Recommended Uniform Screening Panel
Core Conditions
(As of July 2018)

Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
Propionic Acidemia	X					
Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X					
Methylmalonic Acidemia (Cobalamin disorders)	X					
Isovaleric Acidemia	X					
3-Methylcrotonyl-CoA Carboxylase Deficiency	X					
3-Hydroxy-3-Methyglutaric Aciduria	X					
Holocarboxylase Synthase Deficiency	X					
ß-Ketothiolase Deficiency	X					
Glutaric Acidemia Type I	X					
Carnitine Uptake Defect/Carnitine Transport Defect		X				
Medium-chain Acyl-CoA Dehydrogenase Deficiency		X				
Very Long-chain Acyl-CoA Dehydrogenase Deficiency		X				
Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency		X				
Trifunctional Protein Deficiency		X				
Argininosuccinic Aciduria			X			
Citrullinemia, Type I			X			
Maple Syrup Urine Disease			X			
Homocystinuria			X			
Classic Phenylketonuria			X			
Tyrosinemia, Type I			X			
Primary Congenital Hypothyroidism				X		
Congenital adrenal hyperplasia				X		
S,S Disease (Sickle Cell Anemia)					X	
S, βeta-Thalassemia					X	
S,C Disease					X	
Biotinidase Deficiency						X
Critical Congenital Heart Disease						X
Cystic Fibrosis						X
Classic Galactosemia						X
Glycogen Storage Disease Type II (Pompe)						X
Hearing Loss						X
Severe Combined Immunodeficiencies						X
Mucopolysaccharidosis Type 1						X
X-linked Adrenoleukodystrophy						X
Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1						X

Recommended Uniform Screening Panel[1]
SECONDARY[2] CONDITIONS [3]
(As of July 2018)

Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Methylmalonic acidemia with homocystinuria	X				
Malonic acidemia	X				
Isobutyrylglycinuria	X				
2-Methylbutyrylglycinuria	X				
3-Methylglutaconic aciduria	X				
2-Methyl-3-hydroxybutyric aciduria	X				
Short-chain acyl-CoA dehydrogenase deficiency		X			
Medium/short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency		X			
Glutaric acidemia type II		X			
Medium-chain ketoacyl-CoA thiolase deficiency		X			
2,4 Dienoyl-CoA reductase deficiency		X			
Carnitine palmitoyltransferase type I deficiency		X			
Carnitine palmitoyltransferase type II deficiency		X			
Carnitine acylcarnitine translocase deficiency		X			
Argininemia			X		
Citrullinemia, type II			X		
Hypermethioninemia			X		
Benign hyperphenylalaninemia			X		
Biopterin defect in cofactor biosynthesis			X		
Biopterin defect in cofactor regeneration			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.
   Nomenclature for Conditions based upon “Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels.” *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.