Recommended Uniform Screening Panel¹ (RUSP) Secondary² Conditions³ (California Newborn Screening Panel, as of September 2017)

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Category	Condition	Included in California Newborn Screening
Organic Acid Disorders	2-Methyl-3-Hydroxybutyric Aciduria	✓
	2-Methylbutyrylglycinuria	✓
	3-Methylglutaconic Aciduria	4
	Methylmalonic Acidemia with Homocystinuria	•
	Isobutyrylglycinuria	✓
	Malonic Acidemia	✓
Fatty Acid Oxidation Disorders	Carnitine Acylcarnitine Translocase Deficiency	✓
	Carnitine Palmitoyltransferase I Deficiency	✓
	Carnitine Palmitoyltransferase II Deficiency	✓
	Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency	✓
	Glutaric Acidemia Type II	✓
	Short Chain Acyl-CoA Dehydrogenase Deficiency	1
Amino Acid Disorders	Argininemia	✓
	Biopterin Defect in Cofactor Biosynthesis	✓
	Biopterin Defect in Cofactor Regeneration	✓
	Citrullinemia Type II	✓
	Benign Hyperphenylalaninemia	✓
	Hypermethioninemia	✓
	Tyrosinemia Type II	✓
	Tyrosinemia Type III	✓
Hemoglobin Disorders	Various Other Hemoglobinopathies	✓ See Details in Hemoglobinopathies
Other Disorders	T-Cell Related Lymphocyte Deficiencies	~

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https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpa nel/uniformscreeningpanel.pdf

2. Disorders that can be detected in the differential diagnosis of a core disorder.

3. 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.

California Newborn Screening Panel Secondary¹ Conditions² (Hemoglobinopathies) (As of September 2017)

(AS OF September 2017)			
Category	Condition	Included in California Newborn Screening	
Alpha	Alpha Thalassemia Major	✓	
Thalassemias	Hemoglobin H Disease	✓	
Beta Hemoglobin Variants	Hemoglobin C Disease	×	
	Hemoglobin D Disease	✓	
	Hemoglobin E, E	✓	
	Hemoglobin SD Disease	✓	
	Hemoglobin SE Disease	✓	
	Hemoglobin S, Variant	✓	
	Hemoglobin Variant, Variant	✓	
Beta Thalassemias	Beta Thalassemia Major Hemoglobin C Beta-Thalassemia	✓ ✓	
	Hemoglobin D Beta-Thalassemia	✓	
	Hemoglobin E Beta-Thalassemia	✓	
	Hemoglobin E Delta-Beta- Thalassemia Hemoglobin Variant/Beta- Thalassemia	✓ ✓	
Hereditary Persistence of Fetal Hemoglobin	НРЕН/НРЕН	✓	
	S/HPFH	✓	

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