



We are witness to an increasing awareness of Newborn Screening (NBS) among the public in Bangalore. There is a steady increase in the number of samples for routine screening that we are receiving every month.

Goa's "Heel to Heal" NBS Program completed 3 months on September 13, 2008. We have some interesting findings to share. The cumulative worldwide incidence rate for the 45 IEMs in our MS/MS Panel is approximately 1 in 2300. In Goa, the incidence rate is about 1 in 400. The argument that IEMs are few or do not exist in India is not true. They do exist and the limited screening that has been done so far indicate that the incidence rates are high. As the Goa program continues, we will get statistically sound data. Disorders detected so far are Urea Cycle Disorders, MMA/PA, VLCADD, G6PD and CH.

## Symposium on Metabolic Disorders

We organized a symposium on September 8, 2008 at the Goa Medical College in Bambolim to increase awareness and educate health professionals in Goa in treating metabolic disorders. The focus was on practical information that could help in treatment once the diagnosis has been made. The three speakers were, Dr. Maria Silveira, GMC, Dr. Mamta Muranjan, KEM Hospital, Mumbai, and Dr. Rohit Cariappa, NeoGen Labs, Bangalore.

The symposium was sponsored by the Directorate of Health – Goa, Goa Medical College, LabIndia Bio Sciences and NeoGen Labs.

## Very LongChain Acyl-coenzyme A Dehydrogenase Deficiency (VLCADD)

**VLCADD** is a condition that prevents the body from converting certain fats to energy, particularly during periods without food (fasting). Normally, through a process called fatty acid oxidation, several enzymes work in a step-wise fashion to break down fats and convert them to energy. People with VLCADD have inadequate levels of an enzyme that metabolizes a group of fats called very long-chain fatty acids.

Typically, initial signs and symptoms of this disorder occur during infancy or childhood and include low blood sugar (hypoglycemia), lack of energy (lethargy), and muscle weakness. People with an early onset of VLCADD are also at risk of serious complications such as liver abnormalities and life-threatening heart problems. Symptoms that begin in adolescence or adulthood tend to be milder and usually do not involve heart problems. Episodes of VLCADD can be triggered by periods of fasting, illness, and exercise.

## How do people inherit VLCADD?

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 VLCADD?

VLCADD is estimated to affect 1 in 40,000 to 120,000 people.

## Healthcare Professional Resources

1. ACT Sheets

<http://ghr.nlm.nih.gov/condition=verylongchainacylcoenzymeadehydrogenasedeficiency/show/ACTion+Sheet>

The above information is reproduced from NIH

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

## August 2008 Statistics

- 1 case of MMA/PA
- 1 case of MSUD
- 1 case of VLCADD

## Couriers, Logistics & Cut Off Times

We are yet to find a courier that offers a consistent level of service and reliability all over India. Courier service between Bangalore and the 5 major metros is reasonably reliable, usually delivered within 2 days.

We prefer DTDC because they are headquartered in Bangalore and have significant operations here. Once a sample reaches DTDC in Bangalore, they deliver promptly. Other couriers who are reasonably reliable are PAFEX, First Flight and Blue Dart.

**If a sample reaches our laboratory before the cut off time of 1 PM, a result for the 45 disorders analyzed by Tandem Mass Spectrometry will be available by the next morning. If it is urgent, please let us know so that we can try to get a result by the end of the day.**

## Quick Tip

Please make a note of the **Filter Paper Number** before you send it to us. This is the number which we use to track all patient data. When calling us for reports, 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).

As always, we look forward to your feedback.

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