MINNESOTA NEWBORN SCREENING PANEL

Metabolic Disorders

Amino Acid Profile

Arginemia (ARG)
Argininosuccinate acidemia (ASA)
Biopterin cofactor defects (BIOPT-BS and BIOPT-REG)
Citrullinemia type I and II (CIT and CIT-II)
Guanidinoacetate methyltransferase (GAMT) deficiency
Homocystinuria (HCY)
Hypermethioninemia (MET)
Hyperphenylalaninemia (H-PHE)
Maple syrup urine disease (MSUD)
Phenylketonuria (PKU)
Tyrosinemia type I, II, and III (TYR-I, TYR-II, and TYR-III)

Acylcarnitine profile

2-Methyl-3-hydroxybutyric acidemia (2M3HBA)
2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG, SBCAD)
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
3-Methylglutaconyl-CoA hydratase deficiency (3MGA)
Beta ketothiolase deficiency (BKT)
Carnitine acylcarnitine translocase deficiency (CACT)
Carnitine palmitoyltransferase deficiency I (CPT-1)
Carnitine palmitoyltransferase deficiency II (CPT-II)
Carnitine uptake defect (CUD)
Dienoyl-CoA reductase deficiency (DE-RED)
Glutaric acidemia type I (GA-1)
Glutaric acidemia type II (GA-II)
Isobutyryl-CoA dehydrogenase deficiency (IBD, IBG)
Isovaleric acidemia (IVA)
Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
Malonic acidemia (MAL)
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
Medium/short-chain hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
Medium-chain keto acyl-CoA thiolase deficiency (MCKAT)
Methylmalonic acidemia (mutase deficiency)
Methylmalonic acidemia (cobalamin disorders A and B)
Methylmalonic acidemia with homocystinuria
Multiple CoA carboxylase deficiency (MCD)
Propionic acidemia (PROP)
Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
Trifunctional protein deficiency (TFP)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Additional Metabolic Disorders

Biotinidase deficiency (BIOT)
Galactokinase deficiency (GALK)
Galactoepimerase deficiency (GALE)
Galactosemia (GALT)
X-linked adrenoleukodystrophy (X-ALD)

Endocrine Disorders

Congenital adrenal hyperplasia (CAH)
Congenital hypothyroidism (CH)

Hemoglobin Disorders

Alpha thalassemia major
Hemoglobin H disease
Sickle cell disease (Hb S/S)
Sickle-C disease (Hb S/C)
Sickle βeta-plus thalassemia (Hb S/BTh)
Variant hemoglobinopathies

Lysosomal Storage Disorders

Krabbe disease
Mucopolysaccharidosis type I (MPS I)
Pompe disease

Other Disorders

Congenital cytomegalovirus (cCMV)
Cystic fibrosis (CF)
Duchenne muscular dystrophy (DMD)
Severe combined immunodeficiency (SCID)
Spinal muscular atrophy (SMA)
T-cell lymphopenia (TCL)

Point-of-Care Screening

Critical congenital heart disease (CCHD)
Hearing loss

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