

## 2026 Newborn Screening List

### Amino Acid Profile (AA) Disorders

Argininemia (Arginase Deficiency)  
 §Carbamoylphosphate Synthetase I Deficiency  
 Citrullinemia  
 \*Type I (Arginosuccinate Synthetase Deficiency)  
 Type II (Citrin Deficiency)  
 \*Argininosuccinate Lyase Deficiency (Argininosuccinic Aciduria)  
 \*Homocystinuria or variant forms of Hypermethioninemia  
 Hypermethioninemia  
 due to Glycine N-Methyltransferase Deficiency  
 due to S-Adenosylhomocysteine Hydrolase Deficiency  
 due to Methionine Adenosyltransferase Deficiency  
 §Hyperornithinemia-Hyperammonemia-Homocitrullinuria  
 §Hyperornithinemia-Hyperammonemia-Homocitrullinuria w/gyral atrophy  
 \*Phenylketonuria  
 Benign Hyperphenylalaninemia due to:  
 Phenylalanine Hydroxylase Deficiency  
 GTP Cyclohydrolase I Deficiency  
 Pterin-4-Alpha Carbinolamine Dehydratase Deficiency  
 6-Pyruvoyltetrahydropterin Synthase Deficiency  
 Biopterin defect in co-factor biosynthesis  
 Biopterin defect in co-factor regeneration  
 Tyrosinemia  
 \*Type I (Hepatorenal)  
 Type II  
 Type III  
 \*Maple Syrup Urine Disease  
 Type IA  
 Type IB  
 Type II  
 §Ornithine transcarbonylase deficiency (OTC)

### Fatty Acid Profile (FA) Disorders

\*Carnitine Uptake Deficiency (CUD)  
 Carnitine Palmitoyl Transferase Deficiency  
 Type I (CPT I)  
 Type II (CPT II)  
 Short Chain AcylCoA Dehydrogenase Deficiency (SCAD)  
 Glutaric Acidemia Type II (MADD or GAI)  
 \*Medium Chain AcylCoA Dehydrogenase Deficiency (MCAD)  
 Medium/Short Chain AcylCoA Dehydrogenase Deficiency (M/SCHAD)  
 2,4 Dienyl CoA Reductase Deficiency  
 \*Very Long Chain AcylCoA Dehydrogenase Deficiency (VLCAD)  
 \*Long Chain Hydroxyl AcylCoA Dehydrogenase Deficiency (LCHAD)  
 Carnitine /Acylcarnitine Translocase Deficiency (CACTD)  
 \*Trifunctional Protein Deficiency  
 \*Adrenoleukodystrophy (ALD)

### Organic Acid Profile (OA) Disorders

\*Mitochondrial Acetoacetyl CoA Thiolase (Beta Ketothiolase/SKAT) Deficiency  
 \*Propionic Acidemia  
 \*Methylmalonic Acidemia Cobalamin Disorder (CBL A, B)  
 Methylmalonic Acidemia Cobalamin Disorder (CBL C, D)  
 \*Methylmalonyl-CoA Mutase Deficiency  
 \*Multiple CoA Carboxylase Deficiency  
 Malonic Aciduria (MA)  
 Isobutyryl CoA Dehydrogenase Deficiency (IBCD)  
 \*Isovaleric Acidemia (IVA)  
 2 Methylbutyryl Glycinuria (2MBG)  
 2 Methyl 3 Hydroxybutyric Aciduria (2M3HBA)  
 \*3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency (HMG)  
 \*3 Methyl Crotonyl CoA Carboxylase Deficiency (3 MCC)  
 3 Methylglutaconyl CoA Hydratase Deficiency (3MGA)  
 \*Glutaric Acidemia Type I (GAI)  
 Medium/Short Chain 3 hydroxyacyl CoA dehydrogenase deficiency (M/SCHAD)

### Lysosomal Storage Disorders (LSD)

\*Krabbe  
 \*Pompe  
 §Gaucher  
 §Fabry  
 \*Mucopolysaccharidosis I (MPS I)  
 \*Mucopolysaccharidosis II (MPS II)

### Other Disorders

\*Congenital Hypothyroidism  
 \*Congenital Adrenal Hyperplasia  
 \*Biotinidase Deficiency  
 \*Classical Galactosemia  
 Galactokinase Deficiency  
 Galactose Epimerase Deficiency  
 \*Cystic Fibrosis  
 \*Sickle Cell Anemia  
 \*Sickle Beta Thalassemia  
 \*Hemoglobin S/C Disease  
 Other Variant Hemoglobinopathies  
 \*Severe Combined Immunodeficiency  
 T-cell related lymphocyte deficiencies  
 \*Spinal Muscular Atrophy  
 \*Guanidinoacetate Methyltransferase Deficiency (GAMT) - Coming 2026

### Screening Mandated/Information Collection Only

\*Critical Congenital Heart Defect  
 \*Hearing Defect

#### Legend

\* = Core RUSP Conditions  
 § = Other disease screened not core or secondary conditions

## **NBS Disorder/Profile Information**

### **TRANSFUSIONS**

Unless transfusion is marked, the assumption is that the infant has not been transfused.

### **TOTAL GALACTOSE**

Galactose results are based upon the assumption that the infant has had lactose feeding.

### **CYSTIC FIBROSIS**

The CF DNA Mutation analysis is only performed when there is an elevated IRT, or in cases of in utero exposure to CF modulators or a confirmed case of *Meconium ileus*.

### **TANDEM MASS SPECTROMETRY**

(AA): Arginine, Citrulline, Cit/Arg, Leucine, Methionine, Ornithine, Orn/Cit, Phenylalanine, Tyrosine, Phe/Tyr, Valine, Succinylacetone, Argininosuccinic Acid, Asa/Arg

(FA): C0, C4, C5, C5:1, C5DC+C6OH, C6, C8, C8/C10, C10, C10:1, C10:2, C12:1, C14, C14:1, C14:2, C14-OH, C16, C16:1, C16-OH, C18, C18:1, C18:2, C18:1-OH, C0/C16, C0/C18

(OA): C3, C3DC+C4OH, C4, C4DC+C5OH, C5, C5:1, C6-DC, C3/C2, C4DC+C5OH/C8

### **LYSOSOMAL DISORDERS**

(LD): GAA, GALC, GBA, GLA, IDUA, I2S