KINDER | UKE

Parent information

Newborn Screening

Universitätsklinikum Hamburg-Eppendorf UKE HAMBURG

Dear parents,

The birth of your child is still ahead of you or just behind you. We wish all the best for your child. Most children are born healthy and stay that way. However, there are rare congenital diseases that are not yet recognisable by external signs in newborns. If left untreated, such diseases can lead to serious childhood impairments. To prevent this, important early detection examinations (newborn screening) are recommended for all newborns in Germany in the first few days of life. The costs of the examination are covered by statutory health insurance. Participation in newborn screening is voluntary. In order for your child to undergo these examinations, your consent is required and can be given by signing the consent form. Your consent covers newborn screenings for the target diseases listed below and the processing of personal data for the purpose of carrying out the newborn screening.

Newborn screening for congenital disorders of the metabolic, endocrine, vascular, immune and neuromuscular systems

Rare congenital metabolic diseases, hormonal disorders or disorders of the cardiovascular or immune system and the neuromuscular system can lead to organ damage, physical or mental disability, serious infections or even death if left untreated. If they are recognised early, in most cases the administration of medication, adherence to a diet or other specific measures can prevent or alleviate the consequences of the disease. The test is best carried out on the second or third day of life whereby a few drops of blood are dripped onto a filter paper card and sent to a screening laboratory. The exact procedure of the examination and the individual diseases are described beginning on Page 3.

Newborn screening for cystic fibrosis

At the same time as the newborn screening for congenital metabolic, endocrine, vascular, immune and neuromuscular disorders, you will be offered a screening for cystic fibrosis for your child from the same blood sample. In children with cystic fibrosis, viscous mucus is produced in the lungs and other organs. This causes them to become permanently inflamed. As a result, the children are often underweight and grow poorly. In severe cases, lung function can be significantly impaired. The aim of this examination is to diagnose cystic fibrosis at an early stage so that treatment can be started as soon as possible, thereby improving the quality of life and life expectancy of affected children. In accordance with the legal requirements of the German Genetic Diagnostics Act (Gendiagnostikgesetz), a doctor must be consulted before a newborn screening for cystic fibrosis is carried out. Further information on this disease and the exact examination procedure can be found beginning on Page 6.

In order to ensure that the necessary check-ups have been carried out in the event of abnormal findings, we ask that you consent to the transfer of data by the specialist institution providing further care to our screening laboratory until the findings have been clarified.

University Medical Center Hamburg-Eppendorf
Newborn Screening
Building N22
Martinistraße 52, 20246 Hamburg
Telephone: 040 7410 - 53737
Fax: 040 7410 - 57318

Newborn screening for congenital disorders of the metabolic, endocrine, vascular, immune and neuromuscular systems

There are rare congenital disorders of the metabolic, endocrine, vascular, immune and neuromuscular systems that are not yet recognisable by external signs in newborns. These occur in about one in 1000 newborns. If left untreated, they can lead to organ damage, physical or mental disability, serious infections or even death. In order to recognise these diseases, a blood test, known as newborn screening, has been recommended for all newborns in Germany for over 50 years. In recent years, these tests have been continuously developed so that other treatable diseases have been included in newborn screening.

Why is newborn screening carried out?

These congenital metabolic, endocrine, vascular, immune and neuromuscular disorders should be recognised in good time. Early treatment as soon as possible after birth can usually prevent or alleviate the consequences of a congenital disease.

When and how is the examination carried out?

During the second to third day of life (37 to 72 hours after birth), at the latest during your child's second preventive medical checkup, the U2, a few drops of blood (from a vein or heel) are placed on a filter paper card and sent to the screening laboratory immediately after drying. There, the samples are immediately analysed using special, very sensitive examination methods. The costs of the examination are covered by health insurance. To ensure that your child can always have this important examination, we have also drawn up a contingency concept as a quality assurance measure. In the very unlikely event that our own laboratory is unable to process your child's blood samples due to malfunctions, our cooperation partners (other screening laboratories) can take over the test. If you have any further questions about newborn screenings, you can contact your doctor.

What diseases are tested for?

The diseases for which the blood sample may be analysed are set out in binding guidelines issued by the Federal Joint Committee of Doctors and Health Insurance Funds (G-BA). There are 13 metabolic diseases, 2 hormonal disorders, severe combined immunodeficiency, sickle cell disease (SCD) and spinal muscular atrophy (SMA). The consequences and symptoms of these diseases are described in detail below.

