3OTIuMDAwMDBdDQovVHlwZSAvUGFnZQ0KL0NvbnRlbnRzIFs4IDAgUiA5IDAgUl0NCj4+DQoNCmVuZG9iag0
KOCAwIG9iag0KPDwNCi9GaWx0ZXIgL0ZsYXRlRGVjb2RlDQovTGVuZ3RoIDEzNw0KPj4NCnN0cmVhbQ0KeJxdjDsOwjAQRHtLvsOewKwjf+leeqgokeKs40DsmliC4wMBSxHb7JNm3qDAz0H9RjY/sq7SQnC
GzBncOas9pUVr0dmtolyQjVsFzr4GCt2iUbru4xb+o3VTokMAn94y7A7PQsvj2IfL6ClRilMfpnibeyrXcUoxdCWHmKl0sJ85O3H2AifWMDwNCmVuZHN0cmVhbQ0KDQpIbmRvYmoNCjkgMCBvYmoNCjw8D
QovTGVuZ3RoIDANCj4+DQpzdHJlYW0NCg0KZW5kc3RyZWFtDQoNCmVuZG9iag0KMTAgMCBvYmoNCjw8DQovRGlzcGxheURvY1RpdGxlIGZhbHNlDQovTm9uRnVsbFNjcmVlbkJlaGF2aW9ylC9Vc2VOb
25IDQovRml0V2luZG93IGZ93IGZhbHNlDQovQ2VudGVyV2luZG93IGZhbHNlDQovSGlkZVRvb2xiYXIgZmFsc2UNCi9laWRlV2luZG93VUkgZmFsc2UNCi9laWRlTWVudWJhciBmYWxzZQ0KPj4NCg0KZW5kb2JqD
Qo1IDAgb2JqDQo8PA0KL0ZpbHRlciAvRmxhdGVEZWNvZGUNCi9SZXNvdXJjZXMgPDwNCi9Gb250IDw8DQovRXhwZXJ0UGRmX2NmbW5mZmhwYmZoY2tvbmlwbm9sa2tpbmNobGNrbnBpIDExIDAgUg
0KL0V4cGVydFBkZl9pamFsYW9rbWdua2JrZm1ib2NrYmNvbWRmaW9lbG5ibSAxMiAwIFINCi9QRUlUVEwrQXJpYWxNVCAxMyAwIFINCi9FeHBlcnRQZGZfZGVjZ3BwZmltYWxha2Vsb3BwZmtvaGJvaGNs
Y3Bta2IgMTQgMCBSDQovRXhwZXJ0UGRmX2dqaWlubWNuZHBnb2tramlhZmVnaGNjbGhmY2hvY25iIDE1IDAgUg0KPj4NCg0KL1Byb2NTZXQgWy9QREYgL1RleHQgL0ltYWdlQ10NCj4+DQoNCi9CQm94I
FswLjAwMDAwIDAuMDAwMDAgMTA1NC4wMDAwMCAyNjE0LjAwMDAwXQ0KL1N1YnR5cGUgL0Zvcm0NCi9MZW5ndGggMjE0NDkNCi9UeXBlIC9YT2JqZWN0DQovTmFtZSAvRXhwZXJ0UGRmX2IjZW
1IZmhnY2dsaGtvZGVwamlsbWhmYnBuZmhuZXBiDQo+Pg0Kc3RyZWFtDQp4nN29a28cSZIIKux8K6A+zM70+y42FoO7U31Xj/C3e2HuACxKKnFbr5VYU2hAwEWKTlnZSmZyMpNVzfkj+3dvZJIZYU6a0R8R0
RsV09NdwmHJefyEmR3zCPeI8nG5/b9i/09WKnnzR67Z/o+rafFjsfj6q+Lfq/+Wd39+/vVX+F+cez8oG7S8C519/dWPX39181vYns+tf64+N3+3awII+tc//T8Vo+J/ff3Vv9/9SyVgpdT+r5f1HytaiiH4/BYOfp0Kw6
oVOS5xclzi5LhAWVBwh8ppXDidSS1LtzoS0q+qRwOMQ+F5/HDtnMH5ebiMiEamOuMHAw/ygCHj4SJG15ahx6isJYKP6Wx+Ofo5I7OvL+QBx6HwtnVFNPSMY+iv83COJy8P5nQyOWaFxsgBfH4LVwgLGtadK
