



Newborn Screening Conditions List	
Condition Group	Conditions Included
Hemoglobinopathies	- Sickle cell anemia Hb SS - Hb S/C - Hb S/Beta thalassemia - Other Hb variant including genetic trait
Endocrine Disorders	- Congenital adrenal hyperplasia (CAH) - Congenital hypothyroidism
Inborn Errors of Metabolism: Amino Acid Disorders	- Arginase deficiency (Argininemia) - Argininosuccinic aciduria - Biopterin cofactor defects - Citrullinemia, type I - Citrullinemia, type II (Citron deficiency) - Hypermethioninemia - Hyperphenylalaninemia (H-Phe) - Maple syrup urine disease (MSUD) - Phenylketonuria (PKU) - Tyrosinemia, type I - Tyrosinemia, type II - Tyrosinemia, type III

Inborn Errors of Metabolism: Organic Acidemias

- 2-Methylbutyrylglycinuria (2-MBG)
- 3-Hydroxy-3-methylglutaric aciduria (HMG)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC deficiency)
- 3-Methylglutaconic acidemia (3-MGA)
- Beta-ketothiolase deficiency
- Glutaric acidemia, type I (GA type I)
- Isobutyrylglycinuria (IBG)
- Isovaleric acidemia (IVA)
- Malonic aciduria (MAL)
- Methylmalonic acidemia (MUT or methylmalonyl-CoA mutase)
- Methylmalonic acidemia with cobalamin disorders (CblA & CblB)
- Methylmalonic acidemia with homocystinuria (CblC & CblD)
- Propionic acidemia
- 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)

Inborn Errors of Metabolism: Fatty Acid Oxidation

- 2,4-Dienoyl-CoA Reductase Deficiency
- Carnitine-acylcarnitine translocase deficiency (CACT)
- Carnitine palmitoyltransferase deficiency I (CPT IA)
- Carnitine palmitoyltransferase deficiency II (CPT II)
- Carnitine uptake defect (CUD)
- Glutaric acidemia type II (GA type II)
- Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Medium/short chain L-3-hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD)
- Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- Trifunctional Protein Deficiency
- Very long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- Medium-chain ketoacyl-CoA thiolase deficiency (MCAT)

Lysosomal Storage Disorders

- Krabbe
- Pompe
- Mucopolysaccharidosis type I (MPS-I, Hurler Syndrome)

Other Genetic Conditions

- Cystic fibrosis
- Severe combined immunodeficiency (SCID)
- Spinal muscular atrophy (SMA)
- Biotinidase deficiency
- Adrenoleukodystrophy (ALD)
- Galactosemia:
 - Classic Galactosemia (G/G)
 - Galactosemia D/G variant
 - Other Galactosemia variants
- Hearing loss
- Critical congenital heart disease
- (7 different heart defects detected through pulse oximetry screen)