

# **HIE TOOLS – USER GUIDE**

Version: 4

October 10<sup>th</sup>, 2019

## LIST OF REVISIONS

REVISION No.	DATE	REVISED BY:	PAGE/S	DESCRIPTION
1	02/24/2014	GDSP		Draft Version
2	07/08/2016	GDSP		Updated the document to include ALD disorder, SCID Confirmatory, ALD Sequencing mailers and changes for MSMS TPN, CAH II included.
3	5/3/18	GDSP		Updated all screenshots since new disorders Pompe and MPS I support was added in the tool. Added Pompe and MPS I related section.
4	10/10/19	GDSP		Updated all screenshots since changes to HL7 in October 2019

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## INTRODUCTION

The purpose of this document is to provide an encompassing description of the usage of the HIE Tools, a utility to test the data exchanged between GDSP and health care provider through Newborn Screening (NBS) Results Health Information Exchange (HIE) interface. HIE Tools has features to test both the NBS HL7 Result message authored by GDSP as well as the HL7 Acknowledgement generated by the health care provider.

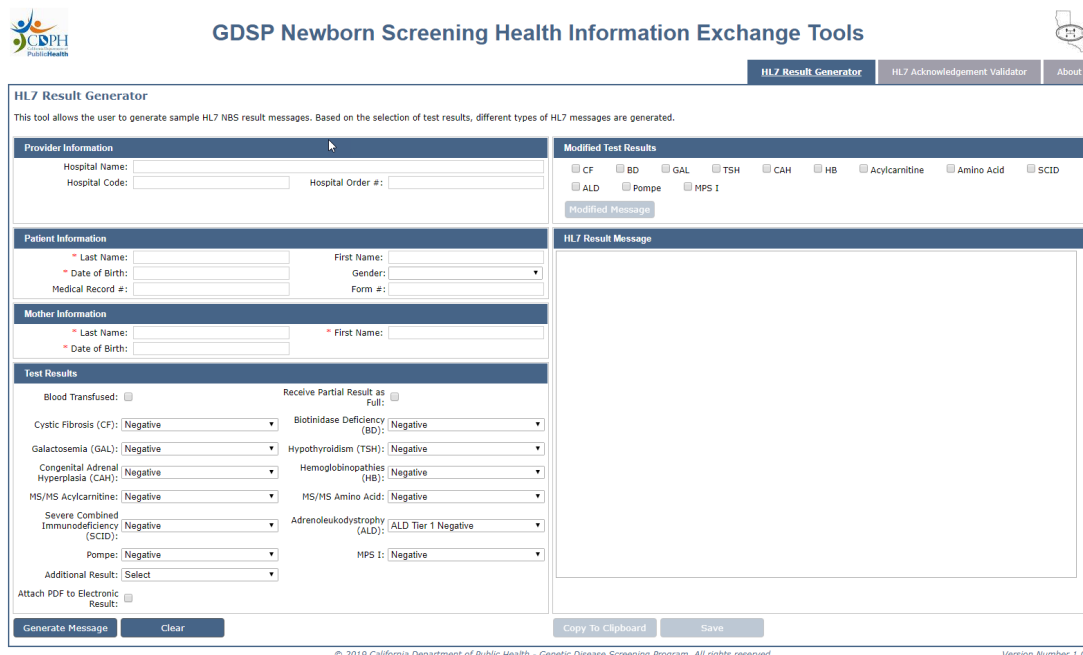
HIE tools will allow new health providers to get quickly on-boarded to Health Information Exchange (HIE) Program by GDSP.

Currently, HIE Tools include the following features:

- **HL7 Result Generator** – Generate sample NBS HL7 Result messages
- **HL7 Acknowledgement Validator** – Validate the HL7 acknowledgement (ACK) messages

Tool	URL
<b>HL7 Result Generator</b>	<a href="https://hiegateway.cdph.ca.gov/GDSPHL7Tools/HL7Message/HL7Generator">https://hiegateway.cdph.ca.gov/GDSPHL7Tools/HL7Message/HL7Generator</a>
<b>HL7 Acknowledgement Validator</b>	<a href="https://hiegateway.cdph.ca.gov/GDSPHL7Tools/HL7Message/HL7Generator">https://hiegateway.cdph.ca.gov/GDSPHL7Tools/HL7Message/HL7Generator</a>

