



Every month we notice a small uptick in the number of samples we receive from all over India. We are also receiving a few samples from abroad. Slowly, but steadily, awareness about Newborn Screening and its benefits are increasing.

In this issue, we will present information on MADD, also known as GA-2. We had detected a case in Kochi, Kerala, in February 2008.

## Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or Glutaric Acidemia 2 or GA2)

MADD is an inherited disorder that interferes with the body's ability to break down proteins and fats to produce energy. Incompletely processed proteins and fats can build up in the body and cause the blood and tissues to become too acidic (metabolic acidosis).

MADD usually appears in infancy or early childhood as a sudden episode called a metabolic crisis, in which acidosis and low blood sugar cause weakness, behavior changes such as poor feeding and decreased activity, and vomiting. These metabolic crises, which can be life-threatening, may be triggered by common childhood illnesses or other stresses.

In the most severe cases of MADD, those affected may also be born with physical abnormalities. These may include brain malformations, an enlarged liver, a weakened and enlarged heart, fluid-filled cysts and other malformations of the kidneys, unusual facial features and genital abnormalities. It may also cause a characteristic odor resembling that of sweaty feet.

Some affected individuals have less severe symptoms that begin later in childhood or in adulthood. In the mildest forms, muscle weakness developing in adulthood may be the first sign of the disorder.

### How do people inherit MADD?

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 MADD?

It is a very rare disorder; its precise incidence is unknown.

### Healthcare Professional Resources

1. ACT Sheets

[http://www.acmg.net/resources/policies/ACT/ACT\\_sheet\\_C4;C5\\_5-3.pdf](http://www.acmg.net/resources/policies/ACT/ACT_sheet_C4;C5_5-3.pdf)

2. A Guide for Parents

[http://mchneighborhood.ichp.edu/pacnorgg/media/Metabolic/glut\\_acid\\_eng.pdf](http://mchneighborhood.ichp.edu/pacnorgg/media/Metabolic/glut_acid_eng.pdf)

The information on MADD is reproduced from NIH <http://ghr.nlm.nih.gov/condition=glutaricacidemia?type=i>

## Diet Update

**Nutricia**, the company that specializes in manufacturing diets for metabolic disorders, has provided NeoGen Labs with infant formula for treating a few select metabolic disorders. We are in the process of clearing it from customs but the law states that it has to be tested by CFTRI (Indian government lab) before it is cleared. This is a small stock for immediate intervention. Nutricia will have to be contacted directly to arrange for long term therapy.

## October 2008 Statistics

- 1 Case of G6PD
- 1 Case of PKU
- 1 Case of MSUD
- 1 Case of VLCADD
- 1 Case of MMA/PA

## Events

We were invited to speak at a CME organized by the IAP in Bhavnagar, Gujarat, on October 4, 2008.

Dr. Rohit Cariappa, Chief Scientist, will be attending the CME organized by Sunrise Hospital, Kakkanad, Kerala on November 30, 2008.

## Partners & Relationships

**Sunrise Hospital** in Kerala has started **routine screening** of newborns.

We have entered into a relationship with **SRL (Super Religare Laboratories)** to process their newborn screening samples. **SRL** is the leading pathology lab chain in India. This is in addition to our relationship with **Piramal Diagnostics**.

## Quick Tip

There are two copies of the sample information form. Please send the original to us and keep the second copy for your records. This allows you to track the sample that was sent to NeoGen. The **Filter Paper Number** is used track patient data at the lab. When calling about a sample, please refer to it.

## 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).

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