

The last issue presented a lot of statistics and we hope they did not overwhelm you. While the sample size was limited, it is a start. We hope that as time goes by and screening continues, we will start getting statistically significant data.

We are continuing to investigate the reason for the high number of Fatty Acid Oxidation Disorders (FAOD) in Goa. Preliminary investigations indicate that they belong to families with 2 or 3 common last names.

Research Partnerships

If you are associated with an organization that does research in subjects related to IEMs, please let us know. We may be able to help with discounted testing prices and data.

Galactosemia (GALT/GALK/GALE)

Galactosemia is a disorder that affects how the body processes a simple sugar called galactose. A small amount of galactose is present in many foods. It is primarily part of a larger sugar called lactose, which is found in all dairy products and many baby formulas. The signs and symptoms of Galactosemia result from an inability to use galactose to produce energy.

Researchers have identified several types of Galactosemia. These conditions are each caused by mutations in a particular gene, and affect different enzymes involved in breaking down galactose. Classic Galactosemia, GALT (also called GALT Type I or galactose-1-phosphate uridylyl transferase deficiency), is the most common and most severe form of the condition and accounts for about 95% of galactosemias. GALK (also called galactokinase deficiency or GALT Type II) and GALE (also called galactose epimerase deficiency or GALT Type III) cause different patterns of signs and symptoms but account for only about 5% of Galactosemia cases.

If infants with classic GALT are not treated promptly with a low-galactose diet, serious complications appear within a few days after birth. Affected infants typically develop feeding difficulties, a lack of energy (lethargy), a failure to gain weight and grow as expected (failure to thrive), yellowing of the skin and whites of the eyes (jaundice), liver damage, and bleeding. Other serious complications of this condition can include overwhelming bacterial infections (sepsis) and shock. Affected children are also at increased risk of delayed development, clouding of the lens of the eye (cataract), speech difficulties, and intellectual disability. Females with classic GALT may experience reproductive problems caused by ovarian failure.

GALT Type II causes fewer medical problems than the classic type. Affected infants develop cataracts,

but may not experience long-term complications. The signs and symptoms of GALT Type III vary from mild to severe and can include cataracts, delayed growth and development, intellectual disability, liver disease, and kidney problems.

How do people inherit GALT?

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 GALT?

Classic GALT occurs in 1 in 30,000 to 60,000 newborns. GALT Type II and III are less common; Type II probably affects fewer than 1 in 100,000 newborns and Type III appears to be very rare.

Resources

1. ACT Sheets

http://www.acmg.net/resources/policies/ACT/Act-Sheet-Galactose+GALT_4-28-06_ljo.pdf

http://www.acmg.net/resources/policies/ACT/Act-Sheet-Galactose_4-27-06_ljo.pdf

2. Patient Resources

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

The information on GALT is reproduced from NIH

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

Oct 2009 Statistics

- 1 Case of G6PD Deficiency
- 1 Case of SCAD
- 1 Case of VLCADD
- 1 Case of Thyroid
- 1 Case of MMA/PA
- 1 Case of Homocystinuria/Liver Disease

Treatment Monitoring

Once a metabolic disorder is identified, it is critical that the patient is monitored while undergoing treatment. The periodic monitoring could be expensive and patients may discount the importance of it. In our effort to ensure positive outcomes, NeoGen Lab will extend special pricing.

IMPORTANT:

Please ensure that the cheque or DD is made out to, **NeoGen Labs Private Limited** payable at Bangalore.

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First Step Newborn Screening Panel

ACYLCARNITINE PROFILE (MS)

Fatty Acid Oxidation Disorders

- Carnitine / Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type I
- 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
- 2,4-Dienoyl-CoA Reductase Deficiency¹
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Multiple Acyl-CoA Dehydrogenase Deficiency
- Neonatal Carnitine Palmitoyl Transferase Deficiency Type II
- Short-chain Acyl-CoA Dehydrogenase Deficiency
- Short chain Hydroxy Acyl-CoA Dehydrogenase Deficiency
- Trifunctional Protein Deficiency
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency

Organic Acid Disorders

- 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
- Glutaric Acidemia Type I
- Isobutyryl-CoA Dehydrogenase Deficiency
- Isovaleric Acidemia
- 2-Methylbutyryl-CoA Dehydrogenase Deficiency
- 3-Methylcrotonyl-CoA Carboxylase Deficiency
- 3-Methylglutaconyl-CoA Hydratase Deficiency
- Methylmalonic Acidemias
- Methylmalonic Acidemias
 - Methylmalonyl-CoA Mutase Deficiency
 - Some Adenosylcobalamin Synthesis Defects
 - Maternal Vitamin B12 Deficiency
- Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
- Propionic Acidemia
- Multiple CoA Carboxylase Deficiency
- Malonic Aciduria

AMINO ACID PROFILE (MS)

Amino Acid Disorders

- Argininemia
- Argininosuccinic Aciduria
- 5-Oxoprolinuria
- Carbamoylphosphate Synthetase Deficiency
- Citrullinemia
- Homocystinuria
- Hypermethioninemia
- Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome
- Hyperornithinemia with Gyral Atrophy
- Maple syrup disease
- Phenylketonuria
 - Classical/Hyperphenylalaninemia
 - Biotin Cofactor Deficiencies
- Tyrosinemia
 - Transient Neonatal Tyrosinemia
 - Tyrosinemia Type I
 - Tyrosinemia Type II
 - Tyrosinemia Type III

OTHER (MS)

- Hyeralimentation
- Liver Disease
- Medium Chain Triglyceride Oil Administration
- Presence of EDTA Anticoagulants in blood specimen
- Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
- Carnitine Uptake Deficiency

BIOCHEMICAL ANALYSIS (BIO)

- Galactosemia
- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- G6PD
- Cystic Fibrosis
- Biotinidase