**Table 1:** HIE Tools location



**Figure 1:** HIE Tools interface

# 1.0 NBS HL7 RESULT GENERATOR

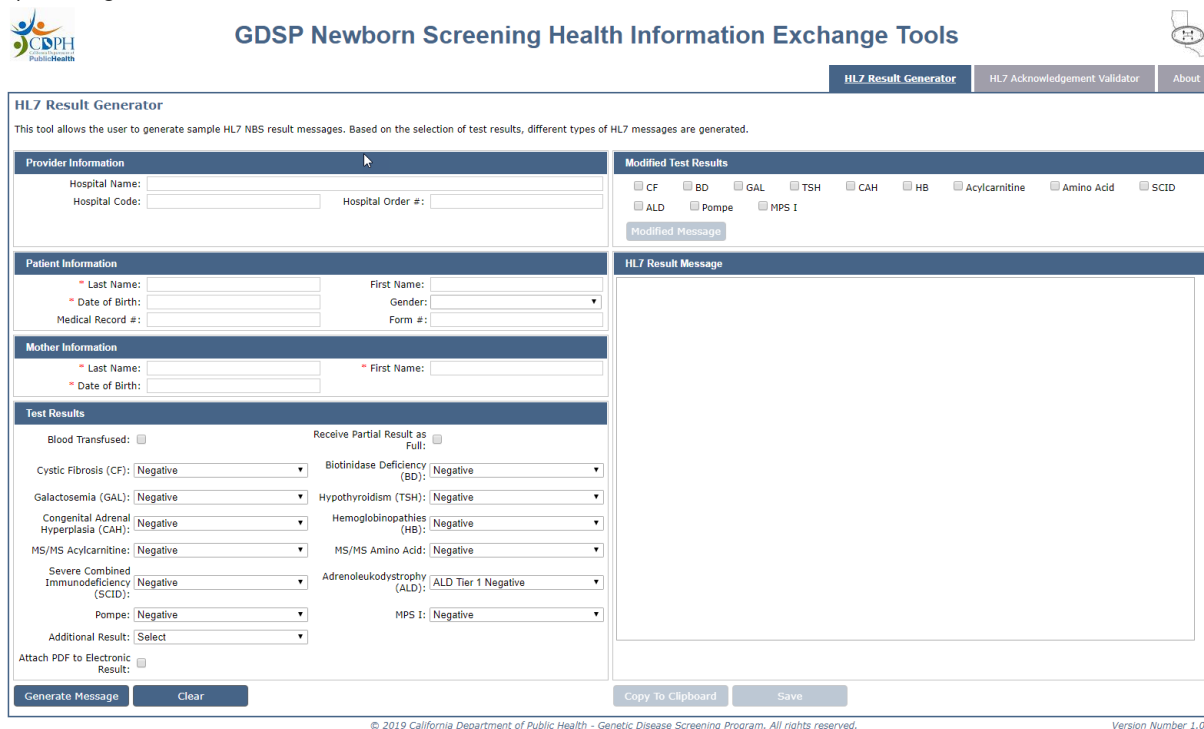
NBS HL7 Result Generator is a tool to generate sample HL7 messages which resemble the actual NBS results sent through Health Information Exchange (HIE) interface. The sample messages contain the newborn screening results in HL7 2.5.1 format. Refer specifications described in the section 3.0 of *4A\_HIE NBS Data Exchange Specification.docx* document from onboarding packet.

HL7 Result Generator tool allows user to generate a variety of NBS HL7 results. Results are generated based on the interpretation selected for each disorder in the tool interface.

## 1.1 STEPS TO GENERATE SAMPLE NBS RESULTS

HL7 Result Generator tool can be used to generate sample NBS HL7 Result messages using the following steps:

- 1) Navigate to NBS HL7 Result Generator tool



**Figure 1.1.1: HL7 Result Generator user interface**

- 2) The input fields marked with asterisk (\*) are mandatory. Enter the data in the input fields as shown below

HL7 Result Generator

HL7 Acknowledgement Validator

About

HL7 Result Generator

This tool allows the user to generate sample HL7 NBS result messages. Based on the selection of test results, different types of HL7 messages are generated.

