Screening-Labor Hannover
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Parents' Information Sheet Neonatal Screening

Für die Patientenakte. Kopie mit Unterschriften an den/die Personensorgeberechtigte aushändigen

Neonatal screening is an essential preventive measure

Neonatal screening was first introduced in 1964. Since then many infants have been identified early enough to be treated successfully. Guidelines of pediatric and medical associations strongly recommend newborn screening. It has been demonstrated worldwide that neonatal screening is essential to prevent potentially catastrophic health problems by early detection of certain metabolic disorders. You too should make sure that your baby has been tested!

Dear parents,

Our very best wishes to you on the birth of your baby.

Unfortunately, a few children are carrying certain metabolic defects. In these cases, mental and physical retardation or signs of disease can often be prevented by simple measures if the disorder has been diagnosed early enough. In neonatal screening for inborn errors of metabolism, newborns are therefore tested on the 3rd day of life (age must be at least 36 hours).

Even when a baby appears to be well, treatment may be required to ensure further normal development. Treatment might consist of a hormone or a vitamin tablet or a special diet might be necessary.

Blood tests

A few drops of blood are taken from your baby's heel and spotted onto special filter paper. The dried blood sample is then sent to the screening-laboratory. According to guidelines provided by German pediatric societies the blood is tested for disorders of hormone metabolism (congenital hypothyroidism, congenital adrenal hyperplasia), of sugar metabolism (galactosemia), for biotinidase deficiency, and a number of disorders of amino acid metabolism (including Phenylketonuria) and for disorders of fatty acid oxidation (including MCAD-deficiency).

The test procedures applied in our lab are undergoing regular revision. We also take part in national and international quality assurance programs.

Additional tests

Further tests may be performed on request including screening for sickle cell disease, glucose-6-phosphate dehydrogenase deficiency and cystic fibrosis.

A single blood sample is usually sufficient

Blood for neonatal screening is usually drawn on the third day of life between the 36th and 72nd hour of life. If you and your child have been discharged from the hospital prior to this, the midwife or your local pediatrician should take a further blood sample at the required time (see above).

A further blood sample may also be required by the laboratory to repeat some of the tests. The fact that the laboratory requests a second sample in some cases does not necessarily mean that your child has been diagnosed with a metabolic disorder. Some babies have not yet completely adapted to postnatal life; this can cause false positive results that have to be checked in order to exclude any inborn error of metabolism.

For the majority of babies the test result will be normal

In these cases, further measures are rarely necessary. However, it is important to keep in mind that the screening tests are not valid to detect inherited diseases under all possible conditions prevailing during the first days of life. If any signs of disease show later in life, your child should be thoroughly examined by a physician/pediatrician.

What to do if a certain disorder has been suspected?

Such children can be treated successfully in almost all cases. First of all, a detailed clinical examination is necessary, either by a pediatrician or in a children's hospital. Usually, further blood and urine tests will then be performed.

If the provisional diagnosis is confirmed, doctors will inform you about the treatment. Sometimes it may be sufficient to take a certain medication, in other cases a special diet may be required to ensure normal development of your child.

Who will receive the test result?

The results are strictly confidential. They are transmitted to the hospital, midwife or physician identified on the order form.

List of disorders:

- Congenital hypothyroidism
- Congenital adrenal hyperplasia (21CAH)
- Galactosemia (galactose-1-p-uridyltransferase deficiency)
- Biotinidase deficiency
- Maple sirup urine disease
- Phenylketonuria
- MCAD deficiency
- VLCAD deficiency
- LCHAD/TFP deficiency
- Isovaleric acidemia
- Glutaric acidemia I
- Defects of carnitine metabolism
- Tyrosinemia type I
- Severe combined immunodeficiency (SCID)

Additional tests:

- Sickle cell disease
- Glucose-6-phosphate dehydrogenase deficiency
- Cystic fibrosis (see back page)
- Additional acylcarnitines and amino acids

Neonatal screening for:

Name: _____

Date of birth: _____

I have been informed about the Neonatal screening by:

_____

I had enough time for consideration.

I have read the information sheet "Parents' Information Sheet Neonatal Screening" (see above). I had the opportunity to ask questions about any tests and procedures described above.

_____

_____

I have received a copy of the parents' information sheet. I can revoke the consent to screening at any time. In this case the analyses will be stopped.

I am aware, of the risk that some diseases may only be detected and treated at a later stage of the disease if I deny Neonatal Screening.

