California Department of Public Health
Genetic Disease Screening Program
Newborn Screening Program

Disorders Detectable by NBS Program as of June 24, 2020

I. Cystic Fibrosis

II. Endocrine Disorders
- primary congenital hypothyroidism
- variant hypothyroidism
- congenital adrenal hyperplasia-salt wasting (21-hydroxylase deficiency)
- congenital adrenal hyperplasia-simple virilizing (21-hydroxylase deficiency)

III. Metabolic Disorders (via tandem mass spectrometry (MS/MS) Screening)

A. Amino Acid Disorders
- classical phenylketonuria (PKU)
- variant PKU
- guanosine triphosphate cyclohydrolase 1 (GTPCH) deficiency (biopterin deficiency)
- 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency (biopterin deficiency)
- dihydropteridine reductase (DHPR) deficiency (biopterin deficiency)
- pterin-4α-carbinolamine dehydratase (PCD) deficiency (biopterin deficiency)
- argininemia/arginase deficiency
- argininosuccinic acid lyase deficiency (ASAL deficiency)
- citrullinemia, Type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- citrullinemia, Type II (citrin deficiency)
- gyrate atrophy of the choroid and retina
- homocitrullinuria, hyperornithinemia, hyperammonemia – HHH
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- methionine adenosyltransferase deficiency (MAT deficiency)
- maple syrup urine disease – (MSUD)
- prolinemia
- tyrosinemia, Type I, II, III, and transient
- ornithine transcarbamylase deficiency (OTC deficiency)
- remethylation defects (MTHFR, MTR, MTRR, Cbl D v1, Cbl G deficiencies)

B. Organic Acid Disorders
- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
- 3-methylglutaconic aciduria (MGA), Type I (3-methylglutaconyl-CoA hydratase deficiency)
- beta-ketothiolase deficiency (BKT)
- ethylmalonic encephalopathy (EE)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- malonic aciduria
- methylmalonic acidemia, mut –
- methylmalonic acidemia, mut 0
- methylmalonic acidemia (Cbl A, B)
- methylmalonic acidemia (Cbl C, D)
- multiple carboxylase deficiency (MCD)
- propionic acidemia (PA)

C. Fatty Acid Oxidation Disorders
- carnitine transporter deficiency
- carnitine-acylcarnitine translocase deficiency (CAT deficiency)
- carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
- carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
- long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency)
- medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- medium/short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD deficiency)
- multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type-2 (GA-2)
- short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
- trifunctional protein deficiency (TFP deficiency)
- very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)
- formiminoglutamic acid (FIGLU) disorder

IV. Other Metabolic Disorders
A. classical galactosemia
B. biotinidase deficiency

V. Severe Combined Immunodeficiency (SCID)

VI. Hemoglobin Disorders
- sickle cell anemia (Hb S/S disease)
- sickle C disease (Hb S/C disease)
- sickle D disease (Hb S/D disease)
- sickle E disease (Hb S/E disease)
- Hb S/ hereditary persistence of fetal hemoglobin (Hb S/HPFH)
- sickle cell disease variant (other sickle cell disease, Hb S/V)
- Hb S/ $Beta^0$ thalassemia
- Hb S/$Beta^+$ thalassemia
- Hb C disease (Hb CC)
- Hb D disease (Hb DD)
- alpha thalassemia major
- Hb H disease (3 gene deletion)
- Hb H/ Constant Spring disease
- beta thalassemia major
- Hb E/ $Beta^0$ thalassemia
- Hb E/$Beta^+$ thalassemia
- Hb E/ Delta Beta thalassemia
- Hb C/ $Beta^0$ thalassemia
- Hb C/$Beta^+$ thalassemia
- Hb D/ $Beta^0$ thalassemia
- Hb D/$Beta^+$ thalassemia
- Hb Variant/ $Beta^0$ thalassemia
- Hb Variant/$Beta^+$ thalassemia
- other hemoglobinopathies (Hb variants)

VII. Adrenoleukodystrophy (ALD)

VIII. Mucopolysaccharidosis Type I (MSP I)

IX. Pompe

X. Spinal Muscular Atrophy (SMA)

[i]Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period the 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.

(Updated by ASC 97 on 7/10/20)