NEWBORN SCREENING
INDIANA

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 • Hypothyroidism
Inborn Errors of Metabolism: Amino Acid Disorders	• Arginase Deficiency (Argininemia) • Argininosuccinic Aciduria • Biopterin Cofactor Defects • Citrullinemia, type I • Citrullienemia, type II (Citron Deficiency) • Hypermethioninenemia • 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) • Isobutrylglycinuria (IBG) • Isovaleric acidemia (IVA) • Malonic aciduria (MAL) • Methylmalonic acidemia (MUT or methylmalonyl-CoA mutase) • Methylmalonic acidemia with cobalamin disorders (CblA & CblB) • Methylmalonic academia with homocystinuria (CblC & CblD) • Propionic acidemia • 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
Inborn Errors of Metabolism: Fatty Acid Oxidation Disorders	• 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 • MPS-I (Hurler Syndrome)
Other Genetic Conditions	• Cystic Fibrosis • Severe Combined Immunodeficiency (SCID) • Spinal Muscular Atrophy (SMA) • Biotinidase deficiency • 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) • Adrenoleukodystrophy (ALD)

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