Provider Information

Hospital Name: Stanford Hospital

Hospital Code:  Hospital Order #:

Patient Information

Last Name: Doe First Name: John

Date of Birth: 10/01/2019 Gender:

Medical Record #:  Form #:

Mother Information

Last Name: Doe First Name: Jane

Date of Birth: 07/14/1988

Test Results

Blood Transfused: ☐ Receive Partial Result as Full: ☐

Cystic Fibrosis (CF):  Biotinidase Deficiency (BD):

Galactosemia (GAL):  Hypothyroidism (TSH):

Congenital Adrenal Hyperplasia (CAH):  Hemoglobinopathies (HB):

MS/MS Acylcarnitine:  MS/MS Amino Acid:

Severe Combined Immunodeficiency (SCID):  Adrenoleukodystrophy (ALD):

Pompe:  MPS I:

Additional Result:

Attach PDF to Electronic Result: ☐

Generate Message

Clear

Copy To Clipboard

Save

Modified Test Results

CF BD GAL TSH CAH HB Acylcarnitine Amino Acid SCID

ALD Pompe MPS I

Modified Message


HL7 Result Message

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Version Number 1.0

Figure 1.1.2: Sample input to HL7 Result Generator

- Click the 'Generate Message' button. HL7 Message will be displayed in the HL7 Result Message textbox as shown below



GDSP Newborn Screening Health Information Exchange Tools



HL7 Result Generator

HL7 Acknowledgement Validator

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Hospital Name: Stanford Hospital

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Mother Information

Last Name: Doe First Name: Jane

Date of Birth: 07/14/1988

Test Results

Blood Transfused: ☐ Receive Partial Result as Full: ☐

Cystic Fibrosis (CF):  Biotinidase Deficiency (BD):

Galactosemia (GAL):  Hypothyroidism (TSH):

Congenital Adrenal Hyperplasia (CAH):  Hemoglobinopathies (HB):

MS/MS Acylcarnitine:  MS/MS Amino Acid:

Severe Combined Immunodeficiency (SCID):  Adrenoleukodystrophy (ALD):

Pompe:  MPS I:

Additional Result:

Attach PDF to Electronic Result: ☐

Generate Message

Clear

Copy To Clipboard

Save

Modified Test Results

CF BD GAL TSH CAH HB Acylcarnitine Amino Acid SCID

ALD Pompe MPS I

Modified Message

HL7 Result Message

MSH|^~\&|SISGOSP|SISGOSP|SISHERCEIVER^11223344^L^H^N^L^H^N^20191009113506||ORU^R01^ORU\_R01|22022~P10|||~^NPI^MR|Doe^John^8||201910010000||2106-3^White|||||2186-5^Not Hispanic or Lat||W1||Doe^Jane^M^Mother|500 Marina Bay Parkway, APT 125^Richmond^CA^94806-4000^USA|^^^916^37201ORC^RE^FormNumber||^HospordNumber||||||^CORTEZ^ALAN^NPI|||||Stanford Hospital|300 PASTOR 1^FormNumber||54089-8^NB Screen Panel Patient AHIC||201910030000|||||James^Smith|||||201OR 2^FormNumber||57128-1^Newborn Screening Report summary panel||201910030000|||||James^SeltOR 1||CE|57721-3^Reason for lab test in Dried blood spot^LN1|LA12421-6^S^Initial screen^S^LN||LN||OR 2||CE|57718-9^Sample quality of Dried blood spot^LN1|LA12432-3^S^Acceptable^S^LN||LN||F||20191OR 3||CE|57130-7^Newborn screening report - overall interpretation^LN1|LA12428-1^S^All screening isOR 4||CE|57131-5^Newborn conditions with positive markers [Identifier] in Dried blood spot^LN1|LA13OR 5||CE|57720-5^Newborn conditions with equivocal markers [Identifier] in Dried blood spot^LN1|LA1OR 6||TX|57724-7^Newborn screening short narrative summary^LN1|NBS Testing Lab - COMMUNITY REG HEDIOR 7||TX|57129-9^Full newborn screening summary report for display or printing^LN1|LN||LN||F||20191OR 8||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 9||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 10||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 11||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 12||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 13||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 14||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 15||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 16||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 17||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 18||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 19||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 20||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 21||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 22||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 23||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood sOR 24||CE|57719-7^Conditions tested for in this newborn screening study [Identifier] in Dried blood s

