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INBORN ERRORS OF METABOLISM (METABOLIC SCREENING FOR YOUR NEWBORN BABY)

The inborn errors of metabolism are a large group of *genetic diseases* involving problems with enzyme activities. The majority are due to defects of single genes coding for enzymes that help to convert various substrates (such as organic acids, amino acids, fatty acids) into products for the normal functioning of the body. In many disorders, problems develop when toxic substances accumulate or interfere with normal function, or when there is reduced ability to manufacture essential compounds. Hence inborn errors of metabolism are often referred to as *inherited metabolic disorders.*

Early Detection Allows Early Treatment

Although individually rare, these inherited metabolic disorders collectively affect about one in 3000 births in Singapore. By screening newborn babies during their first few days of life, such disorders can be treated early, often before any sign or symptom appears.

Which Inherited Metabolic Disorders Are Screened For?

The newborn screening test looks for more than 40 metabolic related disorders in these categories:

- Organic Acid Disorders
- Fatty Acid Oxidation Disorders
- Amino Acid Disorders
- Cystic Fibrosis (this is a genetic condition that leads to accumulation of thick mucus in different organs leading to severe chest infections and poor growth)
- Congenital Adrenal Hyperplasia (this is a hormone disorder that affects metabolism, response to infection, ability to regulate salt levels and gender characteristics)
- Severe Combined Immunodeficiency (this results in extremely poor immunity which places the baby at risk for severe infections)
- Galactosemia (this condition causes the baby to be unable to digest lactose-containing milk feeds and can lead to liver failure)
- Biotinidase Deficiency (this condition causes the baby to be unable to recycle biotin which can lead to seizures, developmental delay, low muscle tone, skin and hair abnormalities)

How Is Newborn Screening For Metabolic Disorders Done?

In order to perform the screening tests, only a small blood sample from your baby is required. A few drops of blood are collected on a special filter paper by gently pricking the heel. The sample is sent to the National Expanded Newborn Screening Laboratory for analysis. Blood samples can be taken from your baby any time between one day (24 hours) and seven days old. The best time for collection is between one to three days old (24 to 72 hours).

How Are The Results For Metabolic Screening Reported?

The results of metabolic screening may be returned in one of three ways:

- *YOUR BABY'S RESULT IS NEGATIVE FOR ALL THE DISORDERS.* More than 99% of the babies fall into this category. It means that your baby is at very low risk of having these disorders. The result will be noted by the doctors and filed into your baby's medical record. On very rare occasions, a disorder may not be picked up on the metabolic screen.
- *A REPEAT SAMPLE IS REQUIRED*. A second sample is needed for about one in 120 babies. It is usually needed because the first sample showed borderline results, which means it was not possible to tell whether or not a problem exists. You will be contacted to arrange for another sample to be taken.
- *YOUR BABY'S RESULT IS POSITIVE FOR ONE OF THE CONDITIONS*. Having a positive result does not necessarily mean that your baby has a disorder. It only means that further testing is required. You will be contacted to make arrangements for a metabolic paediatrician to review and investigate. If a diagnosis is made, your baby will be given appropriate treatment right away. It is important to know that most babies in Singapore are screened negative. These rare but lifethreatening disorders can usually be prevented with early diagnosis and treatment.

Get In Touch

For more information about the metabolic screen in Singapore, speak to your baby doctor at SGH. Remember that early detection can allow early treatment to be given for your baby.

Alternatively, you can contact the National Expanded Newborn Screening Laboratory (telephone: 6394-5049) or write to <Metabolic.Newborn.Screening@kkh.com.sg>