Newborn Screening Program Disorders
Effective September 2021 Utah newborns are screened for the following disorders:

**Newborn Screening Program 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 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 uridylyltransferase enzyme.

**Severe Combined Immunodeficiency 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 hemoglobin 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.

**X-linked Adrenoleukodystrophy (XALD):** an x-linked genetic disorder that mainly affects the nervous system and the adrenal glands.