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Version Number 1.0

Figure 1.1.3: Sample NBS HL7 Result message

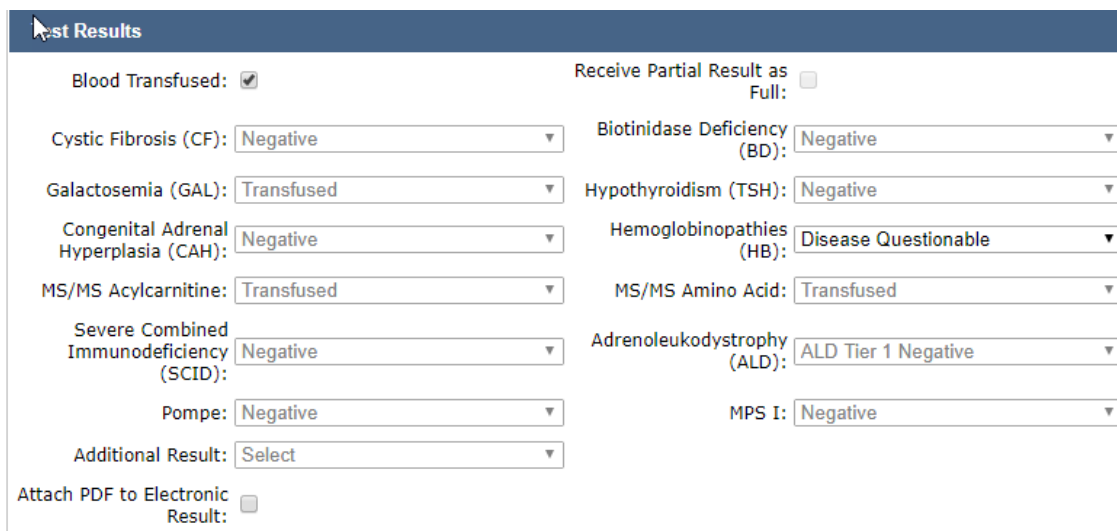
3

4) User can also make different combinations in the Test Results and the HL7 result message will be generated accordingly. Following are some of the scenarios for which HL7 message can be generated:

a) **Blood Transfused:** Select this option if the baby's blood is transfused. When Blood Transfused checkbox is checked, the Test Results are pre-selected with following values

- Cystic Fibrosis = Negative
- Biotinidase Deficiency= Negative
- Galactosemia = Transfused
- Hypothyroidism = Negative
- Congenital Adrenal Hyperplasia = Negative
- MS/MS Acylcarnitine = Transfused
- MS/MS Amino Acid = Transfused
- SCID = Negative
- ALD = ALD Tier 1 Negative
- *Additional Result* dropdown is disabled
- *Hemoglobinopathies* dropdown list has 3 values that can be selected
  - Disease Questionable
  - HB Uninterpretable
  - HB Variant

The generated HL7 message will have Specimen Collection date (every OBR.7) 2 days after baby birth and Blood Transfusion Date (every OBR.14) 3 days after Specimen Collection date.



**Figure 1.1.5:** Blood Transfused option in HL7 Result Generator

b) **Additional Result:** Additional Result types are different from the NBS HL7 results generated for a normal or specific positive case. Structurally the message will be more or less similar but may vary from case to case basis



When any of the Additional Result values is selected, all other fields in Test Results section are disabled shown below.

