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)

Biotinidase Deficiency	22
Congenital Adrenal Hyperplasia	68
Cystic Fibrosis	61
Classic Galactosemia	7
Primary Hypothyroidism	258
Sickling Hemoglobinopathies	193
Non-Sickling Hemoglobinopathies	25
Metabolic Disorders	118
Severe Combined Immune Def.	5
Secondary T-Cell Lymphopenias	70
Total:	827*

**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).

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TEXAS Health and Human Services | Texas Department of State Health Services

Texas Newborn Screening Laboratory

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
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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:

⇒ Eight 3.2mm blood spots are punched from the filter paper and distributed into eight 96-well plates

⇒ Five analytical teams complete the testing:

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)
- Biopterin defect in cofactor biosynthesis (BIOPT BS)
- Biopterin 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)

DSHS Newborn Screening Panel Continued

Hemoglobinopathies

Core Conditions:

- S,S (Sickle Cell Anemia)
- S,C
- S Beta-Thalassemia

Secondary Condition:

- Various other hemoglobinopathies

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)

Severe Combined Immunodeficiency

Core Condition:

- Severe Combined Immunodeficiency (SCID)

Secondary Condition:

- T-cell related lymphocyte deficiencies