What the Laboratory Does

The Tennessee Department of Health's (TDH) Newborn Screening Laboratory works with the Pediatric Case Management group in support of the state newborn screening program. Laboratory testing is provided for early detection of disorders that if not treated could lead to serious complications or even death.

Photo Credit: iStockPhoto

Newborn Screening (NBS) Overview

Every baby born in Tennessee is tested for over 70 genetic disorders.

- ~ 86,000 babies are screened each year
- ~ 100,000 specimens are tested each year
- Screen is collected at 24-48 hours of age; **ideal collection time is 24 hours + 1 minute**
- Federal Recommendations for reporting based on ideal collection and specimen receipt times are:
  - ⇒ 95% of presumed positive results for time-critical conditions should be reported within 5 days of life
  - ⇒ 95% of all newborn screening results for all other conditions should be reported within 7 days of life
  - ⇒ 95% of all newborn screening results for all conditions should be reported within 7 days of life

Newborn Screening Process

Healthcare Provider

- Healthcare provider obtains filter paper for collection from their local health department or the state laboratory
- Healthcare provider distributes parent pamphlet to parents
- Healthcare provider collects specimen and sends to TDH Laboratory within 24 hours of collection
- TDH provides courier services to all birthing facilities to facilitate timely delivery of specimens

State Public Health Laboratory

- Specimen received at state laboratory and a TDH specimen number is assigned
- Demographic information is entered from forms
- Small spots are punched from dried blood spots to initiate the testing:
  1. Eleven 3.2mm blood spots are punched from the filter paper for various testing methods
  2. Testing is performed and completed
  3. Initial results are reviewed and re-tested as indicated to confirm an abnormal screen
  4. Additional second tier testing and/or DNA analysis is completed for some disorders
  5. Abnormal screen results are sent to Pediatric Case Management who follows up with primary care physicians and/or a specialist if necessary
  6. Test results are reported to the provider who submitted the specimen, the listed primary care physician and are available via a secure web portal

Department of Health Authorization No. 343235.
This Electronic publication was promulgated at zero cost.
February 2019

TN Department of Health

Tennessee Newborn Screening Laboratory

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TN Newborn Screening Panel

Amino Acid Disorders

Tests are analyzed quantitatively by Tandem Mass Spectrometry.

- Argininemia
- Carbamoylphosphate Synthethase I Deficiency
- Citrullinemia
- Argininosuccinate Lyase Deficiency
- Nonketotic Hyperglycinemia
- Homocystinuria or variant forms of Hypermethioninemia
- Hypermethioninemia
- Hyperornithinemia-Hyperammonemia-Homocitrullinuria
- Phenylketonuria
- Benign Hyperphenylalaninemia
- Tyrosinemia
- Maple Syrup Urine Disease
- Ornithine transcarbymylase deficiency

Biotinidase Deficiency

Screening is performed by colorimetric methodology.

Cystic Fibrosis

Testing is performed by a quantitative fluoroimmunoassay (FIA) method. Elevated IRT values result in mutation testing.

Galactosemia

Galactosemia testing is performed by a quantitative enzymatic fluorometric method.

- Classic Galactosemia
- Galactokinase Deficiency
- Galactose Epimerase Deficiency

Endocrine Disorders

Testing is performed by a quantitative fluoroimmunoassay (FIA) method.

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia

Fatty Acid Oxidation Disorders

Tests are analyzed quantitatively by Tandem Mass Spectrometry.

- Carnitine Uptake Deficiency
- Carnitine Palmitoyl Transferase Deficiency
- Short Chain AcylCoA Dehydrogenase Deficiency
- Glutaric Acidemia Type II
- Mitochondrial Acetoacetyl CoA Thiolase Deficiency
- Medium Chain AcylCoA Dehydrogenase Deficiency
- Medium/Short Chain AcylCoA Dehydrogenase Deficiency
- 2,4 Dienyl CoA Reductase Deficiency
- Very Long Chain AcylCoA Dehydrogenase Deficiency
- Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency
- Carnitine /Acylcarnitine Translocase Deficiency
- Trifunctional Protein Deficiency
- X-linked Adrenyleukodystrophy

Hemoglobinopathies

Testing is performed by High Performance Liquid Chromatography (HPLC).

- Sickle Cell Anemia
- Sickle Beta Thalassemia
- Hemoglobin S/C Disease
- Other Variant Hemoglobinopathies

Lysosomal Disorders

Enzyme activity is analyzed by Tandem Mass Spectrometry.

- Krabbe
- Pompe
- Gaucher
- Fabry
- Mucopolysaccharidosis I (MPSI)

Organic Acid Disorders

Testing is analyzed quantitatively by Tandem Mass Spectrometry.

- Propionic Acidemia
- Methylmalonic Acidemia
- Multiple CoA Carboxylase Deficiency
- Malonic Aciduria
- Isobutyryl CoA Dehydrogenase Deficiency
- Isovaleric Acidemia
- 2 Methylbutyryl Glycinuria
- 2 Methyl 3 Hydroxybutyric Aciduria
- 3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency
- 3 Methyl Crotonyl CoA Carboxylase Deficiency
- 3 Methylglutaconyl CoA Hydratase Deficiency
- Glutaric Acidemia Type I
- Medium/Short Chain 3 hydroxyacyl CoA dehydrogenase deficiency

Immunodeficiencies

Testing is performed using a real-time PCR method.

- Severe Combined Immunodeficiency
- T-cell Related Lymphocyte Deficiencies

Spinal Muscular Atrophy (coming in 2019)