



A Simple Test That Will Save Your Baby's Life!!!

A Parent's Guide to Newborn Screening

WHAT IS METABOLIC DISORDER?

Metabolic disorders or more specifically "Inborn Errors of Metabolism" are genetically inherited disorders caused by the decrease in the enzyme catalytic activity. Enzymes are proteins essential for the body's metabolism in ensuring the well being of a person. As a result of the defective gene, production of enzyme in the body is altered leading to failure of the metabolic processes. This will lead to the accumulation of substances or metabolites that are toxic to the body. This condition, if left untreated, might affect the mental and physical development of the baby. Some metabolic disorders can be life threatening.

WHAT IS NEWBORN SCREENING (NBS)?

NBS is a test to help your doctor diagnose some of the metabolic disorders your baby might have without the presenting clinical symptoms. With early detection followed by prompt treatment, conditions like mental retardation, severe disability or even death can be avoided. This test has proven to be reliable and highly beneficial in most countries that have implemented it. A number of developed countries have legislation making the test compulsory for all newborns due to its effectiveness as preventive medicine. It has been reported that for these metabolic disorders (consisting of 25 disorders), the incidence is 1:3,500 live births.

HOW IS THE TEST PERFORMED?

The test is performed when your baby is 24-72 hours old and has had proper milk feeding. A few drops of blood, obtained from a gentle prick on the heel of your baby are dropped on a special filter paper. The dried blood on the filter paper is then sent to the laboratory for testing using highly sophisticated instrument known as the tandem mass spectrometer which is only available in specialized laboratory.

HOW AND WHEN WILL I KNOW THE RESULTS OF MY BABY'S TEST?

We will dispatch the results to the attending doctor three (3) days after the receipt of the sample. If your child's test shows an abnormal result, your doctor will be notified immediately for the subsequent follow-up test. Follow the directions of your doctor very carefully as a retest or additional tests are usually necessary to verify if your child has the disorder.

IS TREATMENT AVAILABLE?

The answer is yes for most of the disorders that can be detected by NBS. Your doctor will refer your baby to a medical specialist who will advise you on the management and treatment of your baby.



For further information

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