Trained physicians use the results of StepOne® expanded newborn Screening for the diagnosis and early treatment of many genetic diseases.

In the vast majority of patients signs of the relevant diseases are detected. However, genetic variability, health conditions, and the patient's age at the time of testing can lead to false negative results. If the sample is taken immediately after birth, the probability of a false negative is practically zero.

Disorders Detected by Tandem Mass Spectrometry

**Acylcarnitine Profile**

**Fatty Acid Oxidation Disorders**

- Carnitine/Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type II
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
- 2,4-Dienoyl-CoA Reductase Deficiency
- 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
Amino Acid Profile

Amino Acid Disorders

• Argininemia
• Argininosuccinic Aciduria
• 5-Oxoprolinuria
• Carbamoylphosphate Synthetase Deficiency
• Citrullinemia
• Homocystinuria
• Hypermethioninemia
• Hyperammononemia, Hyperornithinemia, Homocitrullinemia Syndrome
• Hyperornithinemia with Gyral Atrophy
• Maple Syrup Urine Disease
• Phenylketonuria
  · Classical
  · Hyperphenylalaninemia
  · Biopterin Cofactor Deficiencies
• Tyrosinemia
  · Transient Neonatal Tyrosinemia
  · Tyrosinemia Type I
  · Tyrosinemia Type II
  · Tyrosinemia Type III

Other

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

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 (not valid after 2 months of age)
• Cystic Fibrosis (not valid after 3 months of age)
• Galactosemia
  · Galactokinase Deficiency
  · Galactose-1-Phosphate Uridyltransferase Deficiency
  · Galactose-4-Empimerase Deficiency
• Glucose-6-Phosphate Dehydrogenase Deficiency
• Sickle Cell & other Hemoglobinopathies
  (Hemoglobin S, S/C, S/Beta-Thalassemia, C, E Diseases)
• Severe Combined Immunodeficiency (SCID)

Source:

• PerkinElmer Genetics, StepOne® Newborn Screening - Handbook of Metabolic and Other Inherited Disorders, 2008