Recommended Uniform Screening Panel[1] (RUSP)

Core Conditions[2]

(As of September 2018)

Category	Condition	Included in California Newborn Screening
Organic Acid Disorders	Propionic Acidemia	✓
	Methylmalonic Acidemia (Methylmalonyl-CoA Mutase)	✓
	Methylmalonic Acidemia (Cobalamin Disorders)	✓
	Isovaleric Acidemia	✓
	3-Methylcrotonyl-CoA Carboxylase Deficiency	✓
	3-Hydroxy-3-Methylglutaric Aciduria	✓
	Holocarboxylase Synthase Deficiency	✓
	β-Ketothiolase Deficiency	✓
	Glutaric Acidemia Type I	✓
Fatty Acid Oxidation Disorders	Carnitine Uptake Defect	✓
	Medium-chain Acyl-CoA Dehydrogenase Deficiency	✓
	Very Long-chain Acyl-CoA Dehydrogenase Deficiency	✓
	Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency	✓
	Trifunctional Protein Deficiency	✓
Amino Acid Disorders	Argininosuccinic Aciduria	✓
	Citrullinemia Type I	✓
	Maple Syrup Urine Disease	✓
	Homocystinuria	✓
	Classic Phenylketonuria	✓
	Tyrosinemia Type I	✓
Endocrine Disorders	Primary Congenital Hypothyroidism	✓
	Congenital Adrenal Hyperplasia	✓
Hemoglobin Disorders	S,S Disease (Sickle Cell Anemia)	✓
	S, β-Thalassemia	✓
	S,C Disease	✓
Other Disorders	Biotinidase Deficiency	✓
	Cystic Fibrosis[3]	✓
	Classic Galactosemia	✓
	Glycogen Storage Disease Type II (Pompe)	✓
	Mucopolysaccharidosis Type I	✓
	Severe Combined Immunodeficiencies	✓
	X-linked Adrenoleukodystrophy	✓
	Critical Congenital Heart Disease	✸
	Hearing Loss	✸
	Spinal Muscular Atrophy	Planning for 2020

1. https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/uniformscreeningpanel.pdf
2. Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the California Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.
3. Cystic Fibrosis Transmembrane Conductance Regulator (CFTR)-Related Metabolic Syndrome (CRMS) can also be detected by newborn screening (infants with a high level of immunoreactive trypsinogen plus inconclusive CFTR functional and genetic testing)
✸Point-of-care screening tests performed under the auspices of the California Department of Health Care Services