Newborn Screening Program Disorders

Effective January 29, 2018 Utah newborns are screened for the following disorders:

These are disorders that may have significant mortality and morbidity when not diagnosed pre-symptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders.

- **Amino acid disorders:** recessive disorders resulting from an enzyme deficiency needed for amino acid metabolism or transport.
  - Arginase Deficiency
  - Argininosuccinate Lyase Deficiency (ASA)
  - Citrullinemia
  - Homocystinuria
  - Hyperphenylalanemia, including Phenylketonuria (PKU)
  - Maple Syrup Urine Disease
  - Tyrosinemia

- **Fatty Acid Oxidation Disorders**: recessive disorders resulting from an enzyme deficiency needed for the breakdown of fatty acids.
  - Carnitine uptake/transport defects
  - Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)
  - Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
  - Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
  - Long chain 3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
  - Very long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
  - Carnitine-Acylcarnitine Translocase Deficiency
  - Carnitine Palmitoyl Transferase-1 Deficiency
  - Carnitine Palmitoyl Transferase-2 Deficiency

- **Organic Acid Disorders:** recessive disorders resulting from an enzyme deficiency in the intermediary metabolism of amino acids or fatty acids.
  - Beta-Ketothiolase Deficiency
  - Glutaric Acidemia, Type 1
  - Guanidinoacetate methyltransferase (GAMT) deficiency
  - Isobutyryl CoA Dehydrogenase Deficiency
  - Isovaleric Acidemia
  - Malonic Aciduria
  - Methylmalonic Acidemias
  - Propionic Acidemia
  - 3-Hydroxy-3-Methylglutaryl (HMG) CoA Lyase Deficiency
  - 2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase Deficiency
  - 2-Methylbutyryl CoA Dehydrogenase Deficiency
  - Multiple Carboxylase Deficiency

- **Biotinidase deficiency:** a recessive disorder of biotin metabolism.

- **Congenital Adrenal Hyperplasia (CAH):** a genetic disorder in which there are defects in the enzymes of the adrenal cortex required for the biosynthesis of adrenal corticosteroids.

- **Congenital Hypothyroidism:** a disorder in which the newborn is unable to secrete or produce thyroxine normally.

- **Cystic Fibrosis:** a recessively inherited genetic disorder resulting from a protein deficiency that disrupts the epithelial cells.

- **Galactosemia:** a recessively inherited genetic disorder in which the individual is completely or partially incapable of normal metabolism of galactose due to a deficiency of the galactose-1-phosphate uridyltransferase enzyme.

- **Severe Combined Imunodeficiency Disorder (SCID):** affected individuals lack T lymphocytes which help the body fight infections due to a wide variety of viruses, bacteria and fungi.

- **Sickle cell disease & Hemoglobinopathies**: recessively inherited disorders characterized by the presence of abnormal hemoglobins in the blood.
  - FABarts (Alpha Thalassemia carrier)
  - FAS (Sickle Cell carrier)
  - FAC, D, or E (Carrier trait)
  - FS, FC, FE, FSC (Actual disease state)

- **Spinal Muscular Atrophy (SMA):** a recessively inherited disorder, caused by the deletion of the SMN1 gene; resulting in progressive neurodegenerative disease that affects the motor nerve cells in the spinal cord.

Newborn Screening Program
Utah Public Health Laboratory
PO Box 31431
Salt Lake City, UT 84131-9988
ph: 801-584-8256 f: 801-536-0966
newbornscreening.health.utah.gov

UTAH DEPARTMENT OF HEALTH