Recommended Uniform Screening Panel
Core Conditions
(As of July 2024)

X: Condition is in this category --: Condition is not in this category

Core Condition	Metabolic Disorder - Organic acid condition	Metabolic Disorder - Fatty acid oxidation disorder	Metabolic Disorder - Amino acid disorder	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
3-Hydroxy-3-Methyglutaric Aciduria	X	--	--	--	--	--
3-Methylcrotonyl-CoA Carboxylase Deficiency	X	--	--	--	--	--
ß-Ketothiolase Deficiency	X	--	--	--	--	--
Glutaric Acidemia Type I	X	--	--	--	--	--
Holocarboxylase Synthase Deficiency	X	--	--	--	--	--
Isovaleric Acidemia	X	--	--	--	--	--
Methylmalonic Acidemia (Cobalamin disorders)	X	--	--	--	--	--
Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X	--	--	--	--	--
Propionic Acidemia	X	--	--	--	--	--
Carnitine Uptake Defect/Carnitine Transport Defect	--	X	--	--	--	--
Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency	--	X	--	--	--	--
Medium-chain Acyl-CoA Dehydrogenase Deficiency	--	X	--	--	--	--
Trifunctional Protein Deficiency	--	X	--	--	--	--
Very Long-chain Acyl-CoA Dehydrogenase Deficiency	--	X	--	--	--	--
Argininosuccinic Aciduria	--	--	X	--	--	--
Citrullinemia, Type I	--	--	X	--	--	--
Classic Phenylketonuria	--	--	X	--	--	--
Homocystinuria	--	--	X	--	--	--
Maple Syrup Urine Disease	--	--	X	--	--	--
Tyrosinemia, Type I	--	--	X	--	--	--
Congenital adrenal hyperplasia	--	--	--	X	--	--
Primary Congenital Hypothyroidism	--	--	--	X	--	--
S, βeta-Thalassemia	--	--	--	--	X	--
S,C Disease	--	--	--	--	X	--
S,S Disease (Sickle Cell Anemia)	--	--	--	--	X	--
Biotinidase Deficiency	--	--	--	--	--	X
Classic Galactosemia	--	--	--	--	--	X

Core Condition	Metabolic Disorder - Organic acid condition	Metabolic Disorder - Fatty acid oxidation disorder	Metabolic Disorder - Amino acid disorder	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
Critical Congenital Heart Disease	--	--	--	--	--	**X**
Cystic Fibrosis	--	--	--	--	--	**X**
Glycogen Storage Disease Type II (Pompe)	--	--	--	--	--	**X**
Guanidinoacetate Methyltransferase Deficiency	--	--	--	--	--	**X**
Hearing Loss	--	--	--	--	--	**X**
Infantile Krabbe Disease (low galactocerebrosidase [GALC] and psychosine ≥ 10nM)						**X**
Mucopolysaccharidosis Type I	--	--	--	--	--	**X**
Mucopolysaccharidosis Type II	--	--	--	--	--	X
Severe Combined Immunodeficiencies	--	--	--	--	--	X
Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1	--	--	--	--	--	X
X-linked Adrenoleukodystrophy	--	--	--	--	--	X

Recommended Uniform Screening Panel[1] SECONDARY[2] CONDITIONS[3]

(As of July 2024)

Secondary Condition	Metabolic Disorder - Organic acid condition	Metabolic Disorder - Fatty acid oxidation disorder	Metabolic Disorder - Amino acid disorder	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
2-Methyl-3-hydroxybutyric aciduria	**X**	--	--	--	--	--
2-Methylbutyrylglycinuria	**X**	--	--	--	--	--
3-Methylglutaconic aciduria	**X**	--	--	--	--	--
Isobutyrylglycinuria	**X**	--	--	--	--	--
Malonic acidemia	**X**	--	--	--	--	--
Methylmalonic acidemia with homocystinuria	**X**	--	--	--	--	--
2,4 Dienoyl-CoA reductase deficiency	--	**X**	--	--	--	--
Carnitine acylcarnitine translocase deficiency	--	**X**	--	--	--	--
Carnitine palmitoyltransferase type I deficiency	--	**X**	--	--	--	--
Carnitine palmitoyltransferase type II deficiency	--	**X**	--	--	--	--
Glutaric acidemia type II	--	**X**	--	--	--	--
Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency	--	**X**	--	--	--	--

Secondary Condition	Metabolic Disorder - Organic acid condition	Metabolic Disorder - Fatty acid oxidation disorder	Metabolic Disorder - Amino acid disorder	Endocrine Disorder	Hemoglobin Disorder	Other Disorder
Medium-chain ketoacyl-CoA thiolase deficiency	--	**X**	--	--	--	--
Short-chain acyl-CoA dehydrogenase deficiency	--	**X**	--	--	--	--
Argininemia	--	--	**X**	--	--	--
Benign hyperphenylalaninemia	--	--	**X**	--	--	--
Biopterin defect in cofactor biosynthesis	--	--	**X**	--	--	--
Biopterin defect in cofactor regeneration	--	--	**X**	--	--	--
Citrullinemia, type II	--	--	**X**	--	--	--
Hypermethioninemia	--	--	**X**	--	--	--
Tyrosinemia, type II	--	--	**X**	--	--	--
Tyrosinemia, type III	--	--	**X**	--	--	--
Various other hemoglobinopathies	--	--	--	--	**X**	--
Galactoepimerase deficiency	--	--	--	--	--	**X**
Galactokinase deficiency	--	--	--	--	--	**X**
T-cell related lymphocyte deficiencies	--	--	--	--	--	**X**

1. Selection of conditions based upon “Newborn Screening: Towards a Uniform Screening Panel and System.” *Genetic Med*. 2006; 8(5) Suppl: S12-S252” as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that can be detected in the differential diagnosis of a core disorder.
3. Nomenclature for Conditions based upon “Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels.” *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.