



This is a combined issue for April and May 2009. I was travelling and unable to put out the April 2009 issue in a timely fashion. I hope all of you had a chance to review the March 2009 issue as it covered a lot of important information on the way we report.

Dr. Rohit Cariappa presented a seminar entitled "Newborn Screening for IEMs with Tandem Mass Spectrometry" at an IAP forum at Sir Ganga Ram Hospital, New Delhi on May 15, 2009.

As of May 1, 2009, we have started offering Biotinidase Deficiency screening. Our new panels (denoted by +) include Biotinidase.

In this issue we present information on Isovaleric Acidemia. We have seen three cases of this disorder in the samples we have processed at NeoGen.

Isovaleric Acidemia (IVA)

IVA is a rare disorder in which the body is unable to process a certain amino acid properly. It is classified as an organic acid disorder. People with IVA have inadequate levels of an inner mitochondrial matrix enzyme (Isovaleryl-CoA dehydrogenase) that helps break down the amino acid leucine. The finding of elevated five-carbon acylcarnitine (C5) by MS/MS indicates either IVA Deficiency or 2-MethylButyryl-CoA Dehydrogenase (2MGB) Deficiency.

Health problems related to IVA range from very mild to life-threatening. In severe cases, the features of IVA become apparent within a few days after birth. The initial symptoms include poor feeding, vomiting, seizures, and lethargy. These symptoms sometimes progress to more serious medical problems, including seizures, coma, and possibly death. A characteristic sign of IVA is a distinctive odor of sweaty feet during acute illness. It is caused by the buildup of isovaleric acid in affected individuals.

In other cases, the signs and symptoms of IVA appear during childhood and may come and go over time. Children with this condition may fail to gain weight and grow at the expected rate and often have delayed development. In these children, episodes of more serious health problems can be triggered by fasting, infections, or eating an increased amount of protein-rich foods.

Treatment of patients with IVA involves reducing protein intake. During an acute episode, aggressive use of glucose and electrolytes is necessary. Glycine supplementation has proven beneficial because this amino acid is conjugated to isovalerate, forming the less harmful isovaleryl-glycine. Carnitine supplementation is similarly effective.

How do people inherit IVA?

This condition 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 IVA?

In the US, it affects about 1 in 250,000. We do not have statistics for India.

Healthcare Professional Resources

1. ACT Sheets

http://www.acmg.net/resources/policies/ACT/ACT-sheet_C5_5-3-06.pdf

2. Patient Resources

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

The information on IVA is reproduced from NIH
<http://ghr.nlm.nih.gov/condition=isovalericacidemia>

March 2009 Statistics

- 1 Case of SCAD
- 1 Case of VLCADD
- 4 Cases of MMA/PA
- 1 Case of Homocystinuria
- 2 Cases of Tyrosinemia Type I

April 2009 Statistics

- 1 Case of MMA/PA
- 1 Case of MSUD

IMPORTANT: Administrative Notes

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.

Screening Panels

1. **First Step+** (Over 51 IEMs for Rs. 4200)
2. **First Step** (Over 50 IEMs for Rs. 3975)
3. **First Step MS/MS** (45 IEMs, includes Fatty Acid Oxidation Disorders, Amino Acid Disorders, and Organic Acid Disorder panels for Rs. 3250)
4. **First Step Bio+** (6 IEMs - CH, CAH, G6PD, GALT, Cystic Fibrosis, Biotinidase for Rs.1750).
5. **First Step Bio** (5 IEMs - CH, CAH, G6PD, GALT and Cystic Fibrosis for Rs.1500).

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

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