California Newborn Screening Panel

Additional Secondary¹ Conditions²

(As of September 2018)

Category	Condition	Included in California Newborn Screening
Organic Acid	Ethylmalonic Encephalopathy	\checkmark
Disorders	Formiminoglutamic acidemia	\checkmark
Amino Acid Disorders	Carbamoylphosphate Synthetase Deficiency	✓
	Gyrate Atrophy of the Choroid and Retina	\checkmark
	Hyperornithinemia-Hyperammonemia-	
	Homocitrullinuria Syndrome	\checkmark
	Hyperprolinemia Type I	\checkmark
	Hyperprolinemia Type II	\checkmark
	Ornithine Transcarbamylase Deficiency	\checkmark
	Remethylation Defects (MTHFR, MTR,	
	MTRR, Cbl D v1, Cbl G Deficiencies)	\checkmark
	Tyrosinemia, Transient	✓
Other Disorders	Congenital Adrenal Hyperplasia (11β-	
	Monooxygenase Deficiency)	\checkmark
	Duarte Galactosemia	\checkmark

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

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.