In total, one of these diseases is found in approximately one in 1000 newborns. Most of the diseases investigated are hereditary (genetic). Most of the families affected have never had such illnesses before. As the affected children can still appear completely healthy at birth, newborn screening can prevent mental and physical developmental disorders. No conclusions about family risks can be drawn from this study alone.

Who finds out the test results?

In the event of abnormal test results, you will be contacted directly without delay. For the test card, please enter your telephone number and address where you can be reached in the first few days after the birth. Early detection and early treatment for affected newborns is only possible if all parties involved – parents, clinic or paediatrician and screening laboratory – can work together without losing time. Results that indicate that no abnormalities were detected will only be communicated to you upon your personal request. The results of the newborn screening are subject to medical confidentiality and may not be passed on to third parties without your consent.

What do the test results mean?

The results of a screening test are not considered a medical diagnosis. The test results can either be used to largely rule out the analysed disorders or indicate that a further examination may be necessary if a disease is suspected, e.g. by repeating the test. However, it may also be necessary to repeat a test if, for example, the timing of the blood sample was not optimal or the blood on the card was insufficient. In these cases, provided you agree, the same laboratory should carry out the tests.

What should you do if the screening results are abnormal?

A doctor will advise you on which specialist clinic to take your child to for a diagnostic examination. This is necessary in order to verify the screening findings and possibly initiate therapeutic measures. You will receive comprehensive information about the type of examination and treatment options at this center.

How are these diseases treated?

The metabolic diseases, immunodeficiencies, endocrine and neuromuscular disorders mentioned, as well as sickle cell disease, are congenital and therefore cannot be cured in most cases. Timely treatment cannot completely prevent the consequences of all diseases. However, the effects of these congenital disorders can be avoided or at least minimised with appropriate early treatment. Immediate treatment enables the affected child to develop normally in the vast majority of cases.

Depending on the disease in question, treatment may consist of a special diet, the administration of certain medications or counselling and guidance for parents on preventive measures. Specialists are available for counselling and support in the event of the suspicion of or in the case of the actual illness itself.

Since the Genetic Diagnostics Act came into force in 2010, the Genetic Diagnostics Commission (GEKO) at the Robert Koch Institute has been evaluating new screening tests for genetic diseases. The GEKO has endorsed the introduction of screening for tyrosinaemia type I and severe combined immunodeficiency (SCID), sickle cell disease and spinal muscular atrophy (SMA).

Target diseases

Adrenal hyperplasia

Hormonal disorder due to a defect in the adrenal cortex: Masculinisation in girls, fatal outcome possible in salt wasting crises. Treatment with hormones, good prognosis (frequency: approx. 1/15,000 newborns).

Maple syrup urine disease

Defect in the breakdown of amino acids: Mental disability, coma, fatal outcome possible. Treatment with a special diet, usually good prognosis (incidence: approx. 1/180,000 newborns).

Biotinidase deficiency

Defect in the metabolism of the vitamin biotin: Skin changes, metabolic crises, mental disability, fatal outcome possible. Treatment by biotin administration, very good prognosis (frequency: approx. 1/28,000 newborns).

Carnitine inborn errors of metabolism

Defect in the metabolism of fatty acids: Metabolic crises, coma, fatal outcome possible. Treatment with a special diet, very good prognosis (incidence: approx. 1/600,000 newborns).

Galactosaemia

Defect in the metabolism of a lactose component (galactose): Clouding of the lens of the eye, physical and mental disability, liver failure, fatal outcome possible. Treatment with a special diet, usually good prognosis (incidence: approx. 1/77,000 newborns).

Glutaric aciduria type 1

Defect in the breakdown of amino acids: Sudden metabolic crisis with permanent movement disorder. Treatment with a special diet, usually good prognosis (incidence: approx. 1/140,000 newborns).

Hypothyroidism

Congenital hypothyroidism: Severe disturbance of mental and physical development. Treatment by hormone administration, very good prognosis (frequency: approx. 1/3,000 newborns).

Isovaleric acidemia

Defect in the breakdown of amino acids: Mental disability, coma, fatal outcome possible. Treatment with a special diet, very good prognosis (incidence: approx. 1/90,000 newborns).

LCHAD, VLCAD deficiency

Defect in the metabolism of long-chain fatty acids: Metabolic crises, coma, muscle and heart muscle weakness, fatal outcome possible. Treatment with a special diet, avoidance of periods of starvation, usually good prognosis (incidence: approx. 1/80,000 newborns).

