UNIVERSITÄTS
KLINIKUM
HEIDELBERG

NEUGEBORENENSCREENING
HEIDELBERG

NEWBORN SCREENING HEIDELBERG

PARENT INFORMATION

Dear parents,

Your child is about to be born or has just been born, and we wish you all the best for you and your family. Most children are healthy at birth, and remain so. However, there are rare congenital diseases which cannot be clinically detected in newborn infants. Without early detection and treatment such diseases may cause serious health impairments of the child. In order to prevent these, in Germany important early diagnostic examinations (extended newborn screening) for all newborn infants in the first days of life are advised. These examinations are paid for by your health insurance. Participation in newborn screening is voluntary. If you want to have these examinations carried out on your child, you need to fill out the included declaration of consent and sign it (at least one parent or one legal guardian).

Newborn screening for congenital metabolic diseases and endocrine disorders

Without early treatment, certain congenital metabolic diseases and endocrine disorders may cause severe disabilities, or even death. The consequences of a congenital disease can usually be avoided or reduced by administration of certain drugs or by following a special diet. Blood sampling for newborn screening is preferably done between the second to third day of life (37 to 72 hours after birth) from a few drops of blood placed on a filter paper card and sent immediately to a screening laboratory. You can find a full description of the tests and individual disorders starting on page 2.

Newborn screening for cystic fibrosis

Together with newborn screening for congenital metabolic diseases and endocrine disorders we offer screening for cystic fibrosis from the same blood sample. Children with cystic fibrosis suffer from chronic inflammation due to the production of thick and sticky mucus in the lungs and other organs. As a consequence, children often show failure to thrive and reduced growth.

In severe cases the pulmonary function may be significantly affected. The aim of early diagnosis of cystic fibrosis is to start with specific therapy as early as possible, which helps to improve the quality of life and the life expectancy of the child. In accordance with the legal requirements of the German Genetics Diagnostics Act (Gendiagnostikgesetz), a consultation with a doctor is mandatory before newborn screening for cystic fibrosis. More information about the disorder and the test can be found on page 5.

After these examinations have been completed, the test card with the dried blood spots will be retained for 3 months and will then be destroyed in accordance with legal requirements. In case of a conspicuous/abnormal result further testing is required. For this reason we ask for your consent to the data transfer between the continuing care center and our screening center until all findings have been reviewed.

Universitätsklinikum Heidelberg
Zentrum für Kinder- und Jugendmedizin
Dietmar Hopp Stoffwechselzentrum
Neugeborenenscreening
Im Neuenheimer Feld 669
69120 Heidelberg

Tel. 06221 56-8278; 8475
Fax 06221 56-4069

www.neugeborenenscreening.uni-hd.de

UK HD

NEWBORN SCREENING FOR CONGENITAL METABOLIC AND ENDOCRINE DISORDERS

There are rare congenital metabolic diseases and endocrine (hormonal) disorders which cannot be detected by external signs in newborns. These diseases occur in approx. 1 in 1,200 newborns. If left untreated, they lead to organ damage, physical or mental handicaps. In order to detect these diseases, a blood test – known as newborn screening – is recommended for all newborns. This has been successfully performed in Germany for more than 50 years and thousands of children have benefited.

Why is newborn screening performed?

Rare congenital metabolic and endocrine disorders should be diagnosed at an early stage. Severe consequences of a congenital disease can usually be avoided by early treatment, when initiated directly after birth.

When and how is the examination performed?

Between the second to third day of life (37 to 72 hours after birth), possibly together with the second preventive examination of your child, the U2, a few drops of blood (taken from the vein or by heel prick) will be placed on a filter paper card. After drying, the dried blood will be sent immediately to the screening laboratory. Here, the samples will be analyzed on the day of arrival with specialized and highly sensitive examination methods. The costs of the examination will be covered by your health insurance or the clinic.

Which diseases can be detected?

The diseases for which we examine the blood samples are prescribed in a compulsory directive of the Federal Joint Committee (Gemeinsamer Bundesausschuss der Ärzte und Krankenkassen, G-BA). These include 13 metabolic diseases and 2 hormone disorders. The consequences and symptoms of these diseases are described in more detail below.

Overall, one of these diseases is found in about one in every 1,200 newborns. In most families with an affected child these diseases have never occurred before. Since the affected children are still completely healthy at birth, newborn screening can prevent impairments of mental and physical development. No conclusions on family risks can be drawn from this examination alone.

Who will receive the test result?

No matter the result, the sender of the blood sample will receive a written report from the screening laboratory within a few days. The sender informs the parents in case of an abnormal result. In urgent cases, for example when the sender is not available, the laboratory will contact the parents directly. For this reason, your telephone number and the address at which you can be contacted are required on the test card. Early detection and early treatment of affected infants is only possible if all those involved – parents, hospital, pediatrician, and screening laboratory – can work together as quickly as possible.