UdVN51FLU8348rMq+rTaMah8Fx+DOVHBz8eXlQ0UvNM5+cHXsPDDxkPVzG6slahR+lnHCeuL8/kl6cfWqyirq/PoxmHwtvXFd7Q41Lcxee3clYnLwHzVuRKhpMrGU6uLFEWFMw6Uw5WN44WvTRqObpxy
bOvKqQBx6HwPH4C5ecFPyeSgoWjkZpnDj8YeJAHDBkPL2N0Fa1Cj9KPS0lcX5nNL0c/r1gIXl/IA45D4W3rSgIW3cagvw7gxjksSwnYGyWdnFEMJdfgt8gptISQMOtMOaq66TxqWboxo7OvqvJwHclz+RmUX0
QSeuFFRSM1zwx+XuBBHgovsrd0MmGcdacfM5a4vjaXX5Z+qFlGXl+FmyuFZ/ITKD+6+OLljYpLap4Z/Lz4gzwUbrq3dBJhnHWnH22uMpdfln6oWUZeX4WbK4Xfw0/ueWmtTfVPyY1U19ZRI0/BYU+g5V18fgt
nmKeQMG9FjjmcHHM4OaZRFhTsEHI3BIu6I2GcgxtkXOu7+NzHSzAuGAbC3jOJ8rHR0rDi9j+3YtStdPXrNEFDR9BgIZjre8Tg2qBqCCdQNSAOfx8cJ0sOdN63iegYIjqI36ulcBJVRDqGKgJx7wqAcfICBJv5bSI6hog
l4vcqlh2eMYpzVBGIw18lx8ISBJ35bSI6hggL4vfHSNn8ewbykHfxOcS1BnA9iofeUoO2ooattvAJXX2DqoHnPiywKZNwxkMSgloTAN6oANYW5UDAOdSaqE+kBuWBgxBwd9SaQuKNCmAoDxiEgEd2QZvCnqg
avHJwEALujrjld6oAlbygEEleGSxJlieaF4msgA6rqsJUsg51Aq8W2cAxvOQgl1nIWNgLgVVA9XLuaAVhKnlqAZSKl2aw/OQgt1YDRSqBqqXc0ErCFNrmQZDizVQe9JUc3gBo2A3Vm+HqoGy71zQpsLUOiweQ0g
DwfJE8+oXC6BuID3H0KKsqTqGocZuGGrshqGli4TH2g55qjUF35swYexBalmqNamdRs27cnAQApYjbYc81Zqq6k2YcM8gtZZpMLhYa2pPmmpeUMFBCFiOtB3yVGvKvjdhwj6D1LosHkNlA8GyRPPrFwugcjy9
0ICjDFQdjhp7A/s9B8dLFwWPtR3yVAM+wF3Q2MPUeDv3TKPG8fpAwW6k7ZCnGii23AXdM0ytZRoMLdZA7UITjeMFjlLdSNshTzXgBtwF7TNMrcPiMYQ0ECxPNI65OIW68fRCA44yUHUkbuwSN3aJly4KHm
07JHH3IDJo7GFqsp17plGTeH2gYDnWdkjiFiVl0D3D1FqmwdBiDdSeNNUkXsAoWI61HZK4TUkZtM8wtQ6LxxDSQLA80STq4gQqx9MLDTjKQNVRuLEr3NgVXrooeLTtkiNtR2yuEVZF3TPMLWWaTC0WAO1J
001ixcwCnZjbYcsbIPWBe0zTK3D4jGENBAsTzSLujiBuvH0QgOOsqbq2Bl19gae+zBaukh4rO2Qp1pT8L0JE8YepJalWpPaadS8KwcHIWA50nbIU62pA2OCAwMDAwMCBuDQp0cmFpbGVyDQo8PA0KL1Jvb3
QgMSAwIFINCi9TaXplIDI0DQo+Pg0KDQpzdGFydHhyZWYNCjUyMDg1DQolJUVPRg0K|||||F|||20190117154532

