



Happy New Year! PEDICON 2009 is being held in Bangalore from 22nd to 25th January. We would like to extend an invitation to all of you at PEDICON to visit us in Bangalore. Please call Nihal George at 080 2580 5600, if you are planning to come by.

Dr. Cariappa will also be at PEDICON, attending those sessions focusing on Newborn Screening and Inborn Errors of Metabolism (IEMs). Please give him a call at 0 99006 55115 if you wish to meet him or would like to visit our lab.

We completed 6 months of newborn screening in Goa on December 14, 2008. We have collected a lot of data which we plan to present in the next few months.

Fatty Acid Oxidation (FAO) Disorders

These are recessively inherited errors of metabolism. As a group they represent the most common IEMs.

Q: *What are the common presentations of FAO disorders?*

A: Newborns with FAO disorders typically present with:

- Non-ketotic or hypoketotic hypoglycemia
- Metabolic acidosis
- Hepatic failure
- Cardiomyopathy

Late presentations include:

- Episodic Myopathy
- Neuropathy
- Retinopathy
- Arrhythmias

Sudden unexpected death can occur at any age and can be confused with sudden infant death syndrome (**SIDS**).

Some FAO disorders are associated with:

- Intrauterine growth restriction (**IUGR**)
- Prematurity
- Pregnancy complications in the heterozygous mother such as:
 - Severe preeclampsia
 - Acute fatty liver of pregnancy (**AFLP**)
 - Or, hemolysis, elevated liver enzymes, and low platelets (**HELLP**) syndrome.

Maternal pregnancy complications occur in mothers carrying a fetus with Long-Chain 3-hydroxy Acyl-CoA dehydrogenase (**LCHAD**) deficiency or general trifunctional protein (**TFP**) deficiencies.

The application of Tandem Mass Spectrometry (MS/MS) to newborn screening provides an effective means to identify most FAO disorder patients pre-symptomatically.

More information on these disorders can be found on our website. Please click on the following link to get to the page, <http://www.neogenlabs.com/iem.shtml>

Once you are there, click on the disorder of interest and you will get a wealth of information from descriptions to treatments.

Oct 2007 - Dec 2008 Statistics

Sample Size:	1396
High Risk:	686
Screening:	710

High Risk cases are those in which the physician suspects an IEM as a cause of the symptoms. Screening cases are those in which no IEM is suspected (newborns, mainly).

A total of 43 cases of IEMs were detected in this period. Of these, 2 cases (CH and G6PD Deficiency) were detected in the Screening population. The remaining 41 cases were detected from the High Risk population.

- 2 Cases of 3MCC/3MGA/HMG Lyase/BKT
- 2 Cases of CF
- 1 Case of CH
- 1 Case of Citrullinemia
- 2 Cases of G6PD Deficiency
- 1 Case of GA-I
- 1 Case of Galactosemia
- 1 Case of Homocystinuria
- 1 Case of LCHAD
- 1 Case of MADD (GA-II)
- 13 Cases of MMA/PA
- 10 Cases of MSUD
- 2 Case of PKU
- 1 Case of SBCAD
- 1 Case of TYR
- 2 Cases of UCD
- 1 Case of VLCAD

The IEM page referenced above will get you to worldwide incidence rates.

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 hearing from you. Your feedback helps us improve the newsletter.

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