

First Step Newborn Screening Panel

First Step will screen babies for over 45 Inborn Errors of Metabolism (IEM) by Tandem Mass Spectrometry and Biochemical Analysis.

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

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 disease
- Phenylketonuria
 - Classical / Hyperphenylalaninemia
 - Biopterin Cofactor Deficiencies
- Tyrosinemia
 - Transient Neonatal Tyrosinemia
 - Tyrosinemia Type I¹
 - Tyrosinemia Type II
 - Tyrosinemia Type III

Other

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

DISORDERS DETECTED BY OTHER TECHNOLOGIES

- Galactosemia
- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD
- Cystic Fibrosis