

DISORDERS INCLUDED IN THE STEPONE® NEWBORN SCREENING PANEL

DISORDERS DETECTED BY TANDEM MASS SPECTROMETRY

Acylcarnitine Profile

Fatty Acid Oxidation Disorders

Carnitine/Acylcarnitine Translocase Deficiency
Carnitine Palmitoyl Transferase Deficiency Type I¹
3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
2,4-Dienoyl-CoA Reductase Deficiency¹
Medium Chain Acyl-CoA Dehydrogenase Deficiency
Multiple Acyl-CoA Dehydrogenase Deficiency
Neonatal Carnitine Palmitoyl Transferase Deficiency Type II
Short Chain Acyl-CoA Dehydrogenase Deficiency
Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency
Trifunctional Protein Deficiency
Very Long Chain Acyl-CoA Dehydrogenase Deficiency

Organic Acid Disorders

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
Glutaric Acidemia Type I
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleric Acidemia
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3-Methylcrotonyl-CoA Carboxylase Deficiency
3-Methylglutaconyl-CoA Hydratase Deficiency
Methylmalonic Acidemias
 Methylmalonyl-CoA Mutase Deficiency
 Some Adenosylcobalamin Synthesis Defects
 Maternal Vitamin B12 Deficiency
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
Propionic Acidemia
Multiple CoA Carboxylase Deficiency
Malonic Aciduria

X-Linked Adrenoleukodystrophy (X-ALD)

Amino Acid Profile

Amino Acid Disorders

Argininemia
Argininosuccinic Aciduria
5-Oxoprolinuria¹
Carbamoylphosphate Synthetase Deficiency¹
Citrullinemia
Homocystinuria
Hypermethioninemia
Hyperammonemia, Hyperornithinemia, Homocitrullinuria
 Syndrome¹
Hyperornithinemia with Gyral Atrophy¹
Maple Syrup Urine Disease
Phenylketonuria
 Classical/Hyperphenylalaninemia
 Biopterin Cofactor Deficiencies
Tyrosinemia
 Transient Neonatal Tyrosinemia
 Tyrosinemia Type I²
 Tyrosinemia Type II
 Tyrosinemia Type III

Other Observations

Hyperalimentation
Liver Disease
Medium Chain Triglyceride Oil Administration
Presence of EDTA Anticoagulants in blood specimen
Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
Carnitine Uptake Deficiency¹

Lysosomal Storage Disorders (LSD)

Fabry, Gaucher, Krabbe Disease, Mucopolysaccharidosis
Type I (MPS-I), Niemann-Pick, and Pompe

DISORDERS DETECTED BY OTHER TECHNOLOGIES

Biotinidase Deficiency
 Complete Deficiency
 Partial Deficiency
Congenital Adrenal Hyperplasia
 Salt Wasting 21-Hydroxylase Deficiency
 Simple Virilizing 21-Hydroxylase Deficiency
Congenital Hypothyroidism
Cystic Fibrosis (*not valid after 90 days of age*)^{*}

Galactosemia
 Galactokinase Deficiency
 Galactose-1-Phosphate Uridyltransferase Deficiency
 Galactose-4-Epimerase Deficiency
Glucose-6-Phosphate Dehydrogenase Deficiency
Severe Combined Immunodeficiency (SCID)
Sickle Cell and other Hemoglobinopathies
 Hemoglobin S, S/C, S/Beta-Thalassemia, C, & E Diseases
Spinal Muscular Atrophy (SMA)

The analyses conducted by PerkinElmer Genetics produce results that can be used by qualified physicians in the diagnosis of disorders described herein. Evidence of these conditions can be detected in the vast majority of affected individuals; however, due to genetic variability, age of patient at time of specimen collection, quality of specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

1 There is a lower probability of detection of this condition during the immediate newborn period.

2 Succinylacetone (SUAC) is the primary marker for Tyrosinemia Type 1.

* For information on DNA Carrier Testing for children over 90 days of age, please call 866.463.6436.