



2012 Newborn Screening Panel

Amino Acid Disorders

Arginemia (ARG, Arginase deficiency)
Argininosuccinate acidemia (ASA)
Defects of biopterin cofactor biosynthesis (BIOPT-BS)
Defects of biopterin cofactor regeneration (BIOPT-REG)
Citrullinemia type I (CIT-I, argininosuccinate synthetase)
Citrullinemia type II (CIT-II, citrin deficiency)
Homocystinuria (HCY, cystathionine beta synthase)
Hyperphenylalaninemia (H-PHE)
Hypermethioninemia (MET, I/III deficiency)
Maple Syrup Urine Disease (MSUD, branched-chain ketoacid dehydrogenase)
Phenylketonuria
Tyrosinemia type I (TYR-I)
Tyrosinemia type II (TYR-II)
Tyrosinemia type III (TYR-III)

Fatty Acid Oxidation Disorders

Carnitine acylcarnitine translocase deficiency (CACT)
Carnitine uptake defect (CUD, carnitine transport defect)
Carnitine palmitoyltransferase deficiency I (CPT-1a)
Carnitine palmitoyltransferase deficiency II (CPT-II)
Dienoyl-CoA reductase deficiency (DE-RED)
Glutaric acidemia type II (GA-II)
Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
Trifunctional protein deficiency (TFP)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders

2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG, SBCAD)
3-Hydroxy 3-methylglutaric aciduria (HMG, 3-Hydrox 3-methylglutaryl-CoA lyase)
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
3-Methylglutaconic aciduria (3MGA, Type I hydratase deficiency)
Beta ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacyl thiolase)
Glutaric acidemia type I (GA-1)
Isobutyryl-CoA dehydrogenase deficiency (IBG)
Isovaleric acidemia (IVA, Isovaleryl-CoA dehydrogenase deficiency)
Malonic acidemia (MAL, Malonyl-CoA decarboxylase)
Methylmalonic acidemia (CBL A,B; Vitamin B12 Disorders)
Methylmalonic acidemia (CBL C,D)
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)
Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
Propionic acidemia (PROP, propionyl-CoA carboxylase)

Endocrine Disorders

Congenital adrenal hyperplasia (CAH)
Congenital hypothyroidism (CH)

Hemoglobinopathies

Sickle cell disease (HB S/S)
Sickle-C disease (HB S/C)
S- β thalassemia
Variant hemoglobinopathies

Others

Biotinidase deficiency (BIO)
Classic galactosemia (GALT)
Galactose epimerase deficiency (GALE)
Galactokinase deficiency (GALK)
Cystic fibrosis
Hearing