Genetic Disease Screening Program
Electronic Newborn Screening (NBS) Results
NBS Results Onboarding Program
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## Introductions: Key Participants from GDSP

**California Department of Public Health (CDPH)**  
*Genetic Disease Screening Program (GDSP)*

<table>
<thead>
<tr>
<th>Name</th>
<th>Position</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stan Sciortino</td>
<td>Chief of Program Development and Evaluation (PDE)</td>
</tr>
<tr>
<td>Tracey Bishop</td>
<td>Chief of Newborn Screening</td>
</tr>
<tr>
<td>Jorge Palacios</td>
<td>Health Program Specialist</td>
</tr>
</tbody>
</table>
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.

**11 Hospital Organizations Partnering**

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

**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.

**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

Based on 2020 metrics, California represents about **12%** of all births in the United States.
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
The *Electronic Results Onboarding* process will follow the phases outlined below. GDSP will support onboarding provider organizations throughout the implementation process.

<table>
<thead>
<tr>
<th>Project Phase</th>
<th>Key Activities</th>
<th>Activity Owner</th>
</tr>
</thead>
</table>
| Initiate Project               | • Obtain necessary project approvals  
                                 | • Conduct Kick-Off discussions & outline the Onboarding Process. Share necessary documentation | Partner Organization  
                                 |                                       | GDSP                     |
| 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                     |
| 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                     |
| 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                     |
| Go-Live                        | • Begin transmitting HL7 results and monitor interface performance  
                                 | • Begin receiving electronic results and monitor interface performance | Partner Organization  
                                 |                                       | GDSP                     |
| 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
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)

---

1 Go-Live is done a day prior (4/8). However, GDSP will begin publishing HL7 results messages the following day (4/9).
Apart from the above, for any general questions, issues or feedback during the onboarding process, partner organizations should email nbshie@cdph.ca.gov.
### Key Meetings during the Onboarding Process (2 of 2)

<table>
<thead>
<tr>
<th>Meeting Name</th>
<th>Project Phase</th>
<th>Project Timeframe</th>
<th>Description</th>
<th>Frequency</th>
<th>Mandatory?</th>
<th>Duration</th>
</tr>
</thead>
<tbody>
<tr>
<td>Interface Testing Readiness Checkpoint</td>
<td>Develop and Test</td>
<td>Week 13 - 14</td>
<td>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</td>
<td>Once</td>
<td>Yes</td>
<td>0.5 Hours</td>
</tr>
<tr>
<td>Go-Live Readiness Checkpoint</td>
<td>Interface Testing</td>
<td>Week 16 - 17</td>
<td>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</td>
<td>Once</td>
<td>Yes</td>
<td>0.5 Hours</td>
</tr>
</tbody>
</table>

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
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
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)
- **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/](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/](https://hiegatewaystag.cdph.ca.gov/GDSPHL7Tools/)
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.
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)

## Patient Information (PID)

- **Baby's Name**: QM1QWVCNI, **Mother's Name**: QM1QWVCNI
- **Gender**: Male
- **NBS Form**: 50123108
- **Medical Record**: 4422135

## NBS Screen Card (OBR 3)

- **Date of Birth**: 08/24/19
- **Time of Birth**: 10:26
- **Age at Collection**: 24 hours
- **Birth Weight**: 3035 grams
- **Ethnicity**: Asian Pacific Islander
- **Specimen Collection Site**: Stanford/Lucile Packard Children’s Hospital
- **Feeding Type**: Human Milk & Formula, Other
- **Newborn on TPN/Hyperal or Amino Acids**: Yes

## Next of Kin (NK1)

- **Mother's Name**: QM1QWVCNI, **Mother's Name**: QM1QWVCNI
- **Address**: 2502 Ouet Dr, Apt 125, West Sacramento, CA 95691-4520
- **Phone**: (916) 372-0117

## Residing Clinician (MSH.6)

- **Residing Clinician**: Rebecca Colin, MD
- **Testing Laboratory**: Allied Laboratory CAM W 0524 D LB RU HTRA, Mountain View, CA 94040-9569
- **Director**: Rasoul Ali Khani Koupaei, PhD, Genetic Disease Laboratory, (510) 231-1780

## Conditions Tested & Positive Markers (OBR 2)

- **Adrenoleukodystrophy (ALD)**
- **Biotinidase Deficiency (BD)**
- **Congenital Adrenal Hyperplasia (CAH)**
- **Cystic Fibrosis (CF)**
- **Galactosemia (GAL)**
- **MS/MS Aconitine Panel**
- **Hemoglobinopathies Panel (OBR 13)**

