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 I[1]
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
- 2,4-Dienoyl-CoA Reductase Deficiency[1]
- 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 Deficiency[1]
- Citrullinemia
- Homocystinuria
- Hypermethioninemia
- Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome1
- Hyperornithinemia with Gyral Atrophy[1]
- Maple Syrup Urine Disease
- Phenylketonuria
  - Classical/Hyperphenylalaninemia
  - Biopterin Cofactor Deficiencies
- Tyrosinemia
  - Transient Neonatal Tyrosinemia
  - Tyrosinemia Type I[2]
  - 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 Deficiency[1]

Lysosomal Storage Disorders (LSD)

- Fabry, Gaucher, Krabbe Disease, Mucopolysaccharidosis Type I (MPS-I), Niemann-Pick (A/B), 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.

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