Test Results	
Blood Transfused: <input type="checkbox"/>	Receive Partial Result as Full: <input type="checkbox"/>
Cystic Fibrosis (CF): <input type="text" value="Select"/>	Biotinidase Deficiency (BD): <input type="text" value="Select"/>
Galactosemia (GAL): <input type="text" value="Select"/>	Hypothyroidism (TSH): <input type="text" value="Select"/>
Congenital Adrenal Hyperplasia (CAH): <input type="text" value="Select"/>	Hemoglobinopathies (HB): <input type="text" value="Select"/>
MS/MS Acylcarnitine: <input type="text" value="Select"/>	MS/MS Amino Acid: <input type="text" value="Select"/>
Severe Combined Immunodeficiency (SCID): <input type="text" value="Select"/>	Adrenoleukodystrophy (ALD): <input type="text" value="Select"/>
Pompe: <input type="text" value="Select"/>	MPS I: <input type="text" value="Select"/>
Additional Result: <input type="text" value="NBS Result – Repeat (SCID Test)"/>	
Attach PDF to Electronic Result: <input type="checkbox"/>	

**Figure 1.1.6:** Additional Result option in HL7 Result Generator

Different Additional Results are as follows:

**i. NBS Result – Initial (Inadequate Specimen)**

- This type of result is generated when the initial specimen collected from a patient was inadequate for testing and deriving interpretations.
- Newborn Screening Report summary panel in HL7 result message will have *Reason for lab test in Dried blood spot* (OBX with LOINC 57721-3) with reason (Observation Value - OBX.5). as “Subsequent screen - for clarification of initial results (not by law or protocol)”.
- Newborn Screening Report summary panel in HL7 result message will have *Sample quality of Dried blood spot* (OBX with LOINC 57718-9) with Observation Value (OBX.5) as “Specimen quantity insufficient for testing”.
- Narrative Summary in HL7 result message will have –
  - Action Identifier as ACTION REQUIRED and
  - Follow-up message as “\*\*\* This specimen was inadequate for testing for the following reason”
- Other disorder panels will not be present

**ii. NBS Result – Initial (Too Early or Missing Information)**

- This type of result is generated if the specimen collection time was within 12 hours of baby's birth.
- Every OBR segment in HL7 result message must have Specimen Collection date (OBR.7) less than 12 hours from baby birth (PID.7).
- Narrative Summary in HL7 result message will have –
  - Action Identifier as ACTION REQUIRED and
  - Follow-up message
- Other disorder panels will be present

**iii. NBS Result – Repeat**

- This type of result is generated when second specimen is collected since the first specimen collected was inadequate for testing. This result, syntactically is similar to the initial result case
- This result is sent in addition to a previous *NBS HL7 Result – Initial (Inadequate Specimen)* or *NBS Result – Initial (Too Early or Missing Information)*
- Newborn Screening Report summary panel in HL7 result message will have Reason for lab test in Dried blood spot (OBX with LOINC 57721-3) with reason (Observation Value - OBX.5) as “Subsequent screen - for clarification of initial results (not by law or protocol)”
- Newborn Screening Report summary panel in HL7 result message will have Sample quality of Dried blood spot (OBX with LOINC 57718-9) with Observation Value (OBX.5) as “Acceptable”
- Other disorder panels will be present

**iv. NBS Result – Repeat (SCID Test)**

- This type of result is generated when second specimen is collected to specifically re-test for SCID disorder.
- This result is sent in addition to an Initial NBS Result sent earlier.
- The result will have Newborn Screening Report summary panel with Reason for lab test in Dried blood spot as “Subsequent screen - for clarification of initial results (not by law or protocol”.
- Disorder panel present only for the SCID disorder

**v. NBS Result – CFTR DNA Sequencing**

- This type of result is generated when specimen collected is found to be positive for Cystic Fibrosis disorder.

- This result is sent in addition to an Initial NBS Result sent before.
  - Apart from the complete result message, additional CF specific result will be sent.
  - Narrative Summary in HL7 result message will have
    - Action Identifier as ACTION REQUIRED and
    - Follow-up message.
  - Disorder panel present only for the Cystic Fibrosis disorder.
- vi. NBS Result – SCID Confirmatory**
- This type of result is generated when specimen collected is found to be SCID positive or confirmatory test is requested for SCID disorder.
  - This result is sent in addition to Initial NBS Result sent before.
  - Apart from the complete result message, additional SCID specific result will be sent.
  - Narrative Summary in HL7 result message will have
    - Action Identifier as ACTION REQUIRED and
    - Follow-up message.
  - Disorder panel present only for the SCID disorder.
  - Hospital Order Number added on screen is not included in this mailer.
- vii. NBS Result – ALD Sequencing with Mutation**
- This type of result is generated when specimen collected is found to be ALD positive with mutation.
  - This result is sent in addition to NBS Result sent before.
  - ALD sequencing result mailer is sent as an additional result mailer as a correction to NBS result mailer on arrival of ALD sequencing results.
  - Narrative Summary in HL7 result message will have
    - Action Identifier as ACTION REQUIRED and
    - Follow-up message.

