



Newborn Metabolic Screening Disorder List

Adrenoleukodystrophy - 2019

Amino Acid Disorders

- Homocystinuria (HCU)/Hypermethioninemia - 2002
- Maple syrup urine disease (MSUD) - 2002
- Phenylketonuria (PKU)/Hyperphenylalaninemia - 1965
- Tyrosinemia (TYR) - 2002
- 5-Oxoprolinuria (SOXP) - 2002

Biotinidase deficiency (BIO) - 1986

Cystic fibrosis (CF) - 2008

Endocrine Disorders

- Congenital adrenal hyperplasia (CAH) - 1987
- Congenital hypothyroidism (HYP) - 1979

Fatty Acid Oxidation Disorders - 2002

- Carnitine/acylcarnitine translocase deficiency (CACT)
- Carnitine palmitoyl transferase deficiency, type 2 (CPT2)
- Carnitine palmitoyl transferase deficiency, type 1A (CPT1A)
- Carnitine uptake defect (CUD)
- Glutaric aciduria, type 2 (GA2)/Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Isobutyryl-CoA dehydrogenase deficiency (IBCD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHADD)
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCADD)
- Trifunctional protein deficiency (TFP)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)

Galactosemia (GALT) - 1984

Hemoglobinopathies

- Sickle cell disease, trait conditions and other hemoglobinopathies (SC) - 1989
- Alpha thalassemia - 2008
- Beta thalassemia major - 1989

Lysosomal storage diseases (LSD)

- Pompe - 2015
- Fabry - 2015
- Gaucher - 2015
- Krabbe - 2017
- Niemann-Pick - 2015
- MPS I (Hurler's Syndrome) - 2015
- MPS II (Hunter's Syndrome) - 2017

Organic Acid Disorders - 2002

- 2-methylbutyryl-CoA dehydrogenase deficiency (2MBCD)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
- 3-methylglutaconic aciduria (3MGA)
- Beta-ketothiolase deficiency (BKT)
- Glutaric aciduria, type 1 (GA1)
- Isovaleric acidemia (IVA)
- Malonic aciduria (MA)
- Methylmalonic acidemia (MMA)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PA)

Severe Combined Immune Deficiency - 2014

Urea Cycle Disorders - 2002

- Argininemia (ARG)
- Argininosuccinic aciduria (ASA)
- Citrullinemia (CIT)