## Hemoglobinopathies Panel (OBR 13)

- **Hemoglobinopathies**:
  - **Hemoglobin A**: 88.0%
  - **Hemoglobin F**: 12.0%
  - **Hemoglobin C**: 2.0%
  - **Hemoglobin S**: 2.0%
  - **Hemoglobin B**: 0.5%

## Newborn Screening Short Summary (OBR 2)

### NEGATIVE TEST INTERPRETATIONS

<table>
<thead>
<tr>
<th>Condition</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adrenoleukodystrophy (ALD)</td>
<td>MS/MS Amino Acid Panel (Including PKU)</td>
</tr>
<tr>
<td>Biotinidase Deficiency (BD)</td>
<td>Mucopolysaccharidosis I (MPS I)</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (CAH)</td>
<td>Pompe Disease</td>
</tr>
<tr>
<td>Cystic Fibrosis (CF)</td>
<td>Primary Congenital Hypothyroidism (PCH)</td>
</tr>
<tr>
<td>Galactosemia (GAL)</td>
<td>Severe Combined Immunodeficiency (SCID)</td>
</tr>
<tr>
<td>MS/MS Aconitine Panel</td>
<td></td>
</tr>
</tbody>
</table>

## Hemoglobin Pattern (OBR 2)

<table>
<thead>
<tr>
<th>Hemoglobin Pattern</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>FA</td>
<td>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.</td>
</tr>
</tbody>
</table>

## Follow-up:

- None specified.
### HL7 Message Structure: PDF Mailer to HL7 Sample Mapping (2 of 2)

#### Amino Acid Panel (OBR|5)

<table>
<thead>
<tr>
<th>Analyte</th>
<th>Cutoff</th>
<th>Result</th>
<th>Flag</th>
</tr>
</thead>
<tbody>
<tr>
<td>CF</td>
<td>&gt; 68 nM/L</td>
<td>31.00 nM/L</td>
<td></td>
</tr>
<tr>
<td>BO</td>
<td>&gt; 15.00 nM/L</td>
<td>15.00 nM/L</td>
<td></td>
</tr>
<tr>
<td>GAL (Galactose 1 Uridyltransferase (GALT))</td>
<td>&gt; 50 enz units</td>
<td>55.00 enz units</td>
<td></td>
</tr>
<tr>
<td>PCH (Thyroid Stimulating Hormone (TSH))</td>
<td>&gt; 26 nM/L</td>
<td>14.50 nM/L</td>
<td></td>
</tr>
<tr>
<td>SCID (T-cell Receptor Excision Circle (TREC))</td>
<td>&gt; 18 copies/µL</td>
<td>33 copies/µL</td>
<td></td>
</tr>
<tr>
<td>CAH (17-Hydroxyprogesterone (17-OHP))</td>
<td>&gt; 65 nM/L</td>
<td>35 nM/L</td>
<td></td>
</tr>
</tbody>
</table>

#### Cystic Fibrosis Panel (OBR|9)

<table>
<thead>
<tr>
<th>Analyte</th>
<th>Cutoff</th>
<th>Result</th>
<th>Flag</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acylcarnitine</td>
<td>&gt; 8 to &lt; 125 µmol/L</td>
<td>35 µmol/L</td>
<td></td>
</tr>
<tr>
<td>FC ((C16 + C18:1) Ratio)</td>
<td>&lt; 75</td>
<td>37.5</td>
<td></td>
</tr>
<tr>
<td>C-2</td>
<td>&gt; 11 to &lt; 40 µmol/L</td>
<td>20.38 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-3</td>
<td>&gt; 6.15 µmol/L</td>
<td>3.15 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C03 / CO2 Ratio</td>
<td>&gt; 0.3</td>
<td>0.15</td>
<td></td>
</tr>
<tr>
<td>CO3 / CO2 Ratio</td>
<td>&gt; 0.30 µmol/L</td>
<td>0.2 µmol/L</td>
<td></td>
</tr>
<tr>
<td>CO3 / CO3DC Ratio</td>
<td>&gt; 0.2</td>
<td>0.62500</td>
<td></td>
</tr>
<tr>
<td>CO3DC / C10 Ratio</td>
<td>&gt; 0.6</td>
<td>1.50000</td>
<td></td>
</tr>
<tr>
<td>C-4</td>
<td>&gt; 1.7 µmol/L</td>
<td>0.68 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-6</td>
<td>&gt; 0.65 µmol/L</td>
<td>0.46 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C06 / C09 Ratio</td>
<td>&gt; 0.45</td>
<td>0.19073</td>
<td></td>
</tr>
<tr>
<td>C-9</td>
<td>&gt; 0.5 µmol/L</td>
<td>0.3 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C5OH</td>
<td>&gt; 0.65 µmol/L</td>
<td>0.32 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C6H10</td>
<td>&gt; 0.65 µmol/L</td>
<td>0.32 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-12</td>
<td>&gt; 0.45 µmol/L</td>
<td>0.22 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-14</td>
<td>&gt; 2 µmol/L</td>
<td>1 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-16</td>
<td>&gt; 1.2 µmol/L</td>
<td>0.6 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-42</td>
<td>&gt; 0.2 µmol/L</td>
<td>0.1 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-18</td>
<td>&gt; 0.1 µmol/L</td>
<td>0.05 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-18:2</td>
<td>&gt; 0.1 µmol/L</td>
<td>0.05 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-18:3</td>
<td>&gt; 0.1 µmol/L</td>
<td>0.05 µmol/L</td>
<td></td>
</tr>
<tr>
<td>C-18:4</td>
<td>&gt; 0.1 µmol/L</td>
<td>0.05 µmol/L</td>
<td></td>
</tr>
</tbody>
</table>

