Newborn Screening (NBS) Overview

Each baby born in Texas is tested for 52 genetic disorders and hypothyroidism, for a total of 53 disorders
- First screen is collected at 24-48 hours of age
- Second screen is collected at 1-2 weeks of age
- ~380,000 babies are screened each year
- ~760,000 specimens are tested each year

Testing
- Small dots are punched from dried blood spots to initiate the testing
- Initial results are reviewed and re-tested as indicated to confirm whether normal or out-of-range
- The Laboratory reports out-of-range results to NBS Clinical Care Coordination staff who begin follow-up protocols
- Results are reported to the submitting provider

Number of Affected Newborns Identified through Texas Newborn Screening

Calendar Year 2017 (updated 6/13/2018)

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Biotinidase Deficiency</td>
<td>22</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia</td>
<td>68</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>61</td>
</tr>
<tr>
<td>Classic Galactosemia</td>
<td>7</td>
</tr>
<tr>
<td>Primary Hypothyroidism</td>
<td>258</td>
</tr>
<tr>
<td>Sickling Hemoglobinopathies</td>
<td>193</td>
</tr>
<tr>
<td>Non-Sickling Hemoglobinopathies</td>
<td>25</td>
</tr>
<tr>
<td>Metabolic Disorders</td>
<td>118</td>
</tr>
<tr>
<td>Severe Combined Immune Def.</td>
<td>5</td>
</tr>
<tr>
<td>Secondary T-Cell Lymphopenias</td>
<td>70</td>
</tr>
<tr>
<td><strong>Total:</strong></td>
<td><strong>827</strong></td>
</tr>
</tbody>
</table>

*Numbers are subject to change as some disorders may take over a year to diagnose

Timeline of a Specimen in the Laboratory

* Business Days

Day 1*
- Specimen arrives.
- Specimen reviewed and prepared for testing.
- Testing begins.
- Demographic entry begins.

Day 2*
- Demographic entry is completed.
- Testing completed for most specimens; additional testing for some specimens is needed.
- Newborn Screening Clinical Care Coordination staff contact providers about time critical out-of-range results.

Day 3*
- Results for most specimens are completed.
- Clinical Care Coordination staff contact providers about out-of-range results for remaining disorders, except SCID.

Day 4*
- Results for Galactosemia DNA and SCID tests are completed.
- Clinical Care Coordination staff contact providers about out-of-range results for SCID.
- Remaining results reported.
- Completed test result reports are available to healthcare providers (mailed, faxed, HL7 electronic and online).

Providing testing services in support of the Texas Newborn Screening Program.

Early detection of disorders, from a heel stick blood sample, allows early treatment that can prevent serious complications.

Laboratory Services Section, MC 1947
Department of State Health Services
PO Box 149347
Austin, TX 78714-9347
(512) 776-7333
Fax (512) 776-7712
dshs.texas.gov/lab/newbornscreening.shtm
### Newborn Screening Process

**Healthcare Provider**
- Healthcare provider sends order request for newborn screening collection kits
  **Note:** DSHS Laboratory assigns the serial number on the newborn screening kit to the provider, then ships the kit
- Healthcare provider distributes Parent page and Decision form to parent (pages 1 and 2 of NBS kit)
  **Note:** If 1st screen, healthcare provider instructs parents to take Parent page to baby's next appointment
- Healthcare provider instructs parent to read and complete Decision form and return to provider or send to DSHS Laboratory
- Healthcare provider collects specimen and sends to DSHS Laboratory within 24 hours of collection

**DSHS Laboratory**
- DSHS provides courier services to selected areas to facilitate timely delivery of specimens
- Specimen Acquisition receives specimen and assigns a laboratory ID number
- Demographic Entry Group enters information from demographic forms or electronic interface
- NBS Testing Group performs testing on blood spots:
  1. Endocrine and Cystic Fibrosis
  2. Galactosemia and Biotinidase
  3. Hemoglobinopathies
  4. Severe Combined Immunodeficiency
  5. Tandem Mass Spectrometry
- Additional second tier testing is completed for some disorders
- Test results are reported to healthcare provider who submitted the specimen and are available through secure NBS web application
- Specimen Logistics Group provides education to reduce the number of unsatisfactory specimens

### DSHS Newborn Screening Panel

#### Amino Acid Disorders

**Core Conditions:**
- Argininosuccinic Aciduria (ASA)
- Citrullinemia, Type I (CIT)
- Homocystinuria (HCY)
- Maple Syrup Urine Disease (MSUD)
- Classic Phenylketonuria (PKU)
- Tyrosinemia, Type I (TYR I)

**Secondary Conditions:**
- Argininemia (ARG)
- Benign Hyperphenylalaninemia (H-PHE)
- Biotinidase defect in cofactor biosynthesis (BIOPT BS)
- Biotinidase defect in cofactor regeneration (BIOPT REG)
- Citrullinemia, Type II (CIT II)
- Hypermethioninemia (MET)
- Tyrosinemia, Type II (TYR II)
- Tyrosinemia, Type III (TYR III)

#### Biotinidase Deficiency

#### Classic Galactosemia

#### Cystic Fibrosis

#### Endocrine Disorders

- Congenital Adrenal Hyperplasia (CAH)
- Primary Congenital Hypothyroidism (CH)

#### Fatty Acid Oxidation Disorders

**Core Conditions:**
- Carnitine Uptake Defect (CUD)
- Long Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Trifunctional Protein Deficiency (TFP)
- Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

**Secondary Conditions:**
- 2,4 Dienoyl-CoA Reductase Deficiency (DE RED)
- Carnitine Acylcarnitine Translocase Deficiency (CACT)
- Carnitine Palmitoyltransferase Type I Deficiency (CPT I)
- Carnitine Palmitoyltransferase Type II Deficiency (CPT II)
- Glutaric Acidemia Type II (GA2)
- Medium-Chain Ketoacyl-CoA Thiolase Deficiency (MCKAT)
- Medium/Short Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD)
- Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)

#### Severe Combined Immunodeficiency

**Core Condition:**
- Severe Combined Immunodeficiency (SCID)

**Secondary Condition:**
- T-cell related lymphocyte deficiencies

#### Organic Acid Disorders

**Core Conditions:**
- 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
- 3-Hydroxy-3-Methylglutaric Aciduria (HMG)
- Beta-Ketothiolase Deficiency (BKT)
- Glutaric Acidemia Type I (GA1)
- Isovaleric Acidemia (IVA)
- Methylmalonic Acidemia (Cobalamin disorders- Cbl A,B)
- Methylmalonic Acidemia (methylmalonyl-CoA mutase)
- Holocarboxylase Synthase Deficiency (Multiple Carboxylase Deficiency-MCD)
- Propionic Acidemia (PROP)

**Secondary Conditions:**
- 2 Methylbutyrylglycinuria (2MBG)
- 2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)
- 3-Methylglutaconic Aciduria (3MGA)
- Isobutyrylglycinuria (IBG)
- Methylmalonic Acidemia with Homocystinuria (Cbl C, D)
- Malonic Acidemia (MAL)

#### Hemoglobinopathies

**Core Conditions:**
- S,S (Sickle Cell Anemia)
- S,C
- S Beta-Thalassemia

**Secondary Condition:**
- Various other hemoglobinopathies

#### DSHS Newborn Screening Panel Continued