



# NEWBORN SCREENING INDIANA

## Newborn Screening Conditions List

Condition Group	Conditions Included
<b>Hemoglobinopathies</b>	<ul style="list-style-type: none"><li>• Sickle cell anemia Hb SS</li><li>• Hb S/C</li><li>• Hb S/Beta thalassemia</li><li>• Other Hb variant including genetic trait</li></ul>
<b>Endocrine Disorders</b>	<ul style="list-style-type: none"><li>• Congenital adrenal hyperplasia</li><li>• Hypothyroidism</li></ul>
<b>Inborn Errors of Metabolism: Amino Acid Disorders</b>	<ul style="list-style-type: none"><li>• Arginase Deficiency (Argininemia)</li><li>• Argininosuccinic Aciduria</li><li>• Biopterin Cofactor Defects</li><li>• Citrullinemia, type I</li><li>• Citrullinemia, type II (Citron Deficiency)</li><li>• Hypermethioninemia</li><li>• Hyperphenylalaninemia (H-Phe)</li><li>• Maple syrup urine disease (MSUD)</li><li>• Phenylketonuria (PKU)</li><li>• Tyrosinemia, type I</li><li>• Tyrosinemia, type II</li><li>• Tyrosinemia, type III</li></ul>

## 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 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, and other galactosemia variants
- Hearing Loss
- Critical Congenital Heart Disease (7 different heart defects detected through pulse oximetry screen)
- X-Linked Adrenoleukodystrophy (X-ALD) \*screening begins 07/01/2021\*



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