**vii. NBS Result – ALD Sequencing without Mutation**

- This type of result is generated when specimen collected is found to be ALD positive without mutation.
- This result is sent in addition to NBS Result sent before.
- ALD sequencing result mailer is sent as an additional result mailer as a correction to NBS result mailer on arrival of ALD sequencing results.
- Narrative Summary in HL7 result message will have
  - Action Identifier as ACTION REQUIRED and
  - Follow-up message.

**ix. NBS Result – Pompe Sequencing**

- This type of result is generated when specimen collected is found to be Pompe Sequencing positive
- This result is sent in addition to NBS Result sent before.
- Pompe sequencing result mailer is sent as an additional result mailer as a correction to NBS result mailer on arrival of Pompe sequencing results.
- Narrative Summary in HL7 result message will have
  - Action Identifier as ACTION REQUIRED and
  - Follow-up message.

**x. NBS Result – MPS I Sequencing – Conclusive Variant**

- This type of result is generated when specimen collected is found to be MPS I Sequencing with Pathogenic Variant
- This result is sent in addition to NBS Result sent before.
- MPS I sequencing result mailer is sent as an additional result mailer as a correction to NBS result mailer on arrival of MPS I sequencing results.
- Narrative Summary in HL7 result message will have
  - Action Identifier as ACTION REQUIRED and
  - Follow-up message.

**xi. NBS Result – MPS I Sequencing – Pseudo Deficiency**

- This type of result is generated when specimen collected is found to be MPS I Sequencing with Pseudo deficiency Variant
- This result is sent in addition to NBS Result sent before.

- MPS I sequencing result mailer is sent as an additional result mailer as a correction to NBS result mailer on arrival of MPS I sequencing results.
  - Narrative Summary in HL7 result message will have
    - Action Identifier
    - Follow-up message.
- c) **Modified Test Results:** Modified Test Result types are different from the NBS HL7 results generated for a normal or specific positive case. Structurally the message will be more or less similar but may vary from case to case basis

## 1.2 COPY AND SAVE RESULTS

- 1) User can copy the generated HL7 message to clipboard by clicking on 'Copy To Clipboard' button. This feature is available only in Internet Explorer web browser.
- 2) User can save the generated HL7 message by clicking on the 'Save' button. The result message is saved to a file in .hl7 format with default filename as *SampleHL7Result*.
- 3) User can clear the form data (input fields and HL7 Result Message) by clicking the 'Clear' button. A popup appears for user confirmation to clear form data. Click on 'Yes' button to clear the form.

