

Newborn Metabolic Screening Disorder List

Amino Acid Disorders (AA)

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

Biotinidase deficiency (BIO) 1986

Cystic fibrosis (CF) 2008

Endocrine Disorders

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

Fatty Acid Oxidation Disorders (FA) 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 (HGB)

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

Lysosomal storage diseases (LSD) 2015

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

**Although screening for Krabbe and MPS II is required by the Newborn Metabolic Screening Act [410 ILCS 240], the testing method for these disorders are still under development.*

Organic Acid Disorders (OA) 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 (SCID) 2014

Urea Cycle Disorders 2002

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