Newborn Screening Quality Assurance Program
Sickle Cell and Other Hemoglobinopathies
Proficiency Testing Program (HbPT)

In co-sponsorship with Association of Public Health Laboratories (APHL)
Provided by the Newborn Screening and Molecular Biology Branch
Centers for Disease Control and Prevention
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Quarterly Report
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Report Authorization

This report has been reviewed and authorized by Dr. Joanne Mei, Laboratory Chief, Newborn Screening Quality Assurance Program.

Confidentiality Statement

NSQAP participant information and evaluations are strictly confidential and shared only with individual participants, unless written authorization for release is received.

Introduction

This report is the summary of HbPT data reported within the specified period for Quarter 4, 2018. It is distributed to all participants, state laboratory directors, and program colleagues by request. The content includes specimen certification profiles, material distribution information, frequency tables for presumptive phenotypes, clinical assessments, and reported methods. An evaluation of your reported data is attached to this summary.

Certification of PT Specimens

The dried blood spot (DBS) specimens in this panel were prepared from purchased umbilical cord blood. Table 1 lists the hemoglobin presumptive phenotypes and their presumptive clinical assessments.
Table 1. Specimen Certification

<table>
<thead>
<tr>
<th>Specimen</th>
<th>Expected Presumptive Phenotype</th>
<th>Accepted Presumptive Phenotype</th>
<th>Expected Presumptive Clinical Assessment</th>
<th>Accepted Presumptive Clinical Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>418H1</td>
<td>FS</td>
<td>FSE, FSV</td>
<td>Hb SS disease</td>
<td>Hb S with an uncommon variant; Hb SE disease</td>
</tr>
<tr>
<td>418H2</td>
<td>FA</td>
<td></td>
<td>Normal – No abnormal Hb found</td>
<td></td>
</tr>
<tr>
<td>418H3</td>
<td>FAC</td>
<td></td>
<td>Hemoglobin C trait</td>
<td></td>
</tr>
<tr>
<td>418H4</td>
<td>FAS</td>
<td></td>
<td>Hemoglobin S trait</td>
<td></td>
</tr>
<tr>
<td>418H5</td>
<td>FA</td>
<td></td>
<td>Normal – No abnormal Hb found</td>
<td></td>
</tr>
</tbody>
</table>

Distribution of PT Specimens

On September 25, 2018 a PT panel of five DBS specimens was distributed to 46 domestic and 29 foreign laboratories.

Participant Results

We received data from 70 participants by the data reporting deadline. Participants assayed all survey specimens by the analytical schemes they routinely use, and reported, for each specimen, the presumptive phenotype, presumptive clinical assessment, and any other clinical classifications they deem consistent with their analytic results and program operations.

Presumptive phenotypes and presumptive clinical assessments should be reported as directed on the Hemoglobinopathies Data Report Form to avoid point deductions from their overall score. Laboratories should:

- Report one presumptive phenotype derived from results of all methods used by their laboratory for each specimen. Supplement unusual phenotype reports with comments in the Phenotype Comments section of the HbPT Data Report Form.
- List the hemoglobins in the order of their abundance using standard phenotypic nomenclature when reporting the phenotype.
- **Not** insert symbols or blank spaces into the presumptive phenotype nomenclature.
- Report presumptive clinical assessments not listed in the drop-down menu under the comment section in order to receive points for the overall score.

Tables 2a-e show the frequency distribution of participant reported presumptive clinical phenotypes along the frequency of misclassifications for each specimen. Tables 3a-e show the frequency distribution of reported presumptive clinical assessments and the frequency of misclassifications for each specimen.
Table 2a. Frequency Distribution of Reported Presumptive Clinical Phenotypes
Specimen 418H1

<table>
<thead>
<tr>
<th>Presumptive Clinical Phenotype</th>
<th>Phenotype Frequency</th>
<th>#Correctly Classified Phenotype</th>
<th>Misclassified Phenotype</th>
<th>Not Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>FS</td>
<td>64</td>
<td>64</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Other</td>
<td>6</td>
<td>4</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 2b. Frequency Distribution of Reported Presumptive Clinical Phenotypes
Specimen 418H2

<table>
<thead>
<tr>
<th>Presumptive Clinical Phenotype</th>
<th>Phenotype Frequency</th>
<th>#Correctly Classified Phenotype</th>
<th>Misclassified Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>FA</td>
<td>70</td>
<td>70</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 2c. Frequency Distribution of Reported Presumptive Clinical Phenotypes
Specimen 418H3

<table>
<thead>
<tr>
<th>Presumptive Clinical Phenotype</th>
<th>Phenotype Frequency</th>
<th>#Correctly Classified Phenotype</th>
<th>Misclassified Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>FAC</td>
<td>68</td>
<td>68</td>
<td>0</td>
</tr>
<tr>
<td>Other</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 2d. Frequency Distribution of Reported Presumptive Clinical Phenotypes
Specimen 418H4