What does the test result tell us?

The result of a screening test is not yet a medical diagnosis. The test results can either largely exclude the targeted disorders or, in case of a suspected disease, can indicate further diagnostic examinations, for example a repeat of the test. A repetition of the test may also benecessary if the blood sample was not taken at the ideal time or if the amount of blood on the card was insufficient.

Can these diseases be cured?

All mentioned metabolic defects and endocrine disorders are congenital and therefore cannot be cured. However, the effects of these congenital disorders can often be completely avoided or at least reduced by early treatment. The treatments consist of a special diet and / or special medications. Medical specialists are available for consultation and care in case of suspected or identified disease.

Data collection and processing

The test card which is sent to the newborn screening center of the Center for Pediatric and Adolescent Medicine at the University Hospital Heidelberg (Zentrum für Kinder- und Jugendmedizin, Universitätsklinikum Heidelberg) contains information on your child. In addition to the name (important for the correct assignment of the examination results) we need the date of birth and pregnancy week in order to assess the laboratory results correctly. We also ask for your name, address and telephone number on the test card. We need these data to notify you immediately if your child requires urgent medical treatment (this happens only in very few cases). The personal data of your child and your data on the test card will be stored in the laboratory information system of the Heidelberg newborn screening center. The data are retained for 10 years, in accordance with the regulations for medical reports in Germany. Only staff of the newborn screening center will have access to these data when necessary. The data will be deleted after ten years. Your data, your child's data, and collected blood samples will not be used for anything other than the specified purposes, and will not be disclosed to unauthorized third parties.

You have the right to request information on the stored personal data of your child from the responsible person. You also have the right to request correction of inaccurate data, to request erasure of data, and to restrict further processing of data. The person responsible for processing of personal data required as part of newborn screening is:

Prof. Dr. med. Prof. h.c. mult. (RCH) Georg F. Hoffmann
Tel.: 06221 56-4002
E-Mail: georg.hoffmann@med.uni-heidelberg.de

Target diseases

(Prognosis always refers to the results after early diagnosis and consecutive specific therapy)

Biotinidase deficiency (BTD)

Defect in the metabolism of the vitamin biotin: skin changes, metabolic crises, seizures, mental handicap, possibly fatal. Treatment by supplementation of biotin, very good prognosis (Frequency: approx. 1/30,000 newborns).

Congenital adrenal hyperplasia

Hormone disturbance due to defect of the adrenal cortex: masculinisation in females, possibly fatal in case of salt-wasting crises. Treatment by supplementation of hormones, good prognosis (Frequency: approx. 1/14,000 newborns).

Carnitine cycle disorders

Defects in the metabolism of fatty acids: metabolic crises, coma, muscle diseases, possibly fatal. Treatment by special diet, very good prognosis (Frequency: approx. 1/500,000 newborns).

Galactosemia

Defect in galactose metabolism (milk sugar component): cataract, physical and mental handicap, liver failure, possibly fatal. Treatment by special diet, mostly good prognosis (Frequency: approx. 1/70,000 newborns).

Glutaric aciduria type I

Defect in the breakdown of amino acids: sudden metabolic crises with permanent brain damage and movement disorders. Treatment by special diet, mostly good prognosis (Frequency: approx. 1/120,000 newborns).

Hypothyroidism

Congenital deficiency of thyroid hormones: Severe impairment of mental and physical development. Treatment by supplementation of thyroid hormone, very good prognosis (Frequency: approx. 1/3,500 newborns).

Isovaleric acidemia

Defect in the breakdown of amino acids: mental handicap, coma, possibly fatal. Treatment by special diet, very good prognosis (Frequency: approx. 1/100,000 newborns).

LCHAD-, VLCAD-deficiency

Defect in the metabolism of long-chain fatty acids: metabolic crises, coma, muscle and heart muscle weakness, possibly fatal. Treatment by special diet, avoidance of fasting, mostly good prognosis (Frequency: approx. 1/80,000 newborns).

MCAD-deficiency

Defect in energy production from fatty acids: hypoglycemia, coma, possibly fatal. Treatment by avoidance of fasting, very good prognosis (Frequency: approx. 1/10,000 newborns).

Phenylketonuria / Hyperphenylalaninemia

Defect in the metabolism of the amino acid phenylalanine: in phenylketonuria without treatment mental handicap. Successful treatment by special diet, very good prognosis. Mild hyperphenylalaninemia does not require dietary treatment, but necessitates exclusion of BH_4 cofactor defects. (Frequency: approx. 1/5,000 newborns).