## 2.0 HL7 ACKNOWLEDGEMENT VALIDATOR

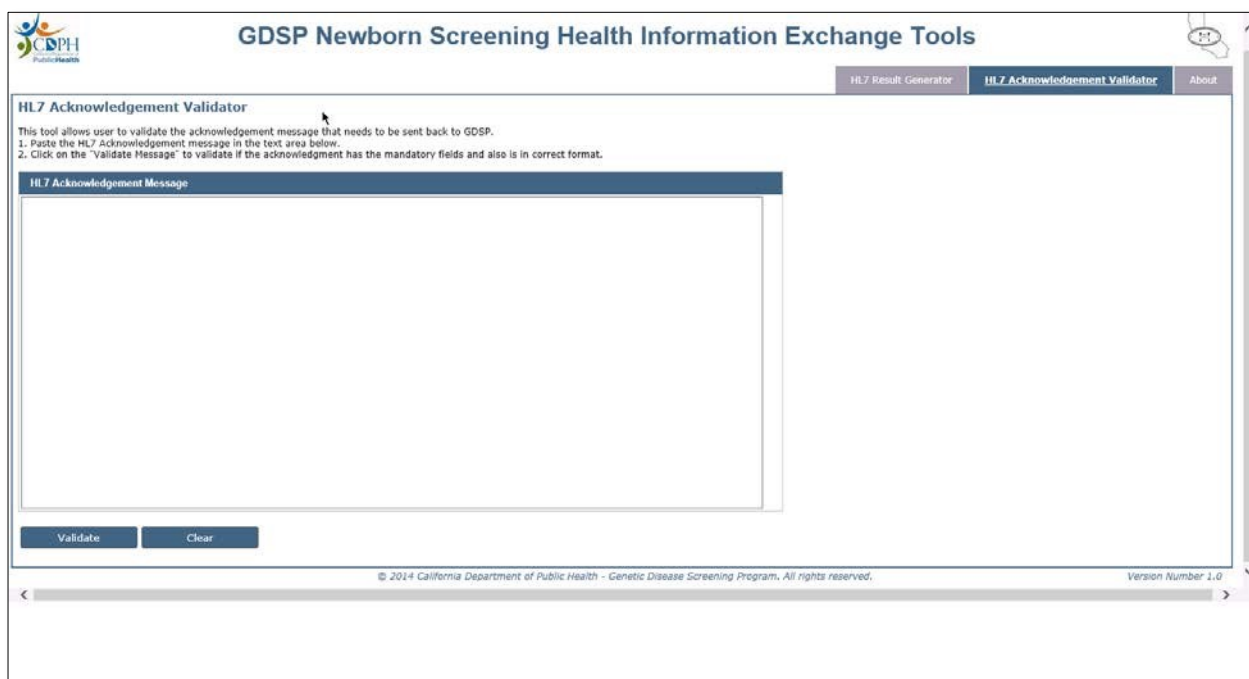
When GDSP sends NBS HL7 Result message to health care provider, it expects to receive an acknowledgement (ACK/NACK) message to confirm successful transmission and processing. GDSP supports the 'Original acknowledgment mode' protocol for message acknowledgement.

HL7 Acknowledgement Validator is a tool to validate HL7 acknowledgements (HL7 2.5.1 format). Refer specifications described in the section 4.0 of *4A\_HIE NBS Data Exchange Specification.docx* document from onboarding packet.

### 2.1 STEPS TO VALIDATE ACKNOWLEDGEMENT MESSAGES

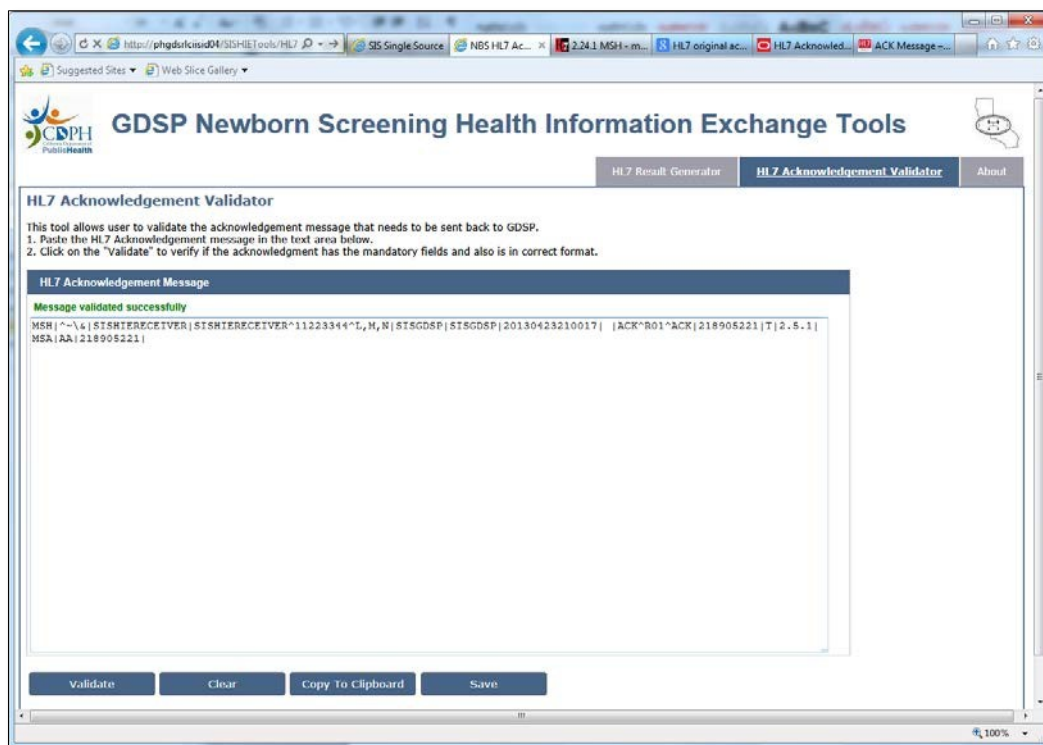
HL7 Acknowledgement Validator tool can be used to validate HL7 Acknowledgement messages using the following steps:

- 1) Navigate to HL7 Acknowledgement Validator tool.

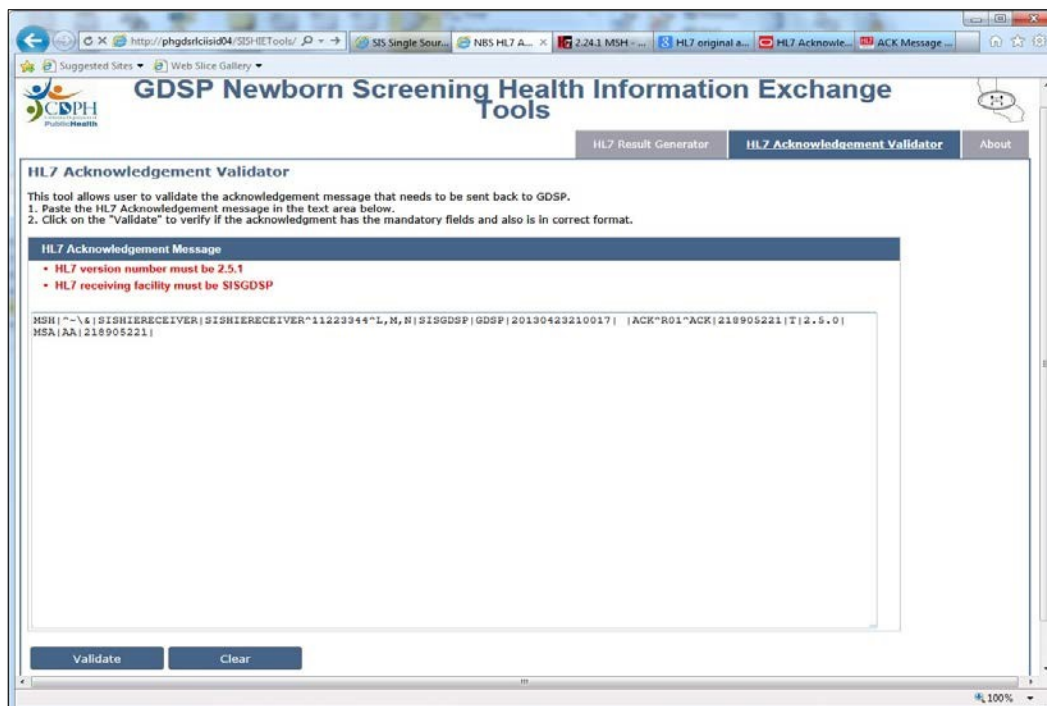


**Figure 2.1.1:** HL7 Acknowledgement Validator user interface

- 2) Paste the acknowledgement in the HL7 Acknowledgement Message text area. Refer to the sample acknowledgement messages in the onboarding packet.
- 3) Click on the Validate button at the bottom of the page. This brings up the validation result message on top of the HL7 Acknowledgement Message panel as shown below.



**Figure 2.1.2:** Successful validation of HL7 Acknowledgement



**Figure 2.1.3:** Unsuccessful validation of HL7 Acknowledgement



## 2.2 COPY AND SAVE ACKNOWLEDGEMENT

- 1) User can copy the generated HL7 message to clipboard by clicking on 'Copy To Clipboard' button. This feature is available only in Internet Explorer web browser.
- 2) User can save the validated acknowledgement by clicking the 'Save' button. The acknowledgement message is saved to a file in .hl7 format with default filename as *SampleHL7Ack*.
- 3) User can clear the acknowledgement message by clicking the 'Clear' button. A popup appears for user confirmation to clear form data. Click 'Yes' button to clear the form.