



Last month we ran out of time and could not put out a newsletter. To make up for it, we have an issue with lots of statistics.

We completed two years of commercial operations in September 2009. In October 2007, our first full month of operations, we did barely 25 samples. It has increased significantly since then.

Conferences

ICBDD 2009 - The 4th International Conference on Birth Defects and Disabilities was held in New Delhi from October 4 to 7, 2009. Dr. Cariappa was a special invitee at a pre-conference workshop sponsored by ICMR, "The Science and Art of Newborn Screening", where he presented "NBS Experiences in Goa".

Dr. Cariappa also presented on the topic "Newborn Screening in Goa using TMS" at the conference. Additionally, a poster, "Goa – A Case Study of a Comprehensive Newborn Screening Program" was also displayed.

This conference was a valuable experience as we were able to meet and share experiences on all aspects of newborn screening with others from India and abroad. The pre-conference ICMR workshop held at AIIMS, Delhi, was especially enlightening since the focus was primarily on newborn screening initiatives in India.

ACBICON 2009 - From November 5 to 7, 2009, the annual conference of the Association of Clinical Biochemists of India will be held at AIMS in Kochi. Dr. Cariappa is an invited speaker and will be presenting "Routine Screening of Newborns for Metabolic Disorders using Tandem Mass Spectrometry". More information on the conference can be found at, www.aimshospital.org/acbicon2009/acbicon2009.html

Goa Newborn Screening Program



The program screens every infant born in a Goa Government healthcare facility. This is a screening program in the truest sense and unique in the number of disorders (50) screened for. The

data presented here may not be indicative of the general population in India. The incidence rate runs around 1 in 200, far higher than the expected rate of 1 in 1000 to 1500 in the world.

Important Note on Interpreting Data:

In the screening program, we count an occurrence of a disorder when we report it as a "Presumptive Positive". These disorders should go through a confirmatory test to confirm the existence of the disorder. For a variety of reasons, some of these,

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especially Fatty Acid Oxidation Disorders (FAOD), have not been through confirmatory testing. The NeoGen Screen, though not a confirmatory test, is very accurate since it is based on data from over 3.5 Million babies.

In the first year of the program, ending June 13, 2009, 8,247 infants were screened. Samples were collected from over 16 healthcare facilities that spanned the entire state of Goa. Within 6 months of the start of the program, over 99% of the births in the government healthcare facilities were covered.

1 Year Goa Statistics (53 Cases)

Of the 53 cases, 29 infants have survived and are being monitored, 21 infants have expired and the status of the remaining 3 is unknown.

- 3 Cases of Thyroid
- 9 Cases of G6PD Deficiency (1**)
- 1 Case of Glutaric Acidemia Type I*
- 4 Cases of MMA/PA*
- 3 Cases of Tyrosinemia/Liver Disease (1*)
- 1 Case of UCD*
- 32 Cases of Fatty Acid Oxidation Disorders
 - 7 Cases of MADD (GA II) (3* 1**)
 - 2 Cases of MADD/VLCADD (1* 1**)
 - 6 Cases of SCADD (2*)
 - 1 Case of SCADD/IBDH*
 - 2 Cases of SCADD/VLCADD (1*)
 - 13 Cases of VLCADD (6*)

* Baby Expired

** Status Unknown

Significantly, several of the FAOD patients (about 20) we have identified are from two families (based on surnames). We have been tracking these patients and have counseled them.

2 Year General Statistics

We look at our screening samples in discrete sets. The Goa data is looked at separately and is presented above. The remaining samples from the rest of India are classified as "High Risk", where the child is suspected of a disorder, and "Routine Screening", where the child is healthy at birth. These following disorders were reported as 'Presumptive Positive'. We have recommended confirmatory testing but do not know if this has been carried out in all cases.

'High Risk' Screening Sample Size: 1,533, Disorders: 64

- 2 Cases of 3MCC/3MGA/HMG Lyase/BKT
- 1 Case of Biotinidase Deficiency
- 1 Case of CAH
- 2 Cases of CF
- 1 Case of CH
- 2 Cases of Citrullinemia
- 1 Case of G6PD Deficiency
- 1 Case of Glutaric Acidemia Type I
- 1 Case of MADD (Glutaric Acidemia Type II)

- 1 Case of Galactosemia
- 6 Cases of Homocystinuria/Liver Disease
- 3 Cases of IVA/2MBG
- 1 Case of LCHAD
- 19 Cases of MMA/PA
- 13 Cases of MSUD
- 3 Cases of PKU
- 3 Cases of Tyrosinemia/Liver Disease
- 2 Cases of UCD (Urea Cycle Disorders)
- 1 Case of VLCADD

Routine Screening Sample Size: 3,803, Disorders: 8

- 1 Case of 3MCC/3MGA/HMG Lyase/BKT
- 1 Case of CAH
- 1 Case of Citrullinemia
- 1 Case of CPT-II/CACT
- 1 Case of G6PD Deficiency
- 2 Cases of Homocystinuria/Liver Disease
- 1 Case of SCHAD

We report multiple disorders (eg.: IVA/2MBG) together. The reason is that the analytes are similar for these disorders and only the confirmatory test will narrow it down.

Screening Panels

MS - Mass Spectrometry Panel (45+ Disorders)

(45 IEMs, includes Fatty Acid Oxidation Disorders, Amino Acid Disorders, and Organic Acid Disorder panels)

Bio - Biochemical Panel (6 Disorders)

CH, CAH, G6PD, GALT, CF, BIOT

1. **First Step PLUS** (Over 51 Disorders)
 - MS + Bio (Rs. 4,250/-)
 - Infants, 0 to 2 months old
 - Biotinidase is valid only until 28 days after birth
2. **First Step** (Over 49 Disorders)
 - MS + CH + CAH + GALT + G6PD (Rs. 3,975/-)
 - Infants, Over 2 months old

3. **First Step MS** (Over 45 Disorders)

- MS (Rs. 3,250/-)
- No Age limit

4. **First Step Bio6** (6 Disorders)

- CH + CAH + GALT + G6PD + CF + BIOT (Rs. 2,000/-)
- Infants, 0 to 2 months old
- Biotinidase is valid only until 28 days after birth

5. **First Step Bio4** (4 Disorders)

- CH + CAH + GALT + G6PD (Rs. 1,500/-)
- No Age limit

Please call or e-mail us if you need to do individual tests or combinations that are not listed above.

IMPORTANT:

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.

Logistics, Kits, Reports

Please send e-mail to info@neogenlabs.com in case you need kits, looking for reports or any general question. You can expect a reply or call within 24 hours (except for holidays) to address your request.

Service Feedback & Complaints

If you feel that we have not lived up to your expectations, please call or e-mail me. We take your concerns very seriously and will address them immediately.

Thomas Mookken

T: +91 99006 55115

E: mookken@neogenlabs.com

www.neogenlabs.com