Metabolic Disorders (Basic)

Phenylketonuria is also called PKU and results when a part of a food protein (phenylalanine) is not broken down by the baby's body. Brain damage that would normally result can be prevented by a special diet low in phenylalanine.

Hypothyroidism results the baby's body does not produce enough of a hormone (substance) called thyroxine. Treatment with thyroxine tablets helps prevent mental and growth retardation.

Congenital Adrenal Hyperplasia (CAH) results when the baby's body does not produce enough of a substance (hormone) called cortisol. Treatment with hormone medications can prevent low blood sugar, salt loss, poor growth and abnormal body changes.

Galactosemia results when milk sugar (galactose) is not broken down due to the lack of a chemical in the body. A diet low in galactose can prevent irreversible brain damage and other complications.

Biotinidase deficiency - A common disorder of an essential water soluble vitamin Biotin. Treatment with biotin will be life saving.

For Further Information :
Please contact your Doctor / Nurse
or Pathology Department
Tel : 2366 7822 / 2366 7825
(9.00 am to 5.00 pm)

New Born Screening For Metabolic Disorders

BREACH CANDY
HOSPITAL TRUST

Give your baby the best start in life

Your care is our Concern

New Born Screening For Metabolic Disorders

BREACH CANDY
HOSPITAL TRUST

Your care is our Concern

Newborn Screening For Metabolic Disorders

We have received information regarding the newborn screening test and would like to get the screening done prior to discharge from the hospital.

☐ Newborn Screening (Basic)

☐ Newborn Screening - Basic + Extended

Yes

No

Signature

Parent's Name

Breach Candy Hospital Trust
Bhulabhai Desai Road,
Mumbai - 400 026.

New Born Screening For Metabolic Disorders

A New Screening Test, for Inborn Errors of Metabolism(IEM) can detect over 50 treatable metabolic disorders in babies. These are disorders that prevent the baby from 'processing' food properly, leading to an accumulation of toxic chemicals in the blood that are harmful to the baby's normal development. Most babies with these disorders look and act normally and seem perfectly healthy. If these remain undetected and untreated at birth, the results could be devastating, leading to mental retardation or even death depending on the disorder.

A Newborn Screening Test can detect abnormalities in the blood **BEFORE** symptoms appear. Treatment in most cases result in a normal healthy individual. However, once symptoms appear in the baby, the damage has already been done and it may be too late for treatment to have a good outcome. Thus it is vital to screen **ALL** babies at birth for these disorders.

In most cases when a disorder is detected, the treatment involves a dietary change or an addition of a vitamin for normal development. In India, due to lack of awareness of the benefits and high costs, only a few babies from families with the history of metabolic disorders are screened. This is woefully inadequate because most often these disorders appear in babies with no family history of the disorder.

How is Newborn Screening Done?

Newborn Screening is performed by pricking the baby's heel and putting a few drops of blood on a special filter paper. The paper is allowed to dry and then sent to the lab where the tests are performed. This test is usually performed when the baby is 24 hours to 72 hours old.

But we have no family history of these disorders...

Parents who have no family history of problems and who have already had healthy children can still have children with these disorders. In fact, most children with these disorders come from families with no previous history of the disorder.

What does a positive result indicate?

Parents should not be alarmed by positive result as the Screening gives preliminary information, albeit with a high degree of accuracy. It should be followed by a precise confirmatory test.

What exactly are Inborn Errors of Metabolism (also called Metabolic Disorders) ?

These are disorders caused by the accumulation of chemicals produced naturally in the body to abnormal levels. The symptoms manifest themselves in a variety of ways: impaired physical development or mental retardation. In some cases, they could result in death. Unfortunately, infants with these disorders show no early signs of these disorders and by the time symptoms appear it is too late for treatment.