What are metabolic disorders?

Metabolism is the process our body uses to make energy from the food that we eat. Food is made up of carbohydrates, fatty acids, and amino acids. Chemicals in our body break them down and convert them into other substances through various metabolic pathways. All these metabolic pathways are controlled by enzymes which help convert one metabolite to another.

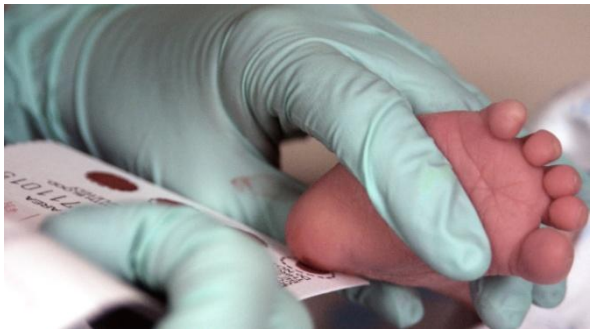
A metabolic disorder occurs when a genetic defect results in the production of a defective enzyme. The defective enzyme does not convert one metabolite to another as expected. When this happens, one may have too much of the first metabolite or too little of the other. In addition the excess metabolites will be converted to other unwanted even toxic metabolites. This leads to an imbalance in various chemical substances in our body resulting in what is referred to as a metabolic disorder.

Why should a baby be screened for metabolic disorders?

1 in 1000 babies has metabolic disorders. These babies are normal at birth. When symptoms and signs appear days or weeks after birth, damage may already have been done to the nervous system, kidneys, eyes, ears, or other organ systems. That is why early detection of these serious disorders in all infants is very important. Early diagnosis and treatment of these disorders can reduce the risk of disease, disability, and even death.

How will my baby be tested?

Your baby's heel will be pricked to obtain a few drops of blood for testing.



It is generally recommended that the sample be taken after the first 24 hours of life have passed. By then the baby would have taken a few feeds and all his metabolic pathways would have begun to function.

What if my baby's test comes as abnormal?

This test is a screening test. Therefore, if a test result comes back as abnormal, there is no need to worry. Other tests are needed to confirm the diagnosis.

A screening test is not the same as diagnostic test. The initial screening provides only preliminary information that must be followed up with more specific diagnostic testing. Sometimes screening may indicate a problem, but after further investigation no problem may be found. However, if a diagnosis is confirmed by testing, your baby will need treatment.

If my baby has one of these disorders, can it be cured?

At the present point in time most metabolic disorders cannot be cured. The aim of early diagnosis and treatment is to prevent more harm being done to the baby due to deficiency of metabolites, excess of metabolites or production of toxic metabolites. The treatment includes medication, special diets and /or other treatments.

How does a baby get a metabolic disorder?

We all have two copies of every gene – one comes from the father, the other from the mother. Metabolic disorders are caused when a gene responsible for producing a particular enzyme transmitted by both the father and the mother have mutations. Such parents are usually normal. Of the two copies of the gene that they have, one is normal and the other is mutated. They are therefore called carriers.

Will my other children have the disorder?

The answer to this question will depend on your child's diagnosis. A doctor can study your family's health history and explain the chance of this happening again. This process is called genetic counseling.

What disorders can you screen for?

We offer screening for the following disorders:

- Congenital hypothyroidism
- Congenital adrenal Hyperplasia
- Galactosemia
- G6PD Deficiency
- Cystic Fibrosis
- Biotinidase
- 11 Fatty Acid Oxidation Disorders
- 14 Amino Acid Disorders
- 12 Organic Acid Disorders

(Please see the separate leaflet with test details and costs)

Human Genetics Unit
Faculty of Medicine
University of Colombo
Kynsey Road, Colombo 8
Phone: 011 2689545
Fax: 011 2689979
E-mail: office@hgucolombo.org
Website: www.hgucolombo.org

This service is provided by the HGU in collaboration with NeoGen Labs, India.

Is my baby healthy?

Metabolic Screening May Provide the Answer

FACTS YOU SHOULD KNOW