

**North Carolina State Laboratory of Public Health**  
**Newborn Screening Laboratory**

List of disorders

April 21, 2006

**Disorders detected by tandem mass spectrometry (MS/MS)**

**Amino Acid disorders**

- Argininosuccinic aciduria (ASA)
- Citrullinemia (CIT I)
- Homocystinuria (cystathionine beta synthase) (HCY)
- Maple syrup urine disease / Branched-chain ketoacid dehydrogenase (MSUD)
- Phenylketonuria / Hyperphenylalaninemia (PKU)
- Tyrosinemia type II (TYR-II)
- Tyrosinemia type III (TYR-III)\*

**Organic Acid Disorders**

- Glutaric acidemia type I (GA-I)
- Multiple carboxylase deficiency (MCD) \*
- 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG) \*
- Isobutyryl-CoA dehydrogenase deficiency (IBD)
- Isovaleric acidemia / Isovaleryl-CoA dehydrogenase deficiency (IVA)
- Beta-ketothiolase (BKT) / Short-chain keto acylthiolase deficiency (SKAT)
- Methylmalonic aciduria (MMA)
- 2-Methylbutyryl-CoA dehydrogenase deficiency (2-MBD)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- Propionic acidemia (PPA, PROP)

**Fatty Acid Disorders**

- Carnitine/acylcarnitine translocase deficiency (CAT) \*
- Carnitine palmitoyltransferase II deficiency (CPT II)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Multiple acyl-CoA dehydrogenase deficiency (GA-II)
- Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- Trifunctional protein deficiency (TFP) \*
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

**Disorders detected by biochemical and other technologies**

Biotinidase deficiency (BIO)

Congenital adrenal hyperplasia (CAH)

Galactosemia/ galactose-1-phosphate uridyl transferase deficiency (GALT)

Primary congenital hypothyroidism (CH)

Hemoglobin C disease (FC)

Hemoglobin E disease (FE)

Sickle cell disease (FS, HB S/S)

Sickle/hemoglobin C disease (FSC, HB S/C)

Sickle/hemoglobin E disease (FSE, HB S/E)

\* no cases yet detected by North Carolina Newborn Screening Laboratory