

Oklahoma Newborn Screening Disorders List

<ul style="list-style-type: none"> • Congenital Hypothyroidism • Classic Galactosemia • Galactosepimerase deficiency (Gale) • Galctokinase deficiency (GALK) • Sickle Cell Disease (Hb SS) • S, C Disease (Hb SC) • S, βThalasemia (Hb S/βth) • Various other hemoglobinopathies • Cystic Fibrosis • Congenital Adrenal Hyperplasia • Biotinidase Deficiency • Severe Combined Immunodeficiency (SCID) • Spinal Muscular Atrophy (SMA) • X-linked Adrenoleukodystrophy (XALD) • Pompe • Mucopolysaccharidosis Type 1 	<p>Organic Acid Disorders</p> <ul style="list-style-type: none"> • Propionic Acidemia (PROP) • Methylmalonic acidemia (MUT) • Methylmalonic acidemia (Cobalamin Disorders) (Cbl A, B) • Methylmalonic acidemia with homocystinuria (Cbl C,D) • Malonic acidemia (MAL) • Isobutyrylglycinuria (Isobutyryl-CoA dehydrogenase deficiency) (IBG) • Isovaleric Acidemia (IVA) • 2-Methylbutyrylglycinuria (2MBG) • 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC) • 3-Methylglutaconic aciduria (3MGA) • 3-Hydroxy-3-methylglutaric aciduria (HMG) • Holocarboxylase synthetase deficiency (multiple carboxylase deficiency) (MCD) • 2-Methyl-3-hydroxybutyric aciduria (2M3HBA) • Beta ketothiolase deficiency (βKT) • Glutaric acidemia type I (GA1)
<p>Amino Acid Disorders</p> <ul style="list-style-type: none"> • Argininemia (ARG) • Argininosuccinic aciduria (ASA) • Citrullinemia type I (CIT) • Citrullinemia type II (CIT II) • Homocystinuria (HCY) • Hypermethioninemia (MET) • Maple Syrup Urine Disease (MSUD) • Phenylketonuria (PKU) • Benign hyperphenylalaninemia (H-PHE) • Biopterin defect in cofactor biosynthesis (BIOPT [BS]) • Biopterin defect in cofactor regeneration (BIOPT [REG]) • Tyrosinemia Type I (TYR I) • Tyrosinemia Type II (TYR II) • Tyrosinemia Type III (TYR III) 	<p>Fatty Acid Disorders</p> <ul style="list-style-type: none"> • Carnitine uptake defect (CUD) • Short-chain acyl-CoA dehydrogenase deficiency (SCAD) • Glutaric acidemia Type II (GAII) • Medium-chain ketoacyl-CoA thiolase deficiency (MCAT) • Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) • Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD) • Carnitine acylcarnitine translocase deficiency (CACT) • Carnitine palmitoyltransferase I deficiency (CPT I) Carnitine palmitoyltransferase II deficiency (CPT II) • Long-chain L-3-hydroxyacyl- CoA dehydrogenase deficiency (LCHAD)Trifunctional protein deficiency (TFP)