Parent Information on Newborn Screening

Screening-Labor Hannover

Early detection of congenital disorders

Dear Parents,
we wish you all the best with the birth of your child!

In the first days of life, special tests are offered to detect congenital disorders as early as possible. The Expanded Newborn Screening for congenital, mostly hereditary diseases is a meaningful examination that should be carried out within the first 36–72 hours of life.

The laboratory tests are performed in accordance with the German Genetic Diagnostics Act (GenDG) and the "Children's Guideline" of the Joint Federal Committee of Physicians and Health Insurance Funds (G-BA).

The result of a screening test is not a medical diagnosis. The results can either largely rule out the disorders that were tested for, or, in case of suspicion of a disease, indicate that further diagnostic examinations are needed. The screening only covers certain congenital disorders defined in the "Children's Guideline".

The blood sample for the preventive examination should be taken during the 2nd to 3rd day of life (36–72 hours after birth). If your child is transferred or discharged from the clinic before the 36th hour of life, the guideline requires an initial blood sample beforehand and a second timely sample later (e.g., taken by a midwife or pediatrician), because some diseases cannot yet be reliably detected in the very first hours of life.

What conditions are being screened for?

- **Adrenogenital syndrome (AGS)**
  Hormonal disorder of the adrenal cortex; life-threatening adrenal crises possible
- **Maple syrup urine disease (MSUD)**
  Disorder of amino acid breakdown; can be life-threatening
- **Biotinidase deficiency**
  Defect in the metabolism of vitamin biotin; may lead to intellectual disability or be life-threatening
- **Carnitine cycle defects**
  Fatty acid metabolism disorder; metabolic crises, coma, potentially fatal
- **Galactosemia**
  Defect in milk sugar metabolism; can cause blindness, physical and mental disability, possibly fatal
- **Glutaric aciduria type I (GA I)**
  Defect in amino acid breakdown; permanent movement disorders, sudden metabolic crises
- **Hypothyroidism**
  Congenital thyroid insufficiency; severe impairment of mental and physical development
- **Isovaleric acidemia (IVA)**
  Defect in amino acid breakdown; intellectual disability, coma
- **LCHAD/VLCAD deficiency**
  Long-chain fatty acid metabolism disorder; metabolic crises, coma, muscle and heart weakness, possibly fatal
- **MCAD deficiency**
  Defect in energy production from fatty acids; metabolic crises, coma, possibly fatal
- **Phenylketonuria (PKU/HPA)**
  Amino acid metabolism disorder (phenylalanine); spasticity, seizures, intellectual disability
- **Tyrosinemia type I (hypertyrosinemia)**
  Disorder of tyrosine metabolism; liver dysfunction, liver cancer, jaundice, bleeding, anemia, potentially severe or fatal course
- **Severe combined immunodeficiency (SCID)**
  Complete absence of immune defense; in infancy, high susceptibility to infections and complications
- **Spinal muscular atrophy (SMA)**
  Genetic defect; increasing muscle weakness, declining motor skills, limited lung function
- **Sickle cell disease (SCD)**
  Disorder of red blood cells; anemia, oxygen deficiency, blood vessel blockage, pain, infections, organ damage
- **Vitamin B12 deficiency**
  Developmental disorders and anemia, also possible due to maternal vitamin B12 deficiency
  Treatment: low-protein diet, vitamin B12
- **Homocystinuria**
  Developmental disorders, seizures, vision and joint problems
  Treatment: medication, diet
- **Propionic acidemia and methylmalonic aciduria**
  Poor feeding, vomiting, heart disease, kidney failure, coma
  Treatment: medication, diet

Approximately 1 in 1,000 children is affected by one of these diseases. Affected children may show no symptoms shortly after birth. Early treatment can prevent severe consequences of these conditions.

What happens if the screening suggests a possible disease?

First, your child will need a detailed examination by a pediatrician or in a specialized children's hospital. Additional blood or urine tests are often required.

All listed metabolic disorders, endocrine disorders, and immune defects are congenital and therefore cannot always be cured. However, early treatment can prevent or reduce the effects of these conditions. Treatment may involve a special diet and/or taking certain medications.

Who receives the laboratory results?

The analysis results are strictly **confidential**. They are protected by **medical confidentiality** and may not be shared with third parties without your consent. The screening laboratory sends the results to the submitting institution (e.g., maternity clinic, children's hospital, or doctor's office). With your permission, your pediatrician may also request a copy of the results.

What happens to the remaining blood sample?

The filter paper containing your child's blood drops is destroyed after the examinations – and any necessary follow-up tests – are completed.

Costs of the examination

Newborn screening examinations are covered by statutory health insurance. Hospital patients with optional services ("chief physician treatment"), private outpatients, and self-paying patients receive an invoice for the individual services according to the German Medical Fee Schedule (GOÄ). The costs are usually at least partially reimbursed by insurance providers and/or public aid offices, depending on the insured tariff.

To carry out the newborn screening, the cystic fibrosis screening, and the screening for other target conditions (study) for:

Name: _____

Date of birth: _____

(Please cross out anything that does not apply)

I have been informed about the examinations and had enough time to think about them.

I have read the parent information on the newborn screening, the cystic fibrosis screening, and the study procedures (text beside this form and on the back). I had the opportunity to ask questions about all examinations and procedures described there.

I have received a **copy of the parent information**. I understand that I can withdraw my consent to the examinations in writing at any time. In that case, the examinations will not be carried out or will be stopped. Any results that have already been produced and sent will be deleted after billing once a written withdrawal is received.

