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
Homocystinuria[2]
Hypermethioninemia
Maple Syrup Urine Disease (MSUD)
Classical MSUD
Intermediate MSUD
Phenylketonuria (PKU)
Classical PKU
Hyperphenylalaninemia
Biopterin 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

Laps Suomen
INNOVIDEA OY
Pietarinkatu 7 LH 2, FIN-00140 Helsinki
p. 040 181 1888
f. 040 181 1000
asiakaspalvelu@LapsSuomen.fi
www.LapsSuomen.fi

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 Pediatrix 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

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