DHMA Alphabetical List of Disorders on the Newborn Screen

Argininemia (Arginase deficiency)
Argininosuccinate aciduria
Beta Ketothiolase (Mitochondrial Acetyl-CoA Thiolase Deficiency
Biotinidase Deficiency
Carnitine Acyl-Carnitine Translocase Deficiency
Carnitine Palmitoyltransferase Deficiency Type 1 (CPT I)
Carnitine Palmitoyltransferase Deficiency Type 1 CPT II
Carnitine Uptake Deficiency
Citrullinemia
Cobalamin C deficiency
Congenital Adrenal Hyperplasia
Congenital Hypothyroidism
Cystic Fibrosis
2,4-dienoyl-CoA Reductase
Galactosemia
Glutaric Acidemia
Glutaric Acidemia Type II
Homocystinuria
3-Hydroxy-3Methylglutaric aciduria
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleric Acidemia
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
Malonic Acidemia
Maple syrup urine disease (Branched-chain ketoacid dehydrogenase deficiency)
Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
2-Methyl-3Hydroxybutyryl-CoA Dehydrogenase Deficiency
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3 Methylcrotonyl- CoA carboxylase deficiency (3-MCC)
Methylmalonic Acidemia
Multiple Carboxylase Deficiency
Phenylketonuria/ hyperphenylalaninemia
Propionic Acidemia
Short chain acyl-CoA dehydrogenase deficiency (SCADD)
Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency (SCHADD)
Sickle Cell Anemia
Trifunctional Protein Deficiency
Tyrosinemia Type I
Tyrosinemia Type I/II/III
Very long chain acyl-CoA Dehydrogenase (VLCADD)

Source:*http://phpa.dhmh.maryland.gov/genetics/Pages/NBS_Disorders_Alphabetical_List.aspx*

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