



As of July 1, 2009, we are making changes to the panels we offer based on feedback received. Since Cystic Fibrosis (CF) screen is not valid after 2 months of birth and Biotinidase (BIOT) is not valid after 28 days of birth, we have reconstituted our panels to take this into account.

We first started offering Newborn Screening (NBS) services over 21 months ago and have been faced with increased costs but have held prices. Unfortunately, we are left with no choice but to revise our prices. The price for the Tandem Mass Spectrometry panel remains unchanged. The prices of other panels have changed slightly to reflect added disorders and higher input costs.

## Screening Panels

### MS - Mass Spectrometry Panel (45+ Disorders)

(45 IEMs, includes Fatty Acid Oxidation Disorders, Amino Acid Disorders, and Organic Acid Disorder panels)

### Bio - Biochemical Panel (6 Disorders)

CH, CAH, G6PD, GALT, CF, BIOT

- 1. First Step PLUS** (Over 51 Disorders)
  - MS + Bio (Rs. 4,250/-)
  - Infants, 0 to 2 months old
  - Biotinidase is valid only until 28 days after birth
- 2. First Step** (Over 49 Disorders)
  - MS + CH + CAH + GALT + G6PD (Rs. 3,975/-)
  - Infants, Over 2 months old
- 3. First Step MS** (Over 45 Disorders)
  - MS (Rs. 3,250/-)
  - No Age limit
- 4. First Step Bio6** (6 Disorders)
  - CH + CAH + GALT + G6PD + CF + BIOT (Rs. 2,000/-)
  - Infants, 0 to 2 months old
  - Biotinidase is valid only until 28 days after birth
- 5. First Step Bio4** (4 Disorders)
  - CH + CAH + GALT + G6PD (Rs. 1,500/-)
  - No Age limit
- 6. First Step CF**
  - CF (Rs. 750/-)
  - Infants, 0 to 2 months old

Please call or e-mail us if you need to do individual tests or combinations that are not listed above. Page 2 presents the detailed panel we offer

## Biotinidase Deficiency (BIOT)

Biotinidase deficiency is an inherited disorder in which the body is unable to reuse and recycle the vitamin biotin. This disorder is classified as a multiple carboxylase deficiency, a group of disorders characterized by impaired activity of certain enzymes that depend on biotin.

The signs and symptoms of BIOT typically appear within the first few months of life, but the age of onset varies. Children with profound BIOT, the more severe form of the condition, often have seizures, weak muscle tone (hypotonia), breathing problems, and delayed development. If left untreated, the disorder can lead to hearing loss, eye abnormalities and loss of vision, problems with movement and balance (ataxia), skin rashes, hair loss (alopecia), and a fungal infection called candidiasis. Immediate treatment and lifelong management with biotin supplements can prevent many of these complications.

Partial BIOT is a milder form of this condition. Affected children experience hypotonia, skin rashes, and hair loss, but these problems may appear only during illness, infection, or other times of stress.

## How do people inherit BIOT?

It is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## How common is BIOT?

Profound or partial BIOT occurs in approximately 1 in 60,000.

## Healthcare Professional Resources

1. ACT Sheets

[http://www.acmg.net/resources/policies/ACT/ACT\\_sheet-biotinidase\\_4-28-06\\_ljo.pdf](http://www.acmg.net/resources/policies/ACT/ACT_sheet-biotinidase_4-28-06_ljo.pdf)

2. Patient Resources

<http://ghr.nlm.nih.gov/condition=biotinidasedeficiency/show/Patient+support>

The information on BIOT is reproduced from NIH

<http://ghr.nlm.nih.gov/condition=biotinidasedeficiency>

## May 2009 Statistics

- 1 Case of MSUD
- 2 Cases of Citrullinemia

## IMPORTANT:

Many of you send payment along with screening samples for analysis. Please ensure that the cheque or DD is made out to, **NeoGen Labs Private Limited** payable at Bangalore.

As always, we look for your feedback to improve this newsletter.

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## First Step Newborn Screening Panel

### ACYLCARNITINE PROFILE (MS)

#### Fatty Acid Oxidation Disorders

- Carnitine / Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type I<sup>1</sup>
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
- 2,4-Dienoyl-CoA Reductase Deficiency<sup>1</sup>
- 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
- 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 (MS)

#### 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 disease
- Phenylketonuria
  - Classical / Hyperphenylalaninemia
  - Biotin Cofactor Deficiencies
- Tyrosinemia
  - Transient Neonatal Tyrosinemia
  - Tyrosinemia Type I<sup>1</sup>
  - Tyrosinemia Type II
  - Tyrosinemia Type III

### OTHER (MS)

- Hyeralimentation
- 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

### BIOCHEMICAL ANALYSIS (BIO)

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