_____

Date, signature of person entitled to custody
Datum und Unterschrift des/der Sorgeberechtigten

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Parents' Information Sheet Cystic Fibrosis (CF) Screening

Für die Patientenakte. Kopie mit Unterschriften an den/die Personensorgeberechtigte aushändigen

Dear Parents,
Together with the existing neonatal screening we now offer cystic fibrosis screening. Since this test may include molecular genetic analysis the Genetic Diagnostics Act has to be applied. The following information will help you to prepare for an informed consent discussion with your doctor.

What is cystic fibrosis?
Cystic fibrosis (CF) is a genetic disorder. About one in 3.300 children is affected. The causative mutation leads to impaired transport of salt in and out of the gland cells. This dysfunction causes production of thick and sticky mucus in the lungs and other organs leading to chronic inflammation. Symptoms vary depending on different mutations. Pancreas function is often impaired. This is why affected children show reduced growth and have problems gaining weight. In severe cases lung functions may significantly be reduced due to chronic inflammation.

How can cystic fibrosis be treated?
Currently there is no curative therapy. However a number of therapeutic measures help to alleviate symptoms, so that life expectancy of cystic fibrosis patients has increased continually over the past years. Therapy includes inhalations and physiotherapy, high caloric diet and certain drugs. In addition regular clinical monitoring can help to tailor the needs of the patients.

Why is cystic fibrosis screening a medically sensible test?
Cystic fibrosis screening allows early diagnosis and start of therapy so that physical development can be significantly improved leading to increased life expectancy and quality.

How is cystic fibrosis screening done?
Cystic fibrosis screening is done at the same time as neonatal screening. No additional test card is required. First immunoreactive trypsin (IRT) is analysed. If this test shows a normal result, no further analyses are required. If the IRT concentration is elevated, a second test (pancreatitis associated protein, PAP) will be performed using the original blood card. Only if PAP is elevated too, a molecular genetic analysis will be performed. In this analysis 31 of the most common mutations are tested. If at least one mutation is detected, the result is "pathological", meaning that further diagnostic tests and clinical investigations are necessary.

In cases of strongly elevated IRT, the screening result is estimated "pathological" based on this test alone and should be followed by further diagnostic procedures and clinical investigations immediately. This combination of test procedures allows the best diagnostic outcome. However, in rare cases the screening may not detect a child suffering from cystic fibrosis. According to the German Genetic Diagnostic Act parents have to give informed consent. If a midwife has taken responsibility for the birth of your child, the screening for cystic fibrosis can be requested by your paediatrician after informed consent within the first 4 weeks after birth. In this case a second blood sample is required.

How are you being notified about the results?
Within 14 days the laboratory provides the result of the cystic fibrosis screening. If further diagnostic tests are necessary, your doctor / hospital will inform you about the results and what to do next. Usually, you will be advised to contact a cystic fibrosis specialist (Cystic Fibrosis Centre). Only one in five children with a pathological result is confirmed to suffer from cystic fibrosis. The diagnosis can usually be verified by performing a "sweat test" in the Cystic Fibrosis Centre. You will get the results of this test immediately afterwards. In some cases further diagnostic procedures may be necessary.

You decide for your child!
Cystic fibrosis screening is an optional test. You may revoke your consent at any time. The results underlie medical confidentiality and cannot be shared with a third party unless you give your consent. After the laboratory tests have been performed the results are directly forwarded to the doctor in charge of contacting you. Your decision to agree or disagree to cystic fibrosis screening should be done after thorough consultation. Please talk to your doctor if any questions arise. Your agreement includes the performance of the respective tests as well as transfer of required personal data.

Costs
Screening tests for newborns are covered by the statutory health insurance.
Patients with private insurance will receive an invoice according to the German Scale of Medical fees (GOÄ).

Cystic fibrosis screening for:

Name: _____

Date of birth: _____

I have been informed about the cystic fibrosis screening by:

_____

I had enough time for consideration.

I have read the information sheet "Cystic Fibrosis Screening" (see above) I had the opportunity to ask questions about any tests and procedures described above.

_____

_____

I have received a copy of the parents' information sheet. Cystic fibrosis screening is usually performed at the same time as the newborn screening. I can revoke the consent to screening at any time. In this case the analyses will be stopped.

I am aware, of the risk that cystic fibrosis may only be detected and treated at a later stage of the disease if I deny cystic fibrosis screening.

_____

Date, signature of person entitled to custody
Datum und Unterschrift des/der Sorgeberechtigten