

ATTACHMENT I to SCOPE OF WORK  
*California Department of Health Services*  
*Genetic Disease Branch*  
*Newborn Screening Program*  
***Metabolic Disorders Detectable by NBS Program's of July 2007***

I. Metabolic Disorders

A. *Carbohydrate Disorders*

- classical galactosemia

B. *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-4 $\alpha$ -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)
- tyrosinemia

C. *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)

#### *D. 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)

#### *E. Biotinidase Deficiency*

- Profound biotinidase deficiency

*F.* In addition, metabolic centers need to report on additional diseases that may be identified but are not on this list including Duarte galactosemia.

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<sup>i</sup> 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.