

# Newborn Screening Digital

FEBRUARY 2013

## WHAT IS NEWBORN SCREENING?

Newborn screening happens when your baby is tested for possible harmful disorders that are not identified in any way other than through blood tests.

These disorders are metabolic disorders (also called “inborn errors of metabolism”) which are passed from one generation to another. If the baby inherits an abnormality this interferes with normal metabolism. Newborn screening can also detect other problems.

Metabolic and other inherited disorders can interfere with your baby’s development. Parents may pass along the gene for a certain disorder without even knowing that they carry the gene.

With a simple and inexpensive blood test, we can detect whether newborns have certain conditions that can cause problems. Even though these conditions are considered rare, early diagnosis and treatment can make the difference between lifelong impairment or disability and normal development.

## WHO NEEDS TO BE TESTED?

All newborn babies should be tested at two to three days old up to seven days of age.

## WHAT ARE THE BENEFITS OF NEWBORN SCREENING?

When diagnosed early, treatment can be started early, minimising the effect of the disorder. These effects can be seen early, in the newborn period, or later, sometimes only in adulthood. The effects of these disorders range from mild to severe, including severe brain damage or enlargement of the heart, liver and spleen, for example.

## HOW IS THE BABY TESTED?

The best time to do the test is at two to three days of age although up to seven days old is acceptable. A heel prick blood specimen is taken and placed on a special filter paper card. The heel prick is a minor procedure that causes minimal discomfort.

## WHAT CAN YOU EXPECT FROM THE TEST?

Once you have decided to have your baby screened, inform your health care provider. A trained Lancet sister will collect the sample. We send it to our central laboratory in Richmond in Johannesburg for testing and the result will be given to your health care provider.

## WHAT DOES THE RESULT MEAN?

Because these disorders are rare, it is most likely that your baby’s test results will be normal.

## ABNORMAL RESULTS

Lancet will contact your health care provider if your screening test is abnormal. They will ask you for another sample for a repeat or confirmatory testing.

An abnormal, or positive result for any substance means that more tests need to be done to rule out false positives. What this means is that your baby could test positive for a genetic disorder but that the result is actually negative and that your baby is healthy.

There are many reasons for false positive results. Sometimes the specimen is incorrectly collected, or the false result relates to the baby’s diet or medical treatment.

Unfortunately, this might mean that new parents worry for a brief period, while the tests are being repeated. But the benefits outweigh the risks of this.

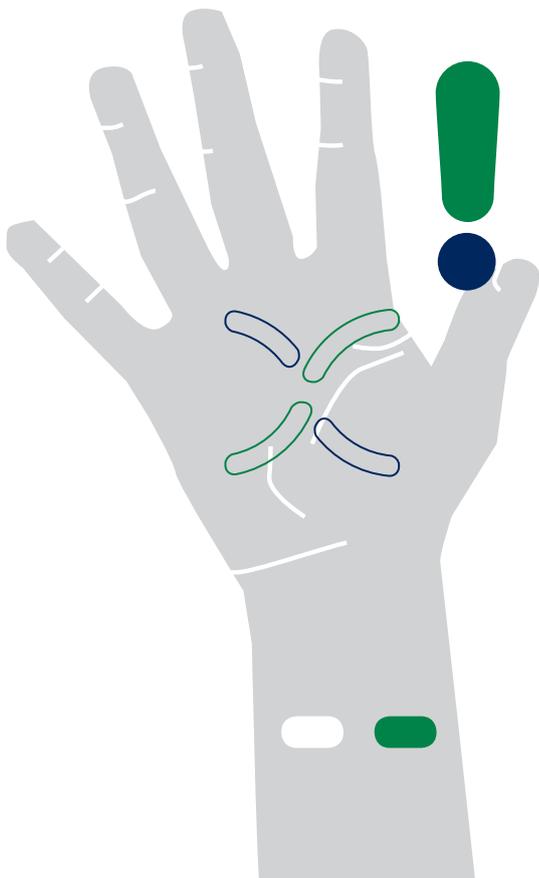
Most parents would rather be anxious about the outcome for brief period of time, to find out about life-threatening illnesses before the symptoms start, rather than risk not testing and not knowing about a critical illness until it’s too late. Most false-positive results are resolved quickly. If the confirmatory test is positive, ask your doctor to advise you on getting the care and advice you need.

## IMPORTANT POINTS TO REMEMBER

This test is done to screen your baby for possible genetic and inherited disorders.

- These disorders are rare.
- If one of these disorders is diagnosed early, treatment may prevent complications.  
An unexpected result needs to be investigated further, with a repeat sample.
- Newborn screening saves lives.

Visit the Lancet website for a Lancet facility near you.



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