I agree that the laboratory may contact the parents to communicate the results. The results may also be shared with a doctor and/or a specialized medical center, which may contact the parents directly if needed. I consent to the transfer of the personal data required for this. The laboratory may also contact the parents to send reminders.

I agree that if an abnormal or positive result is found, the results of further examinations carried out by other medical facilities may be sent to the screening laboratory.

I am aware that if the newborn screening is declined, an existing health condition may only be detected and treated at a later time.

_____
Date

_____
Signature of parent/legal guardian

Medically informed by:

_____

Information in other languages can be downloaded here:

For the patient file. Provide a signed copy to the legal guardians.

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Parent Information on Cystic Fibrosis Screening

Screening-Labor Hannover

Early detection of cystic fibrosis (CF)

What is cystic fibrosis?

Cystic fibrosis is an inherited disease that affects about 1 in 3,300 children. A change in a specific gene causes problems with salt transport in certain cells. As a result, thick, sticky mucus builds up in the airways and other organs, leading to ongoing inflammation. The pancreas is often affected as well, which can make it harder for children to gain weight. In severe cases, repeated lung infections can cause serious and lasting damage to lung function.

How can cystic fibrosis be treated?

There is currently no cure for cystic fibrosis. However, different treatments (such as inhalation therapy, physiotherapy, a high-calorie diet, and medications) can help improve symptoms. Regular care in a specialized cystic fibrosis center is also recommended so that any health problems can be detected and treated early.

Why is screening for cystic fibrosis useful?

If the disease is diagnosed early and treatment begins soon after birth, the child's physical development can improve significantly.

What is special about cystic fibrosis screening?

In about 1 out of every 500 tests, a molecular genetic analysis is also carried out. Because this is required by law, parents must receive medical counseling before the cystic fibrosis screening is done. If the birth was attended only by a midwife, the cystic fibrosis screening can still be completed later, up until the U3 check-up, by a pediatrician using the already collected blood sample. If the result is abnormal, you will be informed in the same way as for the extended newborn screening (see information on the other side).

Cost of the test

The costs are the same as for the extended newborn screening (see information on the other side).

Information in other languages can be downloaded here:

Parent Information on Additional Target Diseases (Study)

Taking part in this study is voluntary. The parameters are measured together with the routine newborn screening. You may withdraw your consent at any time, without giving any reasons and without any disadvantages for you or your child. Please submit your withdrawal in writing to the laboratory. By participating, you agree that the collected data may be used for scientific research.

Conditions included in the study:

Carnitine transporter deficiency

A problem with absorbing carnitine, leading to slowly progressing heart and muscle weakness, low blood sugar, and liver failure. Treatment: carnitine supplements

Citrullinemia

A protein breakdown disorder causing seizures, poor feeding, and life-threatening crises (coma). Treatment: low-protein diet, medication

3-Hydroxy-3-Methylglutaryl-CoA Lyase deficiency

A disorder that prevents the body from making ketones for energy. Symptoms include poor feeding, vomiting, low blood sugar, blood acidosis, reduced awareness, and life-threatening coma. Treatment: avoiding fasting and catabolic stress

Multiple acyl-CoA dehydrogenase deficiency (Glutaric acidemia type II)

A disorder of amino acid and fatty-acid breakdown causing poor feeding, vomiting, blood acidosis, heart problems, and life-threatening coma. Treatment: diet and medication

Remethylation disorders (MTHFR, CblD, CblE, CblG)

Problems in producing the amino acid methionine. Symptoms may include neurological crises, severe intellectual disability, and blood abnormalities. Treatment: vitamins and medication

Hypermethioninemia

A disorder of methionine metabolism, neurological symptoms, intellectual disability.
Treatment: low-methionine diet

Argininosuccinate lyase deficiency

A rare amino-acid metabolism disorder leading to high ammonia and low arginine. Symptoms include low muscle tone, growth problems, vomiting, behavior changes, and life-threatening coma. Treatment: low-protein diet, arginine supplements, medication

Aromatic L-Amino Acid Decarboxylase (AADC) deficiency

An enzyme defect causing low dopamine, serotonin, adrenaline, and noradrenaline. This leads to movement disorders, low muscle tone in the body, dystonia, and global developmental delay

Arginase deficiency

A congenital enzyme deficiency that can cause spasticity, epilepsy, intellectual disability, and high ammonia levels. Treatment: low-arginine diet, enzyme replacement therapy

X-linked adrenoleukodystrophy

An inherited disease on the X chromosome; boys are mainly affected. Symptoms include neurological problems, spasms, difficulty walking, hearing and vision problems, liver damage, and kidney failure. Treatment: gene therapy, stem cell transplantation

Serine deficiency syndrome

A congenital metabolic disorder causing microcephaly, seizures, and delayed development. Treatment: high-dose serine and glycine supplementation

Guanidinoacetate methyltransferase (GAMT) deficiency + Arginine: Glycine amidinotransferase (AGAT) deficiency

Rare disorders of creatine metabolism leading to developmental and speech delays, weak muscles, and seizures. Treatment: creatine, ornithine, low-arginine diet

Adenosine deaminase (ADA) deficiency

This enzyme defect can cause a severe immune deficiency with recurrent serious infections and developmental problems. Treatment: enzyme replacement, gene therapy, stem cell transplantation

2,4-Dienoyl-CoA reductase (DECR) deficiency

A rare congenital defect of the NADPH redox system. Symptoms include failure to thrive, encephalopathy, epilepsy, ataxia, and kidney-related lactic acidosis. Treatment: low-lysine diet, vitamins, carnitine

Cost of the test

The tests are free of charge as part of the study.

Results will be sent either to the healthcare provider who submitted the sample or to a specialized center.

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