

DISORDERS DETECTED BY LAPS SUOMEN METABOLIC SCREENING TEST

1STEP [COMPREHENSIVE]

Disorders Detected by Tandem Mass Spectrometry

Non-ketotic hyperglycinemia (NKH) cannot be reliably detected by MS/MS technology.

ACYLCARNITINE PROFILE

Fatty Acid Oxidation Disorders

Carnitine/Acylcarnitine Translocase Deficiency (Translocase)
 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD)
 Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
 Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or
 Glutaric Acidemia-Type II)
 Neonatal Carnitine Palmitoyl Transferase Deficiency-Type II (CPT-II)
 Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
 Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
 Trifunctional Protein Deficiency (TFP Deficiency)
 Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

Organic Acid Disorders

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)
 Glutaric Acidemia-Type I (GA I)
 Isobutyryl-CoA Dehydrogenase Deficiency
 Isovaleric Acidemia (IVA)
 Acute onset
 Chronic
 2-Methylbutyryl-CoA Dehydrogenase Deficiency
 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Def.)
 3-methylglutaconyl-CoA Hydratase Deficiency
 Methylmalonic Acidemias
 Methylmalonyl-CoA Mutase Deficiency 0
 Methylmalonyl-CoA Mutase Deficiency +
 Some Adenosylcobalamin Synthesis Defects
 Maternal Vitamin B12 Deficiency
 Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
 (3-Ketothiolase Def.)
 Propionic Acidemia (PA)
 Acute onset
 Late onset
 Multiple CoA Carboxylase Deficiency

AMINO ACID PROFILE

Amino Acid Disorders

Argininemia
 Argininosuccinic Aciduria (ASA Lyase Deficiency)
 Acute onset
 Late onset
 Citrullinemia (ASA Synthetase Deficiency)
 Acute onset
 Late onset
 Homocystinuria²
 Hypermethioninemia
 Maple Syrup Urine Disease (MSUD)
 Classical MSUD
 Intermediate MSUD
 Phenylketonuria (PKU)
 Classical PKU
 Hyperphenylalaninemia
 Biotin Cofactor Deficiencies (4)
 Tyrosinemia
 Transient Neonatal Tyrosinemia
 Tyrosinemia Type II (Tyr II)
 Tyrosinemia Type III (Tyr III)

OTHER ABNORMAL PROFILES

Hyperalimentation
 Liver Disease
 Medium Chain Triglyceride (MCT) Oil Administration
 Presence of EDTA Anticoagulants in blood specimen
 Treatment with Benzoate, Pyvalic Acid, or Valproic Acid

Older and High Risk Patients Only (children who are ill or at least 7 days of age)

Carbamoylphosphate Synthetase Deficiency (CPS Def.)
 Carnitine Palmitoyl Transferase Deficiency Type I (CPT-I)
 2,4-Dienoyl-CoA Reductase Deficiency
 5-Oxoprolinuria (Pyroglutamic Aciduria)

Hyperammonemia, Hyperornithinemia, Homocitrullinemia
 Syndrome (HHH)
 Hyperornithinemia with Gyral Atrophy
 Malonic Aciduria

Disorders Detected by Other Technologies

Biotinidase Deficiency
Complete Deficiency
Partial Deficiency

Congenital Adrenal Hyperplasia
Salt Wasting 21-Hydroxylase Deficiency
Simple Virilizing 21-Hydroxylase Deficiency

Cystic Fibrosis (not valid after 3 months of age)

Galactosemia
Galactokinase Deficiency
Galactose-1-Phosphate Uridyltransferase Def.
Galactose-4-Epimerase Deficiency

Glucose-6-Phosphate Dehydrogenase Deficiency

Congenital Hypothyroidism (not valid after 2 months of age)

Sickle Cell Hemoglobinopathies
Hemoglobin S Disease
Hemoglobin S/C Disease
Hemoglobin S/Beta-Thalassemia Disease
Hemoglobin C Disease
Hemoglobin E Disease

1. The analyses conducted by Pediatrx Screening produce results that can be used by qualified physicians in the diagnosis of disorders described herein. While evidence of these conditions will be detected in the vast majority of affected individuals, due to genetic variability, it may not be detected in all.
2. Due to genetic variability, the possible lack of detectability in newborns for this disorder, and/or statistical uncertainty, results may not detect the disorder in a higher percentage of the sampled population, when compared with the results achievable in the newborn screening program, as a whole.