MCAD deficiency

Defect in energy production from fatty acids: Hypoglycaemia, coma, fatal outcome possible. Treatment by avoiding periods of starvation, very good prognosis (incidence: approx. 1/10,000 newborns).

Phenylketonuria

Defect in the metabolism of the amino acid phenylalanine: Mental disability if left untreated. Successful treatment with a special diet, very good prognosis (incidence: approx. 1/10,000 newborns).

Tyrosinemia type I

Disorder in the breakdown of the amino acid tyrosine, which without treatment from the first days of life can lead to severe liver dysfunction with jaundice and a tendency to haemorrhage, impaired kidney function and neurological crises. Treatment with a drug (nitisinone) and a low-protein diet, good prognosis (incidence: approx. 1/135,000 newborns).

Severe combined immunodeficiency (SCID)

Complete lack of immune defence: high susceptibility to infection coupled with complications due to infection even in infancy. Treatment by strict hygienic precautions. Therapy with bone marrow or stem cell transplantation, enzyme replacement therapy. Avoid breastfeeding, live vaccinations or transfusion of untreated blood products. If left untreated, most affected children die within 1 to 2 years (frequency: approx. 1/32,500 newborns)

Sickle cell disease (SCD)

Deformation of the red blood cells (sickle cells) leads to anaemia, increased viscosity of the blood and a poorer oxygen supply to the organs. Long-term organ damage. Acute complications include cerebral infarction, kidney failure, spleen infarction, septicaemia and anaemia. The treatment approach includes education and guidance on behavioural measures, infection prophylaxis (e.g. vaccinations), administration of hydroxycarbamide, transfusions if necessary and, if necessary, stem cell transplantation as a further treatment approach. If left untreated, it can occur from around the age of 3. Symptoms may occur as early as the third month of life (frequency: approx. 1/3,950 newborns).

Spinal muscular atrophy (SMA)

Deficiency of a certain protein (survival motor neuron (SMN) protein) leads to increasing muscle weakness with a decline in motor development and impairment of lung function. Treatment is carried out with medication and symptomatic therapy (physiotherapy, rehabilitation, orthopaedics, psychology). The first symptoms of the disease in children with infantile SMA (the most common and most severe form) appear by the age of 6. If left untreated, these children die within 1 to 2 years (frequency: approx. 1/6,000 to 1/11,000 newborns).

Newborn screening for cystic fibrosis

At the same time as the newborn screening for congenital metabolic, endocrine, vascular, immune and neuromuscular disorders, you will be offered a screening for cystic fibrosis for your child. The aim of this examination is to diagnose cystic fibrosis at an early stage so that treatment can be started as soon as possible, thereby improving the quality of life and life expectancy of children with cystic fibrosis. Newborn screening for cystic fibrosis is subject to the special regulations of the German Genetic Diagnostics Act. The following information is intended to help you prepare for a consultation with your doctor.

What is cystic fibrosis?

Cystic fibrosis is a hereditary disease that affects approximately 1 in 3,300 children. A gene mutation in the so-called *CFTR* gene leads to a disruption of salt exchange in glandular cells. This is the cause of the formation of viscous mucus in the airways and other organs, which become permanently inflamed. The severity of the symptoms can vary due to different genetic mutations. The function of the pancreas is often restricted. As a result, affected children are often underweight and grow poorly. In severe cases, lung function can be significantly impaired as a result of repeated cases of severe pneumonia.

How can cystic fibrosis be treated?

The symptoms of cystic fibrosis can be improved or alleviated by various therapeutic approaches, with the result that the life expectancy of cystic fibrosis patients has risen continuously. The treatment of cystic fibrosis consists of inhalations and physiotherapy, a needs-based diet and medication. It also makes sense to have regular check-ups at specialised cystic fibrosis clinics so that early changes can be treated in good time.

Why is screening for cystic fibrosis useful?

Newborn screening for cystic fibrosis enables early diagnosis. Starting treatment early can improve the physical development of affected children. This also increases the chance of a longer and healthier life.

How is newborn screening for cystic fibrosis carried out?

Screening for cystic fibrosis does not usually require an additional blood sample. Screening for cystic fibrosis takes place at the same time and from the same blood sample that is taken from your child for newborn screening for congenital diseases of the metabolic, endocrine, vascular, immune and neuromuscular systems. To do this, a few drops of blood are dripped onto a filter paper card and sent to a screening laboratory.