#### Very Long Chain Fatty Acid Tier 1 (ALD)

<table>
<thead>
<tr>
<th>Fatty Acids and Ratios</th>
<th>Result</th>
<th>Reference Range</th>
<th>Cutoff</th>
<th>Unit</th>
<th>Flag</th>
</tr>
</thead>
<tbody>
<tr>
<td>C26</td>
<td>0.2</td>
<td>(0.16-0.38)</td>
<td>&gt;0.42</td>
<td>µmol/L</td>
<td></td>
</tr>
</tbody>
</table>

#### Lysosomal Storage Diseases

<table>
<thead>
<tr>
<th>Analyte</th>
<th>Result</th>
<th>Cutoff</th>
<th>Unit</th>
<th>Flag</th>
<th>Interpretation Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pompe: Acid alpha-glucosidase (GAA)</td>
<td>12.923</td>
<td>&lt;2.079</td>
<td>µmol/Lh</td>
<td></td>
<td>The acid alpha-glucosidase Enzyme activity level is above the 16% of the daily patient median and suggests it is screen negative for Pompe disease.</td>
</tr>
<tr>
<td>MPS I: Alpha-L-iduronidase (DUA)</td>
<td>3.117</td>
<td>&lt;1.2204</td>
<td>µmol/Lh</td>
<td></td>
<td>The alpha-L-iduronidase Enzyme activity is above the 16% of the daily patient median and suggests it is screen negative for Mucopolysaccharidosis I (MPS I) disease.</td>
</tr>
</tbody>
</table>

#### ALD Tier-1 Panel (OBR|19)

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

#### Pompe Panel (OBR|16)

- MPS I Panel (OBR|17)

#### Galactosemia Panel (OBR|12)

#### PCH Panel (OBR|11)

#### SCID Panel (OBR|15)

#### Biotinidase Panel (OBR|14)

#### CAH Panel (OBR|15)
Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results.
Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results
Below is a sample HL7 message for SIS GDSP newborn screening initial negative test results:

```hl7
<table>
<thead>
<tr>
<th>1</th>
<th>NM</th>
<th>33160-8</th>
<th>Propionylcarnitine</th>
<th>C3:15:1 µmol/L</th>
<th>0.31</th>
<th>[0.28,0.39]</th>
<th>[0.28,0.39]</th>
<th>[0.28,0.39]</th>
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<tbody>
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<td>2</td>
<td>NM</td>
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<td>Propionylcarnitine</td>
<td>C3:15:1 [NÄTAT]</td>
<td>0.31</td>
<td>[0.28,0.39]</td>
<td>[0.28,0.39]</td>
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<td>3</td>
<td>NM</td>
<td>67705-8</td>
<td>Hydroxofururylcarnitine</td>
<td>C4:0 [N:1] µmol/L</td>
<td>0.31</td>
<td>[0.28,0.39]</td>
<td>[0.28,0.39]</td>
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<td>Hydroxofururylcarnitine</td>
<td>C4:0 [N:1] µmol/L</td>
<td>0.31</td>
<td>[0.28,0.39]</td>
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<td>5</td>
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<td>3-hydroxyacylcarnitine</td>
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<td>[0.28,0.39]</td>
<td>[0.28,0.39]</td>
<td>[0.28,0.39]</td>
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<td>C5:0 [N:1] µmol/L</td>
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<td>12</td>
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<td>[0.28,0.39]</td>
<td>[0.28,0.39]</td>
<td>[0.28,0.39]</td>
</tr>
</tbody>
</table>
```

Updated: 12/10/2021
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:

```
--- 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:

```

Updated: 12/10/2021
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|||||||198011020000

- **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
Below is a sample ACK Message

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