PathCare
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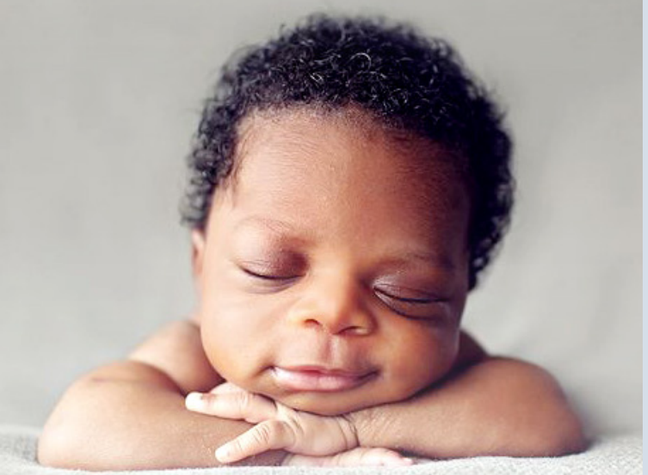

Your newborn baby's screening test

A blood test to screen for genetic- and metabolic conditions

Answers to Frequently Asked Questions

What is Newborn Screening?

Newborn screening is a process where infants are screened for specific conditions shortly after birth. These conditions may not always be clinically evident in the newborn period. Newborn screening entails different kinds of tests including hearing tests, cardiology examinations and blood tests. The laboratory Newborn Screening test aims to detect certain rare, but serious, metabolic and endocrine conditions in the newborn. Early detection of these conditions before the baby becomes symptomatic can reduce the risk of severe illness, disability and even death. It also allows for earlier monitoring and treatment, resulting in better health outcomes for most diagnosed babies.

Who should be tested?

Although the disorders screened for are often the result of inherited gene mutations, most affected babies identified through Newborn Screening are from families with no history of the disorder. Since we do not know which child may be at risk of a metabolic disease, international medical practice recommends that all children undergo Newborn Screening.

What is the Newborn Screening status in South Africa?

Although Newborn Screening is accepted worldwide as a routine public health measure aimed at early screening, diagnosis and management of selected treatable inherited diseases, this is not yet the case in South Africa. A Newborn Screening test profile relevant to the South African population is available through the private health system on request of the parents.

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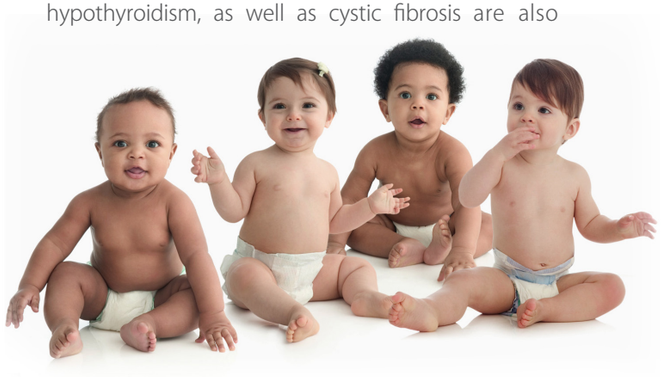
When should my newborn be screened?

For term babies the ideal time is between 24 – 72 hours after the first feed, but up to one week of age is still acceptable. The reason for the delay is that the baby's metabolism needs to break down the proteins, carbohydrates and fat present in breast or formula milk, before some of the metabolic disorders can be detected. Different screening times may apply for premature and sick neonates.

What conditions are screened for?

The newborn screening test screens for a group of disorders called Inborn Errors of Metabolism (IEM), some endocrine disorders and Cystic Fibrosis. IEM refers to metabolic disorders that results from the absence or abnormality of an enzyme or its co-factor, leading to either the accumulation of toxic breakdown products or the deficiency of a specific nutrient required for normal body function. These include amino acid, carbohydrate, organic acid and fatty acid oxidation pathway disorders. Endocrine disorders such as congenital adrenal hyperplasia and congenital hypothyroidism, as well as cystic fibrosis are also included in the screening. More information on the specific disorders tested may be obtained from your local PathCare laboratory.



How common are these disorders?

These disorders are individually rare, but cumulatively they affect approximately one out of every 2000 newborns in South Africa.

What if a Newborn Screening test is abnormal?

As this is only a screening test, any abnormal tests will be followed up with confirmatory tests to exclude or confirm any condition. In the case of an abnormal test result, the pediatrician will discuss the result, and the need for further testing with the parents.

What happens if an infant is diagnosed with a disorder?

If the follow-up tests come back positive for any of the disorders screened, the laboratory will immediately contact the pediatrician, who will inform the parents, and discuss a treatment plan. Genetic counselling is strongly advised as most of these disorders are hereditary and have a possible recurrence risk in the family. Contact the Pathcare Genetics team on geneticconsult@pathcare.org or **021 596 3655** for the details of a Genetic Counsellor in your area.

How are these disorders treated?

In most instances, treatment consists of dietary modifications, dietary supplementation, hormones and sometimes medication. If an infant has one of these disorders, it is very important to start treatment as soon as possible.

How is the screening performed?

The test can be requested by your pediatrician through most private laboratories in South Africa. You can discuss the test with your gynaecologist during pregnancy or pediatrician soon after delivery. A few drops of blood are collected from a heel prick onto a special absorbent paper called a Guthrie card. The dried blood card will be sent to the North-West University Newborn screening laboratory for testing. Any positive result will immediately be reported to the pediatrician.

In conclusion: Newborn Screening is a blood test that requires only a few drops of blood, collected 1-3 days after a baby's 1st feed. This screening test will detect certain rare, but serious metabolic conditions that may not be apparent at birth. Early diagnosis and treatment may prevent debilitating complications.