

TO PARENTS

A heel prick blood test to screen for certain serious disorders is available for all newborn babies in Denmark.



STATENS
SERUM
INSTITUT

*prevents and controls
infectious diseases,
biological threats and
congenital disorders*

BLOOD TEST FOR BABIES



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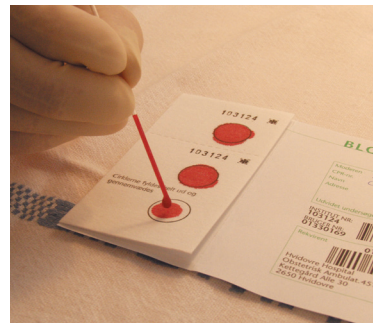
INFORMATION ON THE HEEL PRICK TEST

A simple blood test can be carried out 48-72 hours after birth to check whether your baby has any congenital disorders that may need urgent treatment.

This blood test cannot be performed unless you, as parents, give your permission. If you say no, the test will not be carried out.

How is it done?

A few drops of blood are collected on a filter paper card after pricking the skin on one of the baby's heels. This rarely causes the baby any discomfort.



How important is this test for your baby?

Even if a baby appears completely healthy at birth, he or she may, in rare cases, have a congenital disorder, such as a metabolic defect. As long as babies are still in the womb, they are protected by the mother's metabolism. Disorders of this type therefore do not become apparent until after birth. The disorder may develop slowly as harmful metabolic products build up in the blood. Or it may develop suddenly, in the form of metabolic crises that are potentially fatal for the baby. The longer treatment is delayed, the greater the risk of the baby dying or sustaining lasting mental or physical harm. For the baby's sake, it is therefore very important to detect such a disease and start treatment as quickly as possible.

What specific disorders are screened for?

On the Statens Serum Institut website www.ssi.dk/nyfoedte you will find a list of the rare congenital disorders currently screened for, and more detailed information on the individual disorders. Some disorders are also described at the back of this booklet. If your baby has one of these disorders, you will be informed immediately and asked to bring your baby in for further examination and treatment at the local paediatric department, often in cooperation with the Kennedy Centre and the clinical genetics departments of Copenhagen University Hospital and Skejby Hospital.

How reliable is the test?

As it is a 'screening' test, in rare cases, results might be obtained that suggest a disorder is present when it is not, particularly in the case of premature babies. This will quickly become apparent in the more detailed examination that immediately follows. Nor can screening rule out all disorders. Firstly it is not technically possible to screen for all congenital disorders, and secondly screening is carried out only for disorders for which early detection means better treatment options that benefit the baby.

OTHER INFORMATION

What happens to the blood after the test has been performed?

The sample will be kept frozen in locked and secure facilities in the Neonatal Screening Biobank at Statens Serum Institut. Storing the sample in a biobank means that the test can be repeated if there is any subsequent doubt about the diagnosis, or additional analyses can be performed that were not available at the time of birth. In rare cases, the sample has also proved important in the reliable identification of a person who is the victim of an accident, natural disaster, crime, etc. later in life. The aim is always to have an adequate sample for these purposes. Any surplus sample material, as in all other laboratories that analyse blood samples, is used in regular quality assurance and the development of new analysis methods.

The biobank also serves as a national resource of great importance to medical research. Use for research purposes, however, depends on the approval of a research ethics committee and the Danish Data Protection Agency. The biobank's management committee must also approve this use.

The individual blood sample is given a code number at Statens Serum Institut. Unauthorised individuals cannot therefore identify the source of the sample. Information on the child's name, date of birth and birth weight and the length of the pregnancy, as well as the mother's name and civil registry number, are kept separately from the sample itself.

You decide what happens to the sample on behalf of your baby

If you do not want the sample to be used in medical research for ethically approved purposes, you can inform the tissue use registry of the Danish National Board of Health. You can find further information on this at www.sundhedsstyrelsen.dk/vaev, where you will also find an article entitled 'Dit væv, dit valg' ('Your tissue, your choice').

If you do not wish the sample to be retained at all, write to the Section for Neonatal Screening, KBI, Statens Serum Institut and the sample will then be destroyed.

Statens Serum Institut's website www.ssi.dk/nyfoedte also has more detailed information on the Neonatal Screening Biobank.

EXAMPLES OF CONGENITAL DISORDERS

Hormonal disorders

Congenital hypothyroidism (underactive thyroid) is due to the inability of the baby to produce enough of a hormone that controls metabolism. If left untreated, this disorder leads to stunted growth and brain damage.

If the disease is detected early, the child can develop normally with hormone treatment in the form of tablets.

Congenital hypothyroidism occurs in around one in 3,400 babies.

Metabolic disorders

This large group of congenital disorders arises due to the inability to convert certain substances. The disorders are difficult to detect if screening is not carried out, and can lead to organ damage, severe retardation of mental development and death. The treatment usually consists of a special diet with restricted intake of the foodstuff the baby cannot tolerate. One example is phenylketonuria (PKU), in which the amino acid phenylalanine cannot be converted. With another disorder, MCAD, the baby is unable to burn fat, which together with insufficient food intake, can result in acutely low blood sugar levels leading to convulsions and death. It is therefore important to know about this disorder so that such cases can be prevented simply by giving sugar. Metabolic disorders altogether occur in one in 3,000 babies.

If screening indicates that your child has one of these disorders, you will be informed immediately and will be asked to take your baby to the nearest paediatric department for further examination and treatment.

This information was produced in consultation with the Danish National Board of Health.