Recommended Uniform Screening Panel¹ (RUSP) Core Conditions² (As of September 2017)

Propionic Acidemia Methylmalonic Acidemia (Methylmalonic Acidemia (Methylmalonic Acidemia (Cobalamin Disorders) Jesualeric Acidemia Organic Acid Disorders 3-Methylcrotonyl-CoA Carboxylase Deficiency 3-Hydroxy-3-Methylglutaric Aciduria Holocarboxylase Synthase Deficiency β-Ketothiolase Deficiency Glutaric Acidemia Type I Carnitine Uptake Defect Medium-chain Acyl-CoA Dehydrogenase Deficiency Very Long-chain Acyl-CoA Dehydrogenase Deficiency Trifunctional Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Maple Syrup Urine Disease Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Fendocrine Disorders Hemoglobin Disorders Biotinidase Deficiency S, Disease (Sickle Cell Anemia) S, C Disease Biotinidase Deficiency Valiase Adrenoleukodystrophy Valiase Deficiency Valiase Adrenoleukodystrophy	Category	Condition	Included in California Newborn Screening
Methylmalonyl-CoA Mutase	Organic Acid	Propionic Acidemia	✓
Methylmalonic Acidemia (Cobalamin Disorders) Organic Acid Disorders 3-Methylcrotonyl-CoA Carboxylase Deficiency 3-Hydroxy-3-Methylglutaric Aciduria Holocarboxylase Synthase Deficiency β-Ketothiolase Deficiency Glutaric Acidemia Type I 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 Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Findocrine Disorders Hemoglobin Disorders S, S Disease Sickle Cell Anemia) S, C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies V Critical Congenital Heart Disease ** ** ** ** ** ** ** ** **		Methylmalonic Acidemia	
Cobalamin Disorders Sovaleric Acidemia Soval		(Methylmalonyl-CoA Mutase)	✓
Organic Acid Disorders 3-Methylcrotonyl-CoA Carboxylase Deficiency 3-Hydroxy-3-Methylglutaric Aciduria Holocarboxylase Synthase Deficiency β-Ketothiolase Deficiency Glutaric Acidemia Type I Carnitine Uptake Defect Medium-chain Acyl-CoA Dehydrogenase Deficiency Very Long-chain Acyl-CoA Dehydrogenase Deficiency Trifunctional Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Hemoglobin Disorders Hemoglobin Disorders Hemoglobin Disorders Hemoglobin Disorders Other Disorders Other Disorders Other Disorders South South South South		Methylmalonic Acidemia	
Disorders 3-Methylcrotonyl-CoA Carboxylase Deficiency 3-Hydroxy-3-Methylglutaric Aciduria Holocarboxylase Synthase Deficiency β-Ketothiolase Deficiency Glutaric Acidemia Type I 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 Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Findacrine Disorders Hemoglobin Disorders Hemoglobin Disorders S, S Disease (Sickle Cell Anemia) S, β-Thalassemia S, C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease ** ** ** ** * ** ** ** ** *		(Cobalamin Disorders)	✓
3-Methylcrotonyl-CoA Carboxylase Deficiency 3-Hydroxy-3-Methylglutaric Aciduria √ Holocarboxylase Synthase Deficiency √ β-Ketothiolase Deficiency √ Glutaric Acidemia Type √ Carnitine Uptake Defect √ Medium-chain Acyl-CoA Dehydrogenase Deficiency √ Very Long-chain Acyl-CoA Dehydrogenase Deficiency √ Very Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase √ Very Long-chain Acyl-CoA Dehydrogenase √ Very Long		Isovaleric Acidemia	✓
3-Hydroxy-3-Methylglutaric Aciduria Holocarboxylase Synthase Deficiency Factothiolase Deficiency Glutaric Acidemia Type	Disorders		
Holocarboxylase Synthase Deficiency β-Ketothiolase Deficiency Glutaric Acidemia Type I Carnitine Uptake Defect Medium-chain Acyl-CoA Dehydrogenase Deficiency Very Long-chain Acyl-CoA Dehydrogenase Deficiency Very Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Trifunctional Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Findocrine Disorders Hemoglobin Disorders Biotinidase Deficiency Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease		3-Methylcrotonyl-CoA Carboxylase Deficiency	✓
B-Ketothiolase Deficiency Glutaric Acidemia Type		3-Hydroxy-3-Methylglutaric Aciduria	✓
Glutaric Acidemia Type I 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 Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Endocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease ** ** Carnitine Uptake Defect ** Medium-chain Acyl-CoA Dehydrogenase /* /* /* /* /* /* /* /* /* /		Holocarboxylase Synthase Deficiency	✓
Carnitine Uptake Defect		β-Ketothiolase Deficiency	✓
Fatty Acid Oxidation Disorders		Glutaric Acidemia Type I	✓
Deficiency Very Long-chain Acyl-CoA Dehydrogenase Deficiency Deficiency Deficiency Deficiency Deficiency Deficiency Deficiency Deficiency Deficiency Trifunctional Protein Deficiency Very Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Very Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Very Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Very Long-chain Acyl-CoA Dehydrogenase Very Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Very Long-chain Acyl-CoA Dehydrogenase Deficiency Very Long-chain Acyl-CoA Dehydrogenase Very Long-chain Acy	Oxidation	Carnitine Uptake Defect	✓
Fatty Acid Oxidation Disorders Very Long-chain Acyl-CoA Dehydrogenase Deficiency Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Trifunctional Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Maple Syrup Urine Disease Disorders Homocystinuria Classic Phenylketonuria ✓ Tyrosinemia Type I ✓ Endocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia ✓ S, S Disease (Sickle Cell Anemia) ✓ S, β-Thalassemia ✓ S, C Disease ✓ Biotinidase Deficiency ✓ Cystic Fibrosis³ ✓ Classic Galactosemia ✓ Severe Combined Immunodeficiencies ✓ V-linked Adrenoleukodystrophy ✓ Critical Congenital Heart Disease **		Medium-chain Acyl-CoA Dehydrogenase	
