

# 1<sup>st</sup> STEP Newborn Screening Panel

NeoGen Labs screens newborns for over 50 Inborn Errors of Metabolism (metabolic disorders) from a dried blood spot collected 24 hours AFTER birth.

## Disorders Detected By Tandem Mass Spectrometry Acylcarnitine Profile - Fatty Acid Oxidation Disorders

- Carnitine / Acylcarnitine Translocase Deficiency
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Neonatal Carnitine Palmitoyl Transferase Deficiency Type II
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type I<sup>1</sup>
- 2,4-Dienoyl-CoA Reductase Deficiency<sup>1</sup>
- Multiple Acyl-CoA Dehydrogenase Deficiency
- Short-chain Acyl-CoA Dehydrogenase Deficiency
- Trifunctional Protein Deficiency
- Short chain Hydroxy Acyl-CoA Dehydrogenase Deficiency

## Acylcarnitine Profile - 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<sup>1</sup>
- Carbamoylphosphate Synthetase Deficiency<sup>1</sup>
- Citrullinemia
- Homocystinuria
- Hypermethioninemia
- Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome<sup>1</sup>
- Hyperornithinemia with Gyral Atrophy<sup>1</sup>
- Maple Syrup Urine Disease
- Phenylketonuria
  - Classical / Hyperphenylalaninemia
  - Biopterin Cofactor Deficiencies
- Tyrosinemia
  - Transient Neonatal Tyrosinemia
  - Tyrosinemia Type I<sup>1</sup>
  - Tyrosinemia Type II
  - Tyrosinemia Type III

## Other

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

<sup>1</sup> There is a lower probability of detection of this disorder during the immediate newborn period.

## Disorders Detected By Other Technologies

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



Giving babies the best start in life!



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# 1<sup>st</sup> STEP Newborn Screening Test

What...  
Why...  
When...  
Where...  
How...

# 1<sup>st</sup> STEP Newborn Screening Test

\*A newborn screening test at birth identified Ayden with G6PD deficiency. It is a very simple disorder to treat. Today, he is a healthy five year old who loves chocolates, icecreams and cakes.