# Changes to Newborn Screening Cystic Fibrosis Testing and Result Reporting Statements

Newborn screening laboratory – Department of State Health Services

The Texas DSHS Newborn Screening (NBS) Laboratory is pleased to announce full in-house Cystic Fibrosis 2nd-tier DNA testing has resumed. Several Newborn Screening Cystic Fibrosis result reporting statements will be updated to reflect the change in methodology. These updates are scheduled to go into effect Tuesday, May 24, 2016. All changes are indicated in red. A full list of all possible results including all Cystic Fibrosis results including mutation names can be found here: [http://www.dshs.texas.gov/lab/docs/SubmitterMailer-May2016.pdf](http://www.dshs.texas.gov/lab/docs/SubmitterMailer-May2016.pdf).

<table>
<thead>
<tr>
<th>Overall Specimen Result</th>
<th>Disorder</th>
<th>Screening Result</th>
<th>Analyte(s)</th>
<th>Analyte Result</th>
<th>Screening Result Note</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>Cystic Fibrosis</td>
<td>Normal</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>Normal</td>
<td>0 Mutations Detected No further evaluation necessary unless clinically indicated. The immunoreactive trypsinogen (IRT) result was normal. Tests for a 40-mutation panel in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were negative, however, the presence of other mutations cannot be ruled out.</td>
</tr>
<tr>
<td>Abnormal</td>
<td>Cystic Fibrosis</td>
<td>Result may be Abnormal or Inconclusive</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>IRT Elevated CFTR Mutation Panel may be 0, 1, or 2 Mutations Detected</td>
<td>Revised Screening Result for Cystic Fibrosis. Additional testing using a CFTR 40 Mutation Panel has been performed. <em>(Note: Result notes vary depending on the results applied for CFTR Mutation Panel)</em> [The specimen was originally reported as Indeterminate for Cystic Fibrosis showing Immunoreactive Trypsinogen as Elevated. The original screening result note read “Please repeat the newborn screen within 7 days. Many unaffected infants have an elevated immunoreactive trypsinogen (IRT) level on the first specimen. The second screening specimen (collected after 7 days of age) is required to determine if result is significant.”]</td>
</tr>
<tr>
<td>Abnormal</td>
<td>Cystic Fibrosis</td>
<td>Inconclusive</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>Elevated</td>
<td>0 Mutations Detected No further evaluation necessary unless clinically indicated. None of the 40 CFTR mutations in the DSHS panel were detected, but there is a minimal risk for Cystic Fibrosis due to a mutation not included in the panel. Clinical evaluation not necessary unless symptomatic.</td>
</tr>
<tr>
<td>Abnormal</td>
<td>Cystic Fibrosis</td>
<td>Abnormal</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>Very Elevated 0 Mutations Detected</td>
<td></td>
</tr>
<tr>
<td>----------</td>
<td>----------------</td>
<td>----------</td>
<td>-------------------------------------------------</td>
<td>----------------------------------</td>
<td></td>
</tr>
<tr>
<td>Abnormal</td>
<td>Cystic Fibrosis</td>
<td>Abnormal</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>Elevated or Normal 1 Mutation Detected</td>
<td></td>
</tr>
<tr>
<td>Abnormal</td>
<td>Cystic Fibrosis</td>
<td>Abnormal</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>Elevated or Normal 2 Mutations Detected</td>
<td></td>
</tr>
<tr>
<td>Abnormal</td>
<td>Cystic Fibrosis</td>
<td>Abnormal</td>
<td>Immunoreactive Trypsinogen CFTR 40 Mutation Panel</td>
<td>Elevated or Normal 2 Mutations Detected</td>
<td></td>
</tr>
</tbody>
</table>

**Reminders:**
- Read Screening Result Notes fully before taking action.
- Contact the laboratory with any questions:
  - Telephone: 1-888-963-7111 X7585 or x2638
  - Email: NewbornScreeningLab@dshs.state.tx.us