High Risk or Low Risk Results

When the sample has been analysed and results are available, one of the following will occur:

**Your baby’s result is Negative for all the disorders.**

More than 99 per cent 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 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 1000 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 needed. You will be contacted to make arrangements for a metabolic physician 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 life-threatening disorders can usually be prevented with early diagnosis and treatment.

You can also write to us at Metabolic.Newborn.Screening@kkh.com.sg

If you are delivering your baby at KK Women’s and Children’s Hospital (KKH) or Singapore General Hospital (SGH), your baby will be screened. Please note that you have the right to refuse the metabolic screen. If this is your wish, please speak to your healthcare provider.

If you are delivering in a hospital, other than KKH or SGH, you may enquire with your healthcare provider for the test.

Get in Touch

For more information about the metabolic screen in Singapore, speak to your healthcare provider, or contact the National Expanded Newborn Screening Laboratory

Tel: 6394-5049

Useful telephone number

Central Appointments 6294-4050

KK Women’s and Children’s Hospital

100 Bukit Timah Road

Singapore 229899

Tel: 6394-5049

Fax: 6394-3773

Website: www.kkh.com.sg

www.facebook.com/kkh.sg
A New Beginning of Life – Congratulations to the arrival of your new bundle of joy. As a parent, your baby's health is your first priority. To ensure that your baby gets the best start in life and stays healthy, your newborn can be screened for more than 40 rare medical disorders.

Although most newborns with these disorders look healthy at birth, they may be at risk of having serious health problems later in life. These may include learning difficulties, recurrent sickness and even death if their disorder is not detected and treated early.

Early Detection allows Early Treatment
These disorders are very rare and they affect about one in 3000 births in Singapore. By testing newborns during their first few days of life, such disorders can be treated early, often before any sign or symptom surfaces.

This pamphlet will provide you with information about newborn screening in Singapore, its benefits to your newborn, and answer some questions you may have in mind.

The disorders in the newborn screening include:

- **Inborn Errors of Metabolism**
  When the body cannot successfully breakdown (metabolise) some substances in food/milk such as fats, protein or sugar, they can accumulate and become toxic to the body. This can cause serious health problems.

The newborn screening looks for more than 30 metabolic related disorders in the following 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 disorder of hormone production that may affect baby's metabolism, response to infection, ability to regulate salt levels and sex characteristics.

- **Severe Combined Immune Deficiency (SCID)**
  This condition results in extremely poor immunity and the child can develop severe infections.

- **Galactosemia**
  Babies with galactosemia are unable to process lactose containing milk feeds and ingestion may lead to liver failure.

- **Biotinidase Deficiency**
  This is a metabolic disorder whereby the body is unable to recycle biotin leading to a deficiency. This can result in complications such as seizures, developmental delay and low muscle tone. Babies can also have skin and hair abnormalities.

It is important to note that although these are all serious medical conditions, early diagnosis and treatment can lead to a good outcome.

A Small Test with Big Benefits
In order to perform the screening tests, a small blood sample is needed from your baby. A few drops of blood are collected on 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).