



The 4th International Conference on Birth Defects and Disabilities in the Developing World is being held in New Delhi. The conference is sponsored by the CDC and the March of Dimes. Dr. Rohit Cariappa, Chief Scientist at NeoGen Labs, will be speaking at the conference. Additionally, we will have a poster presentation.

The conference is being held from Oct 4 to 7, 2009 at the Le Meridien Hotel, New Delhi. Visit the web, <http://www.4icbddw2009.com/> for more details.

In early July, Dr. Cariappa made a presentation to the Goa Health Advisory Committee, summarizing 1 year of data for the Newborn Screening Program in Goa.

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 in these cases.

Congenital Adrenal Hyperplasia (CAH)

CAH (also known as 21-hydroxylase deficiency) is an inherited disorder that affects the adrenal glands. These glands are located on top of the kidneys and produce a variety of hormones that regulate many essential functions in the body. Two of these hormones, cortisol and aldosterone, are produced from cholesterol through the activity of an enzyme called 21-hydroxylase. Cortisol has numerous functions such as maintaining blood sugar levels, protecting the body from stress, and suppressing inflammation. Aldosterone, sometimes called the salt-retaining hormone, acts on the kidneys to regulate the levels of salt and water in the body, which affects blood pressure. People with CAH have a shortage of the 21-hydroxylase enzyme, which impairs the conversion of cholesterol to cortisol and aldosterone. When the precursors of cortisol and aldosterone build up in the adrenal glands, they are converted to male sex hormones called androgens. Androgens are normally responsible for the appearance of secondary sex characteristics in males (virilization). Elevated levels of androgens can affect the growth and development of both males and females.

There are three types of CAH. Two types are classic forms, known as the simple virilizing and salt-loss types. Simple virilizing CAH causes a buildup of potent androgens that leads to the masculinization (development of male characteristics) of external genitalia in females at birth. The development of the internal reproductive organs (uterus and ovaries) in these patients is normal. Salt-loss CAH results from an almost complete loss of enzyme activity. In these

cases, so little aldosterone is produced that the kidneys do not reabsorb sodium (a component of salt). In the third type of CAH, known as the nonclassic form, levels of functional 21-hydroxylase enzyme are moderate. Both males and females with the nonclassic type can display signs and symptoms of androgen excess after birth.

How do people inherit CAH?

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 CAH?

The classic form of CAH appears in 1 in 15,000 newborns. The prevalence of the non classic form of CAH is estimated to be 1 in 100 individuals. The prevalence of both forms may vary among different ethnic populations.

Resources

1. ACT Sheets
http://www.acmg.net/resources/policies/ACT/Act_sheet_CAH_4-28-06_ljo.pdf
2. Patient Resources
<http://ghr.nlm.nih.gov/condition=21hydroxylasedeficiency/show/Patient+support>

The information on CAH is reproduced from NIH
<http://ghr.nlm.nih.gov/condition=21hydroxylasedeficiency>

June 2009 Statistics

- 1 Case of Homocystinuria/Liver disease
- 1 Case of MADD

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.

As always, we look for your feedback to improve this newsletter.

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.

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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