Tyrosinemia type I

Defect in breakdown of tyrosine, if untreated leading to liver failure within the first days of life with icterus and bleeding tendency, kidney dysfunction, and neurological crisis. Treatment by special medication (Nitisinone) and by a protein controlled diet, good prognosis (Frequency: approx. 1/135,000 newborns).

Note

Early treatment cannot completely prevent all symptoms in all cases of the abovementioned disorders. However, most children will develop normally after timely initiation of treatment.

For information and concerns about data processing and data protection you can contact our data protection officer:
Datenschutzbeauftragter Universitätsklinikum Heidelberg
Im Neuenheimer Feld 672, 69120 Heidelberg
E-Mail: Datenschutz@med.uni-heidelberg.de

In case of illegal data procession you have the right to complain to the supervisory authority:
Der Landesbeauftragte für den Datenschutz und
die Informationsfreiheit Baden-Württemberg
Postfach 10 29 32, 70025 Stuttgart
Königstraße 10a, 70173 Stuttgart
Tel.: 0711 615541-0, Fax: 0711 615541-15
E-Mail: poststelle@lfdi.bwl.de
Internet: http://www.baden-wuerttemberg.datenschutz.de

Right of revocation

Participation in newborn screening is voluntary. You may revoke your consent at any time. In this case test cards of your child will be destroyed, and your personal data in our computer system will be blocked, preventing further access.

Residual blood sample

The test cards with the remaining blood will be destroyed after a storage time of 3 months.

If you agree to all provided examinations, please sign the declaration of consent on the back.

Only fill out this page if you do NOT fully agree to the screening program.

Name of child: _____

Date of birth: _____
(or adhesive label)

I have been informed about newborn screening for congenital metabolic diseases / hormone disorders and cystic fibrosis. I have been informed about the possible negative consequences for my child in case of my rejection of selected parts of newborn screening.

Differentiated declaration of consent

(Please sign at each point you agree with.)

I agree to the following examinations and the transfer of data required for this purpose:

◯ Newborn screening for congenital metabolic diseases and hormone disorders (pages 2-3)

_____

Date, name (ITALICS) and signature of at least one legal guardian

◯ Newborn screening for cystic fibrosis (pages 5-6)

_____

Date, name (ITALICS) and signature of at least one legal guardian

◯ In case of a newborn screening result requiring subsequent examinations, I agree to the data transfer to the newborn screening center Heidelberg by the continuing care center until the findings have been clarified (Data transfer tracking).

_____

Date, name (ITALICS) and signature of at least one legal guardian

_____

Date, name (ITALICS) and signature of responsible physician according to §8 subsection 1, of the German Genetic Diagnostics Act

This declaration of consent remains with the sender of the sample.
The consent for newborn screening or rejection of selected parts of the screening program must be noted on the filter paper card for newborn screening in the fields provided.

NEWBORN SCREENING FOR CYSTIC FIBROSIS

Together with newborn screening for inherited metabolic and endocrine disorders, we offer newborn screening for cystic fibrosis from the same blood sample. The aim of this examination is the early diagnosis of cystic fibrosis, in order to start treatment as early as possible and thus improve quality of life and life expectancy of affected children. Screening for cystic fibrosis is subject to the regulations of the German Genetic Diagnostics Act (Gendiagnostikgesetz). The following information will help you to prepare for an informed consent discussion with your doctor.

What is cystic fibrosis?

Cystic fibrosis is a genetic disorder affecting approximately 1 in 4,800 children. The causative gene mutations in the so-called CFTR gene lead to an impairment of salt exchange in glandular 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 gene mutations. Pancreatic function is also often impaired. This causes that affected children show reduced growth and have problems gaining weight. Lung function is mostly impaired due to chronic inflammation and recurrent lung infections.

How can cystic fibrosis be treated?

Currently there is no curative therapy for cystic fibrosis. However, various therapeutic approaches are used to alleviate symptoms of the disease so that health and life expectancy of patients with cystic fibrosis patients have continuously increased. Therapy includes inhalations and physiotherapy, a particularly high caloric diet and certain drugs. In addition, regular clinical monitoring in cystic fibrosis centers helps to initiate and adapt required therapies timely.

Why is newborn screening for cystic fibrosis useful?

Newborn screening for cystic fibrosis allows for early diagnosis and start of treatment, so that physical development of the affected children can be improved. This helps to improve the health of affected children and raises the prospects of a longer life expectancy.

How is screening for cystic fibrosis performed?

Newborn screening for cystic fibrosis does generally not require additional blood sampling. The screening test is done at the same time and from the same blood sample which is taken from your child for newborn screening for metabolic and endocrine disorders. For this, a few drops of blood (taken from the vein or by heel prick) are spotted on a filter paper card which is sent to a screening laboratory.

