

**California Newborn Screening Program**  
Disorders Detectable by Tandem Mass Spectrometry (MS/MS)  
Using Newborn Screening Dried Blood Spots (to be added to NBS Program mid-2005)

**Primary Disorders (41)**

***Amino Acid Disorders (14)***

- argininemia/arginase deficiency
- argininosuccinic acid lyase deficiency (ASAL deficiency)
- bipterin disorders (4)
- citrullinemia type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- citrullinemia type II (citrin deficiency)
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- hypermethioninemia/MAT deficiency
- hyperphenylalaninemia – Classical Phenylketonuria (PKU)
- hyperphenylalaninemia – variant PKU
- hyperphenylalaninemia – benign PKU
- maple syrup urine disease – (MSUD)

***Organic Acid Disorders (17)***

- 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-methylglutaconic aciduria (MGA) type II
- 3-methylglutaconic aciduria (MGA) type III
- 3-methylglutaconic aciduria (MGA) type IV
- beta-ketothiolase deficiency (BKD)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- methylmalonic acidemia, mut –
- methylmalonic acidemia, mut 0
- methylmalonic acidemia (Cbl A, B)
- methylmalonic acidemia (Cbl C, D)
- propionic acidemia (PA)
- multiple carboxylase deficiency (MCD)

***Fatty Acid Oxidation Disorders (10)***

- 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)
- 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)

### **Secondary Disorders (13)**

- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- 5-oxoprolinuria
- ethylmalonic encephalopathy(EE)
- homocitrullinuria, hyperornithinemia, hyperammonemia -HHH
- gyrate atrophy of the choroid and retina
- malonic aciduria
- non-ketotic hyperglycinemia
- prolinemia type I
- prolinemia type II
- tyrosinemia type I (TYR-I)
- tyrosinemia type II (TYR-II)
- tyrosinemia type III (TYR-III)
- tyrosinemia, transient