Oxidation Disorders Very Long-chain Acyl-CoA Dehydrogenase Deficiency Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Argininosuccinic Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Maple Syrup Urine Disease Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Endocrine Disorders Congenital Hypothyroidism Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, β-Thalassemia ✓ S,C Disease ✓ Biotinidase Deficiency ✓ Cystic Fibrosis³ ✓ Classic Galactosemia ✓ Severe Combined Immunodeficiencies ✓ W-Iniked Adrenoleukodystrophy ✓ Critical Congenital Heart Disease **		Deficiency	✓
Deficiency Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency Trifunctional Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Endocrine Disorders Congenital Adrenal Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease		Very Long-chain Acyl-CoA Dehydrogenase	
Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency ✓ Trifunctional Protein Deficiency ✓ Argininosuccinic Aciduria ✓ Citrullinemia Type I ✓ Amino Acid Disorders Maple Syrup Urine Disease ✓ Homocystinuria ✓ Classic Phenylketonuria ✓ Tyrosinemia Type I ✓ Endocrine Disorders Primary Congenital Hypothyroidism ✓ Congenital Adrenal Hyperplasia ✓ S,S Disease (Sickle Cell Anemia) ✓ S, β-Thalassemia ✓ S,C Disease ✓ Biotinidase Deficiency ✓ Cystic Fibrosis³ ✓ Classic Galactosemia ✓ Severe Combined Immunodeficiencies ✓ V-Iinked Adrenoleukodystrophy ✓ Critical Congenital Heart Disease **		Deficiency	✓
Trifunctional Protein Deficiency Argininosuccinic Aciduria Citrullinemia Type I Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Findocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease		Long-chain L-3-Hydroxyacyl-CoA Dehydrogenase	
Argininosuccinic Aciduria Citrullinemia Type I Maple Syrup Urine Disease Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Findocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, B-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis ³ Classic Galactosemia Severe Combined Immunodeficiencies V Critical Congenital Heart Disease		Deficiency	✓
Citrullinemia Type I Amino Acid Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Endocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, B-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies V Critical Congenital Heart Disease		Trifunctional Protein Deficiency	→
Amino Acid Disorders Maple Syrup Urine Disease ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓ ✓		Argininosuccinic Aciduria	✓
Disorders Homocystinuria Classic Phenylketonuria Tyrosinemia Type I Endocrine Disorders Congenital Adrenal Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies V Critical Congenital Heart Disease		Citrullinemia Type I	✓
Classic Phenylketonuria Tyrosinemia Type I Endocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S, B-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies V- Critical Congenital Heart Disease		Maple Syrup Urine Disease	✓
Tyrosinemia Type I Endocrine Disorders Congenital Adrenal Hypothyroidism Congenital Adrenal Hyperplasia Hemoglobin Disorders S,S Disease (Sickle Cell Anemia) S,β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease		Homocystinuria	✓
Endocrine Disorders Primary Congenital Hypothyroidism Congenital Adrenal Hyperplasia Hemoglobin Disorders S,S Disease (Sickle Cell Anemia) S,β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies V-Iinked Adrenoleukodystrophy Critical Congenital Heart Disease		Classic Phenylketonuria	✓
Disorders Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S,β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease		Tyrosinemia Type I	✓
Disorders Congenital Adrenal Hyperplasia S,S Disease (Sickle Cell Anemia) S,β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease	Endocrine	Primary Congenital Hypothyroidism	✓
Hemoglobin Disorders S, β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease **	Disorders		✓
Hemoglobin Disorders S, β-Thalassemia S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies V-linked Adrenoleukodystrophy Critical Congenital Heart Disease	•	S.S Disease (Sickle Cell Anemia)	✓
Disorders S,C Disease Biotinidase Deficiency Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies X-linked Adrenoleukodystrophy Critical Congenital Heart Disease *		,	✓
Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies ✓ Other Disorders X-linked Adrenoleukodystrophy Critical Congenital Heart Disease **		7.1	✓
Cystic Fibrosis³ Classic Galactosemia Severe Combined Immunodeficiencies ✓ Other Disorders X-linked Adrenoleukodystrophy Critical Congenital Heart Disease **	Other Disorders	Biotinidase Deficiency	✓
Classic Galactosemia Severe Combined Immunodeficiencies ✓ Other Disorders X-linked Adrenoleukodystrophy Critical Congenital Heart Disease **			✓
Severe Combined Immunodeficiencies ✓ Other Disorders X-linked Adrenoleukodystrophy Critical Congenital Heart Disease **			· · · · · · · · · · · · · · · · · · ·
Other Disorders X-linked Adrenoleukodystrophy ✓ Critical Congenital Heart Disease *			√
Critical Congenital Heart Disease			· · · · · · · · · · · · · · · · · · ·
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Glycogen Storage Disease Type II (Pompe) Planning for 2018			Planning for 2019
Mucopolysaccharidosis Type I Planning for 2018			

^{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