HL7 NBS Results: Message Structure Information

Below are the key segments that will be included in HL7 NBS electronic test result messages published by GDSP :

- **Message Header (MSH) –** Specifies details about the message (Message Type - Newborn Screening Results, Sender, HIE Receiver, etc.)
  e.g. MSH|^~\&|SISGDSP|SISGDSP|SISHIERECEIVER^11223344^L,M,N|^NPI123456^L,M,N|20140324140740||ORU^R01^ORU_R01|220220550|T|2.5.1

- **Patient Identifier (PID) –** Patient demographic information (Name, MRN, DOB, Sex, Ethnicity, etc.)
  e.g. PID|1||3242112234^^^NPI123456&NPI^MR||Johnson^^^^^^B||201312020000|M||2106-3^White||||||||||||2186-5^Not Hispanic or Latino^^^^||N|1

- **Next of Kin (NK1) –** Next of Kin, providing information details of the mother (Name, Address, Number, etc.)
  e.g. NK1|1|Johnson^Mitchell|MTH^Mother|2592 DUET DR, # APT125&^^W SACRAMENTO^CA^95691-4520^USA|^^^^^916^3720117||||||||||198011020000

- **Common Order (ORC) –** Transmits details common to all test results (Clinician, Hospital information on TRF Form Number, Hospital Order ID etc.)
  *e.g.*ORC|RE|25989006^FormNumber||W12312312^HospOrderNum|||||||11356370^CUNNINGHAM^REBECCA^^^^^^NPI|||

- **Observation Request (OBR) –** Transmits information about an exam, diagnostic study/observation, or assessment that is specific to an order or result (like Cystic Fibrosis, Galactosemia)
  e.g. OBR|9|24680^FormNumber||54078-1^Cystic fibrosis newborn screening panel|||20131204000000|||||||||NPI123456^KIM^ JULEANN^^^^^^NPI||||||20140529110321|||F

- **Observation Result (OBX) –** Carries the value of measured and computed results of the diagnostic observation (Birth weight and time, Acylcarnitine newborn screen interpretation, Fatty acid oxidation defects, etc.)
  e.g. OBX|1|NM|8339-4^Birthweight ^LN|1|3543|g |>2500|N|||F|||25052011|

- **Note (NTE) –** Provides additional testing related comments, as needed.
  e.g. NTE|1||Cutoff: >=0.42

HL7 ACK Message Sample

Below is a sample ACK Message

```
MSH|^~\&|SISHIERECEIVER|^1801088422^L,M,N|SISGDSP|SISGDSP|20130614210011||ACK^R01^ACK|220270737|T|2.5.1|MSA|AA|220270737|
```

HL7 Acknowledgement/Error: Message Information

Below are some of the important segments that need to be included in HL7 ACK/ NACK messages from the partner system to GDSP:

- **Message Header (MSH) –** Specifies details about the message (Message Type - Newborn Screening Results, Sender, HIE Receiver, etc.)

  e.g. MSH|^~\&|SISHIERECEIVER|^1538484316^L,M,N|SISGDSP|SISGDSP|20130912210027||ACK^R01^ACK|220949844|P|2.5.1|

- **Message Acknowledgement (MSA) –** Includes HL7 message reference identifier and acknowledgement code, such as:
  - Application Accept(AA) - A positive code that indicates message that was accepted correctly
  - Application Error (AE) – A negative code that indicates that were either a problem with the message structure, or the message itself
  - Application Reject (AR) – A negative code that indicates that message was rejected

  e.g. MSA|AA|1234567890

- **Error (ERR)* –** Provides detailed text on the exact error of the message

  e.g. ERR||OBR^1|100^Segment sequence error^HL70357|E|||Missing required OBR segment|Email help desk for further information on this error||||^NET^Internet^helpdesk@hl7.org

  **Note: Error Segment is only required if acknowledgement code is AE or AR*