



In this newsletter we discuss other benefits of performing a Newborn Screen. The obvious benefit is that if a disorder is detected early on in life and treated, there is a very good possibility that the child will lead a normal healthy life. The following case synopsis discusses other benefits which are not as obvious but just as powerful.

1. An explanation for Unexplained Deaths

A baby died without the cause of death identified. Fortunately, the doctor had a suspicion that it may have been afflicted by an IEM and had collected a sample a day before the baby expired. The sample was analyzed in our lab and we were able to accurately identify that **Citrullinemia** was the cause. Even though the outcome was tragic, there was a silver lining. The parents were counseled about the implications of having a second child and the consequences. If they choose to have another child, there is a 25% probability that the child would be affected.

2. Speedy Diagnosis

A baby was in the NICU for nearly 2 weeks without a diagnosis. One of the doctors suggested a NBS Test. A blood sample was collected and within 6 hours of the sample reaching our lab, we were able to confirm that the baby had **Maple Syrup Urine Disease (MSUD)**. If the NBS Test was done the very first day the baby entered the NICU, the diagnosis would have been faster and NICU costs would have been much lower. If the NBS Test is positive, the metabolic disorder is identified. If it is negative, it rules out over 50 metabolic disorders. A negative result helps in the differential diagnosis as bacterial infections, sepsis and IEMs have several overlapping symptoms.

3. Effectiveness of Treatment

A child was diagnosed with **Methylmalonic Acidemia (MMA)**. The doctor wanted to find out the effectiveness of the treatment and requested a 'follow up' NBS Test. The initial screen showed a high level of metabolites that were associated with MMA. The second test, done after a few days of treatment, indicated normal levels of metabolites, supporting the effectiveness of the treatment.

April 2008 Statistics

- 1 Case of **Propionic Acidemia** (1:100,000)
- 1 Case of **MSUD** (1:185,000)
- 1 Case of elevated **C5OH**

Maple Syrup Urine Disease (MSUD)

MSUD is a disorder which we are encounter quite frequently in India. The worldwide incidence rate is very rare (1:185,000) but we are seeing evidence that

it is much higher in India. We have detected 4 positive cases in 8 months.

Description

MSUD is an inherited disorder in which the body is unable to process certain protein building blocks (branched chain amino acids) properly. Beginning in early infancy, this condition is characterized by poor feeding, vomiting, lack of energy (lethargy), seizures, and developmental delay. The urine of affected infants has a distinctive sweet odor, much like burned caramel that gives the condition its name. Maple syrup urine disease can be life-threatening if untreated.

MSUD can be classified by its pattern of signs and symptoms or by its genetic cause. The most common and most severe form of the disease is the classic type, which appears soon after birth. Variant forms of the disorder appear later in infancy or childhood and are typically milder, but still involve mental and physical retardation if not treated.

How do people inherit MSUD?

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.

Healthcare Professional Resources

1. ACT Sheets

<http://ghr.nlm.nih.gov/condition=maplesyrupurinedisease/show/ACTION+Sheets>

2. Gene Reviews

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

The above information is reproduced from NIH

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

Screening Panels

We offer 3 panels to reflect demand:

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.1250).

As always, we look for your feedback on how this newsletter can be improved. Let us know specific topics you would like us to cover.

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