There, the enzyme immunoreactive trypsin (IRT) is determined first. If the value is elevated, a second test for pancreatitis-associated protein (PAP) is carried out on the same blood sample. If the second test result is also elevated, a DNA test (genetic test) is carried out on the same sample to look for the most common genetic changes that occur in cystic fibrosis. If one or two gene mutations are found, the screening result needs to be verified. If the first test (IRT) is already very abnormal, the screening results alone need to be verified and the other tests are then no longer carried out. The combination of test steps maximises the accuracy and reliability of the results. Nevertheless, it can very rarely happen that a child has cystic fibrosis and this is not recognised in an early diagnosis.

In accordance with the legal requirements of the German Genetic Diagnostics Act, a doctor must be consulted before newborn screening for cystic fibrosis is carried out. If the birth is conducted by a midwife, the screening for cystic fibrosis in your child can be carried out by a doctor up to the age of 4 weeks (for example at the U2). A further blood sample must then be taken for this purpose. In contrast to screening for cystic fibrosis, newborn screening for congenital diseases of the metabolic, endocrine, vascular, immune and neuromuscular

systems should ideally be carried out within the 37th to 72nd hour of life, since, unlike in cystic fibrosis screening, immediate initiation of therapy is crucial for the majority of the diseases tested.

To ensure that your child can always have this important examination, we have also drawn up a contingency concept as a quality assurance measure. In the very unlikely event that our own laboratory is unable to process your child's blood samples due to malfunctions, our cooperation partners (other screening laboratories) can take over the test.

How will you be informed about the screening results and what happens afterwards?

The laboratory will let you know within 14 days whether the results are normal or need to be checked. You will only be informed of a normal result if you expressly request it. If you have a result that needs to be verified, a doctor will tell you which specialist clinic you can take your child to for a check-up. This is necessary in order to verify the screening findings and possibly initiate therapeutic measures. You will receive comprehensive information about the type of examination and treatment options at this center. A result that needs to be verified does not yet mean that your child has cystic fibrosis. Only one in five children with a result that needs to be verified actually has cystic fibrosis. However, there is an increased probability of what is known as a carrier. The carriers are healthy, but can pass this trait on to their offspring. In any case, you will be offered genetic counselling so that you can obtain detailed information about the significance of this result.

At the specialist clinic for cystic fibrosis, a confirmatory examination, usually a sweat test, is carried out first and everything else is discussed with you. This sweat test is harmless and painless and does not put any strain on your child. You will be informed of the result the day after the examination at the latest. Further investigations may be necessary.

You decide for your child

Participation in cystic fibrosis screening is voluntary. The costs of the examination are covered by statutory health insurance. The results of the examination are subject to medical confidentiality and may not be passed on to third parties without your consent. You have the right to withdraw your consent to cystic fibrosis screening at any time. A decision in favour of or against screening for cystic fibrosis should be made on the basis of sound information. You always have the opportunity to discuss your questions with doctors. Your consent covers newborn screening for cystic fibrosis and the processing of the personal data required for this purpose.

This genetic screening test for cystic fibrosis is endorsed by the Genetic Diagnostics Commission at the Robert Koch Institute.

Information about the processing of your personal data

Information about the processing of your personal data can be found at uke.de/data-protection/patients or by using the QR code here. At your request, we can also provide the data protection information in printed form.

Right of cancellation

Participation in newborn screening is voluntary. You can revoke your consent at any time with effect for the future.

Residual blood sample

If you give your consent, the residual blood sample will be stored for a period of 12 months after receipt by the laboratory for quality assurance purposes and for the purpose of verifying the results at a later date. Otherwise, your child's blood sample will be stored for 3 months after all tests have been completed in accordance with legal requirements and then destroyed.

Declaration of consent for newborn screening

If you would like to have your child screened for congenital metabolic, endocrine, vascular, immune and neuromuscular disorders, as well as cystic fibrosis, please sign on this page.

Name of the child: _____

Date of birth: _____ (or adhesive label)

- ○ I consent to the newborn screening for congenital metabolic, endocrine, vascular, immune and neuromuscular disorders, as well as cystic fibrosis, in my child and to the processing of the information and data provided for this purpose by the UKE and by cooperation partners of the UKE as part of the contingency concept. If abnormalities are detected, I hereby consent to the laboratory doctor forwarding the findings to me/us. In the event of abnormal findings, I also agree to the forwarding of the findings to a specialist clinic to be selected by me/us, where the diagnostic examination can be carried out, and to the forwarding of my/our contact details to this clinic for the purpose of contacting me/us.