First the enzyme immunoreactive trypsin (IRT) is determined. If this test shows a normal result, no further analyses are required. If the IRT concentration is elevated, a second test for the pancreatitis associated protein (PAP) will be performed using the original blood card. Only if PAP is also elevated will a DNA test (molecular genetic analysis) for the most common gene mutations be performed from the filter paper card. If one or two gene mutations are detected, the screening result is reported as abnormal. This means that further diagnostic work-up is required as explained below. If the first test (IRT) is very conspicuous due to strongly elevated IRT, the screening result is already at this step labeled “abnormal” based on this test alone and the other laboratory tests are not performed. This combination of test procedures allows for the best diagnostic efficiency. However, in rare cases a child affected by cystic fibrosis may not be detected by newborn screening.

According to the legal regulations of the German Genetic Diagnostics Act, counseling of parents by a physician is compulsory before newborn screening for cystic fibrosis is performed. If a midwife alone was the primary caregiver at delivery, newborn screening for cystic fibrosis can be requested by your pediatrician after informed consent within the first 4 weeks after birth. In this case a second blood sample is required. However, newborn screening for congenital metabolic and endocrine disorders should still be done within the first 72 hours of life, as in contrast to cystic fibrosis an immediate initiation of treatment is crucial for the majority of these disorders.

Your child’s blood sample will be stored for 3 months after performing the tests and will be subsequently destroyed.

How are you notified of the screening result and what has to be done subsequently?

The laboratory informs the sender (physician) of the blood sample within 14 days of receipt of the sample whether the results of cystic fibrosis screening are abnormal or normal. In case of a normal result you will be informed only on your explicit request. If further diagnostic work-up is necessary due to an abnormal screening result, your doctor/hospital will contact you and refer you to a cystic fibrosis center. A positive result does not necessarily mean your child is affected by cystic fibrosis. Only one in five children with an abnormal screening result is finally confirmed to be affected by cystic fibrosis. However, children with "false-positive" cystic fibrosis screening have an increased probability to be carriers of one (heterozygous) mutation in the CFTR gene. These gene carriers are healthy, however, they can pass the mutation on to their children. In either case a genetic counseling will be offered to you, so that you can inform yourself about the relevance of the results.

To verify the suspected diagnosis, the cystic fibrosis center will usually perform a sweat test as first examination. On your appointment you will receive further information and counseling. This sweat test is safe and painless and does not stress your child. You will get the results of this test immediately afterwards. In some cases further diagnostic procedures may be necessary.

You decide for your child!

Participation in cystic fibrosis screening is voluntary. The costs are covered by the statutory health insurance. The results of the examination are subject to medical confidentiality and will not be disclosed to third parties without your consent. The screening laboratory transmits the results directly to the responsible physician, who is required to contact you in a case of an abnormal result. You may revoke your consent to cystic fibrosis screening at any time. Your decision to agree or disagree to cystic fibrosis screening should be based on substantiated information. Please talk to your doctor if any questions arise.

Newborn screening for cystic fibrosis in its present form has been approved by the Genetic Diagnostics Commission (Gendiagnostik-Kommission) at the Robert-Koch-Institute, Berlin.

ID29055_Unternehmenskommunikation/Medienzentrum Universitätsklinikum Heidelberg

DECLARATION OF CONSENT FOR NEWBORN SCREENING

If you wish your child to receive newborn screening for congenital metabolic diseases / hormone disorders and cystic fibrosis, please sign on this page.

Name of child: _____

Date of birth: _____
(or adhesive label)

◯ I have received the information material on newborn screening and have been adequately informed.

◯ I agree to newborn screening for congenital metabolic diseases / hormone disorders and cystic fibrosis on my child and to the transfer of data.

◯ In case of a newborn screening result requiring subsequent examinations, I agree to the data transfer to the newborn screening center Heidelberg by the continuing care center until the findings have been clarified (Data transfer tracking).

◯ I have been informed about the necessity of a second examination at the latest by _____ (Date). (A second examination is only necessary in very few children.)

◯ I do not consent to newborn screening for my child. I have been informed about the possible negative consequences for my child (undetected disease possibly leading to handicap and death).

_____

Date, name (ITALICS) and signature of at least one legal guardian

_____

Date, name (ITALICS) and signature of responsible physician according to §8 subsection 1, of the German Genetic Diagnostics Act

If you wish only selected examinations to be performed on your child, please complete and sign the declaration on the back.

This declaration of consent remains with the sender of the sample.
The consent for newborn screening or rejection of selected parts of the screening program must be noted on the filter paper card for newborn screening in the fields provided.