



January 2009 was a busy month for us. Dr. Cariappa was at PEDICON in Bangalore and met a few of you. IEM and newborn screening had more time devoted to it with a prime time session sponsored by PerkinElmer. The Delhi Government has installed a Tandem Mass Spectrometer at Lok Nayak Hospital.

## Diets

Diets play an important role in treating metabolic disorders. One of the challenges is that today none of these products are easily available in the market due to high customs duties. And, no screening program can be successful without access to diets for the affected individuals.

We have been working with Nutricia, a subsidiary of Danone in France, to get access to diets in India. They are in the process of registering their products in India so that they can be made easily available.

Today at NeoGen, we have a small quantity of diets for the following disorders (thanks to Nutricia's generosity);

- Maple Syrup Urine Disease
- Methylmalonic Acidemia
- PKU
- Tyrosinemia

These are limited quantities that will be available to parents who are committed to saving the infant's life. This stock will sustain the baby until the paperwork can be put together to import diets for ongoing treatment. We expect the diets to be replaced by the parents so that we can extend this benefit to others. We will also work with and guide parents through the paperwork, if requested.

Please contact **Chris Popma** at Nutricia Middle East & Africa directly if you have questions regarding diets.

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## Propionic Acidemia (PA)

PA is an inherited disorder in which the body is unable to process certain parts of proteins and lipids properly. It is classified as an organic acid disorder, a condition that leads to an abnormal buildup of organic acids. Abnormal levels of organic acids in the blood (organic acidemia), urine (organic aciduria), and tissues can be toxic and can cause serious health problems.

In most cases, the features of PA become apparent within a few days after birth. The initial symptoms include poor feeding, vomiting, appetite loss, weak muscle tone, and lack of energy. These symptoms sometimes progress to more serious medical

problems, including heart abnormalities, seizures, coma, and possibly death.

Less commonly, the signs and symptoms of PA appear during childhood and may come and go over time. Some affected children experience intellectual disability or delayed development. In children with this later-onset form of the condition, episodes of more serious health problems can be triggered by prolonged periods without food, fever, or infections.

## How common is PA?

We have seen **13** cases of **PA/MMA** in our screening in India. The condition appears to be common in several populations, including the Inuit population and Saudi Arabians.

## How do people inherit PA?

This condition is inherited in an autosomal recessive pattern. The parents of an individual with an autosomal recessive condition are carriers of one copy of the mutated gene but do not show signs and symptoms of the condition.

## Healthcare Professional Resources

1. ACT Sheets

<http://ghr.nlm.nih.gov/condition=propionicacidemia/show/ACTion+Sheets2>

2. Gene Reviews

<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=oa-overview>

The above information is reproduced from NIH  
<http://ghr.nlm.nih.gov/condition=propionicacidemia>

## January 2009 Statistics

- 3 Cases of Fatty Acid Oxidation Disorders

## Administrative Notes

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

## Screening Panels

- **First Step** (Over 50 IEMs for Rs. 3975)
- **First Step MS/MS** (45 IEMs, includes Fatty Acid Oxidation Disorders, Amino Acid Disorders, and Organic Acid Disorder panels for Rs. 3250)
- **First Step Bio** (5 IEMs which include 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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