<table>
<thead>
<tr>
<th>Presumptive Clinical Phenotype</th>
<th>Phenotype Frequency</th>
<th>#Correctly Classified Phenotype</th>
<th>Misclassified Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>FAS</td>
<td>68</td>
<td>68</td>
<td>0</td>
</tr>
<tr>
<td>Other</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

Table 2e. Frequency Distribution of Reported Presumptive Clinical Phenotypes
Specimen 418H5

<table>
<thead>
<tr>
<th>Presumptive Clinical Phenotype</th>
<th>Phenotype Frequency</th>
<th>#Correctly Classified Phenotype</th>
<th>Misclassified Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>FA</td>
<td>70</td>
<td>70</td>
<td>0</td>
</tr>
</tbody>
</table>
Table 3a. Frequency Distribution of Reported Presumptive Clinical Assessments
Specimen 418H1

<table>
<thead>
<tr>
<th>Presumptive Clinical Assessment</th>
<th>Assessment Frequency</th>
<th>#Correctly Classified Assessment</th>
<th>Misclassified Assessment</th>
<th>Not Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hb SS Disease</td>
<td>65</td>
<td>65</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Other</td>
<td>5</td>
<td>2</td>
<td>1</td>
<td>2</td>
</tr>
</tbody>
</table>

Table 3b. Frequency Distribution of Reported Presumptive Clinical Assessments
Specimen 418H2

<table>
<thead>
<tr>
<th>Presumptive Clinical Assessment</th>
<th>Assessment Frequency</th>
<th>#Correctly Classified Assessment</th>
<th>Misclassified Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal – No abnormal Hb found</td>
<td>70</td>
<td>70</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 3c. Frequency Distribution of Reported Presumptive Clinical Assessments
Specimen 418H3

<table>
<thead>
<tr>
<th>Presumptive Clinical Assessment</th>
<th>Assessment Frequency</th>
<th>#Correctly Classified Assessment</th>
<th>Misclassified Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hemoglobin C trait</td>
<td>70</td>
<td>70</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 3d. Frequency Distribution of Reported Presumptive Clinical Assessments
Specimen 418H4

<table>
<thead>
<tr>
<th>Presumptive Clinical Assessment</th>
<th>Assessment Frequency</th>
<th>#Correctly Classified Assessment</th>
<th>Misclassified Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hemoglobin S trait</td>
<td>70</td>
<td>70</td>
<td>0</td>
</tr>
</tbody>
</table>

Table 3e. Frequency Distribution of Reported Presumptive Clinical Assessments
Specimen 418H5

<table>
<thead>
<tr>
<th>Presumptive Clinical Assessment</th>
<th>Assessment Frequency</th>
<th>#Correctly Classified Assessment</th>
<th>Misclassified Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal – No abnormal Hb found</td>
<td>70</td>
<td>70</td>
<td>0</td>
</tr>
</tbody>
</table>
Total Specimen Error Frequency by Testing Algorithm

Table 4 shows the frequency of errors per testing algorithm for all specimens. Algorithms reported by less than three participants are not shown.

<table>
<thead>
<tr>
<th>Primary</th>
<th>Secondary</th>
<th>Tertiary</th>
<th>Total Specimens</th>
<th>Presumptive Phenotype Errors</th>
<th>Presumptive Clinical Assessment Errors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isoelectric Focusing</td>
<td>Bio-Rad Screening HPLC</td>
<td>Primus Ultra 2 HPLC</td>
<td>25</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Bio-Rad Screening HPLC</td>
<td></td>
<td></td>
<td>45</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Bio-Rad Screening HPLC</td>
<td>Isoelectric Focusing</td>
<td></td>
<td>25</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Bio-Rad Screening HPLC</td>
<td>Isoelectric Focusing</td>
<td></td>
<td>65</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

*Methods are designated as “Other” when less than three participants report results for a given method. Currently those methods include:

IEC-HPLC
MS/MS
Capillars—ALERE
Sebia capillars Neonatal Haemoglobin

Evaluations

Overall, participants reported three Presumptive Phenotype misclassifications and one Presumptive Clinical Assessment misclassification.

Future Shipments

The Newborn Screening Quality Assurance Program will ship next quarter’s HbPT specimens on January 15, 2019.

Acknowledgments

The specimens for this program were prepared from umbilical cord blood samples supplied by Cleveland Cord Blood Center, Cleveland, Ohio; LifeSouth Community Blood Centers, Inc., Gainesville, FL; and Life Line Stem Cell, New Haven, IN. Patient specimens were provided by Children’s Hospital Oakland Research Institute (CHORI).

The content of this report may also be located on our website at:

http://www.cdc.gov/labstandards/nsqap_reports.html
This NEWBORN SCREENING QUALITY ASSURANCE PROGRAM report is an internal publication distributed to program participants and selected program colleagues. The laboratory quality assurance program is a project cosponsored by the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories.

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