Your baby's first test, because the best protection you can give is early detection

IMPORTANT FACTS:

Tests for metabolic and genetic disorders that are not apparent at birth

Early detection saves lives

Repeat tests may be required in up to 10% of cases

These disorders are rare

Other services along the Next Biosciences Pregnancy Journey:

TriScreen
Non-invasive Prenatal Testing

Netcells
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FirstScreen

Newborn Screening

What is Newborn Screening?

Newborn screening is recognised all over the world as a form of preventative health care. Newborn screening tests babies in their first few days of life for selected genetic and metabolic diseases that are often not apparent at birth. These congenital (from birth) diseases can progress rapidly to severe illness if not detected early and treatment started. Severe complications such as brain damage, enlargement of the heart, liver, spleen and possibly even death, can be prevented by this simple test.

Next Biosciences offers FirstScreen – a newborn screening test done in collaboration with North-West University and includes tests relevant to the South African population.

Should all newborns be screened?

These diseases are rare. However, since we do not know which child may be at risk of a metabolic disease, international practice recommends that all children undergo newborn screening.
By testing all children, we are assured of finding the few that are affected, for whom early detection and treatment of disease is vital.

When should newborns be screened?

The ideal time is **between 24 and 72 hours after the birth** but up to a week after birth is still acceptable. The baby needs to have had a few feeds and started the digestion and metabolism of proteins, carbohydrates and fats (all found in breast milk and formula milk) to bring the disorder to light.

Tests done outside the ideal timeframe may still be useful but they become less accurate.

How is the screening performed?

1. The test must be done **1-3 days after birth**.
2. **Ask your paediatrician** to order the test.
3. A trained sister will come to you to **take the test**.
4. **A few drops of blood** will be taken from your baby's heel (in some cases, instead of a heel prick, blood may be drawn from the baby's vein) and put on a special blotting card
5. The card will be couriered to **North-West University** where the testing will be done.
6. Next Biosciences will inform you and your **paediatrician of the result**.

What does it mean if a re-test is required?

Sometimes, the laboratory at North-West University may request that the test is repeated. It does not necessarily mean there is anything wrong with the baby – it may be that the sample was not collected properly and not enough blood was available for testing. Repeat tests may be requested in about 10% of cases.

What if my baby's result is abnormal?

If something is found to be abnormal with the results, a repeat test and sometimes additional blood and urine tests may be required. The request for further testing must not alarm you as it does not mean that there is something wrong with the baby. Often, when the first test suggests a problem, the results cannot be considered final until a repeat test is done. This requires a new blood sample. Your paediatrician will discuss the need for further tests with you.

When will I know the results of the test?

You will be notified of the result within 7 working days of the test being done.

What happens if my baby is diagnosed with a disorder?

If a test comes back positive for any of the disorders, your paediatrician will contact you immediately. Each disorder is treatable in its own way even though they are not curable. Your paediatrician will guide you through the explanation of the disease and the ways in which it can be treated. Next Biosciences is also able to organise a genetic counsellor to counsel you about the implications of the disease.

How are these conditions treated?

In most instances, treatment consists of dietary modifications, dietary supplementation, hormones and sometimes medication. If your baby has one of these conditions, it is very important that treatment is started as soon as possible.

Are there risks involved in Newborn Screening?

The baby will feel a little discomfort during the blood collection procedure and may cry a bit. The testing is not harmful at all.

What conditions are screened for?

24 conditions are screened for in the Newborn Screen, including Cystic Fibrosis and Galactosaemia.

Below is a full list of the disorders that are screened.

Amino Acid Disorders

1. Citrullinaemia, Type I
2. Classic Phenylketonuria
3. Homocystinuria
4. Maple Syrup Urine Disease
5. Tyrosinaemia, Type I

Organic acid disorders

6. 3-Hydroxy-3-Methyglutaric Aciduria
7. 3-Methylcrotonyl-CoA Carboxylase Deficiency
8. Glutaric Acidaemia Type I
9. Holocarboxylase Synthase Deficiency
10. Isovaleric Acidaemia
11. Methylmalonic Acidaemia (Cobalamin disorders)
12. Methylmalonic Acidaemia (methylmalonyl-CoA mutase)
13. Propionic Acidaemia
14. ß-Ketothiolase Deficiency

Disorders of fatty acid oxidation

15. Carnitine Uptake Defect/Carnitine Transport Defect
16. Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
17. Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or Glutaric Acidaemia II)
18. Medium-chain Acyl-CoA Dehydrogenase Deficiency
19. Very Long-chain Acyl-CoA Dehydrogenase Deficiency

Disorders of carbohydrate metabolism

20. Classic Galactosaemia

Endocrine disorders

21. Congenital Adrenal Hyperplasia
22. Primary Congenital Hypothyroidism

Other Disorders

23. Biotinidase Deficiency
24. Cystic Fibrosis

Visit the Next Biosciences website for more information on these disorders and to book a test - **www.nextbio.co.za**