  For further clarification of a finding (e.g. if a blood sample is taken too early, suspicion of the presence of a target disease) and if a second screening card required for this is not received, I agree to the laboratory doctor contacting me/us to send a reminder. The specialist clinic providing further treatment may inform the screening laboratory if I/we have not made or attended an appointment for a diagnostic examination so that they can contact me/us.

- ○ The specialist clinic providing further care may transmit the result of the diagnostic examination to the screening laboratory for quality assurance purposes (data transmission of diagnostic examination). *(In case of refusal, this must be noted on the screening card).*

- ○ I **agree that** residual blood samples will be stored for a period of 12 months after receipt by the laboratory for quality assurance purposes and for the purpose of subsequent verification of the results. If the sample is rejected, it will be destroyed 3 months after all tests have been completed. (This must also be noted on the screening card).

- ○ I **agree** that the anonymised sample may be used for the further development of medical diagnostics, for quality assurance or for similar scientific purposes. *(In case of refusal, this must be noted on the screening card).*

- ○ I refuse to have my child undergo the newborn screening. I was made aware of the possible negative consequences for my child (undetected illness that could lead to disability and death).

_____

Date, name in block letters, signature of at least one person with parental authority

_____

Date, name in block letters, signature of the doctor providing information in accordance with § 8(1) of the German Genetic Diagnostics Act

If you only wish to have individual examinations carried out on your child, please complete and sign the declaration on the reverse side.

This declaration of consent remains with the sender of the sample.
Consent to the newborn screening or rejection of individual parts of the screening programme must be noted on the filter paper card for newborn screening in the spaces provided.

Differentiated declaration of consent for the newborn screening procedures

If you agree to all offered examinations, please sign the declaration of consent on the reverse side.
If you do not wish to consent to the screening programme in full, please complete this page.

Name of the child: _____

Date of birth: _____ (or adhesive label)

I was informed about newborn screening for congenital metabolic, endocrine, vascular, immune and neuromuscular disorders, as well as newborn screening for cystic fibrosis. I was made aware of the possible negative consequences for my child if I refuse individual parts of the newborn screening programme.

Differentiated declaration of consent

(Please tick the section(s) of the newborn screening programme to which you agree)

- ○ Newborn screening for congenital metabolic, endocrine, vascular, immune and neuromuscular disorders (Pages 3-5)
- ○ Newborn screening for cystic fibrosis (Pages 6-7)

  I consent to the newborn screening of my child and to the processing of the information and data provided for this purpose by the UKE and by cooperation partners of the UKE as part of the contingency concept. If abnormalities are detected, I hereby consent to the laboratory doctor forwarding the findings. In the event of abnormal findings, I also agree to the forwarding of the findings to a specialist clinic to be selected by me/us, where the diagnostic examination can be carried out, and to the forwarding of my/our contact details to this clinic for the purpose of contacting me/us.
  For further clarification of a finding (e.g. if a blood sample is taken too early, suspicion of the presence of a target disease) and if a second screening card required for this is not received, I agree to the laboratory doctor contacting me/us to send a reminder. The specialist clinic providing further treatment may inform the screening laboratory if I/we have not made or attended an appointment for a diagnostic examination so that they can contact me/us.
- ○ The specialist clinic providing further care may transmit the result of the diagnostic examination to the screening laboratory for quality assurance purposes (data transmission of diagnostic examination). (In case of refusal, this must be noted on the screening card).
- ○ I **agree that** residual blood samples will be stored for a period of 12 months after receipt by the laboratory for quality assurance purposes and for the purpose of subsequent verification of the results. If the sample is rejected, it will be destroyed 3 months after all tests have been completed. (This must also be noted on the screening card).
- ○ I **agree** that the anonymised sample may be used for the further development of medical diagnostics, for quality assurance or for similar scientific purposes. (In case of refusal, this must be noted on the screening card).

_____

Date, name in block letters, signature of at least one person with parental authority

_____

Date, name in block letters, signature of the doctor providing information in accordance with § 8(1) of the German Genetic Diagnostics Act

This declaration of consent remains with the sender of the sample. Consent to the newborn screening or rejection of individual parts of the screening programme must be noted on the filter paper card for newborn screening in the spaces provided.

Legal notice: University Medical Center Hamburg-Eppendorf (UKE), Martinistraße 52, 20246 Hamburg | Design: SW | Photo: EH | Current as of: April 2025