Newborn Screening Program Disorders

Effective July 1, 2013 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 break down 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
- 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.
- **SCID:** Severe Combined Imunodeficiency disorder; 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)

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