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DAkkS Deutsche Akkreditierungsstelle D-ML-19642-02-00

# Information for Parents on Newborn Screening

## Early detection of congenital health problems

Für die Patientenakte. Kopie mit Unterschriften an Personensorgeberechtigte aushändigen

**Dear Parents,**
We wish you and your little one all the best on the birth of your child!

In the first days of life, specific examinations are offered to detect congenital diseases at an early stage. *Expanded newborn screening* for congenital, mostly hereditary diseases is a very useful examination that should be performed within the first 36 – 72 hours of life.

Laboratory tests are carried out in accordance with the provisions of the Genetic Diagnostics Act (GenDG) and the "Children's Directive" of the Federal Joint Committee of Physicians and Health Insurance Funds (G-BA).

The results of a screening test do not yet constitute a medical diagnosis. The test results can either largely rule out the disorders in question, or, if a disease is suspected, they can indicate that further diagnostic testing is required. Only certain congenital diseases specified in the "Children's Directive" are captured in the screening.

The blood for the screening test is usually taken during the 2nd – 3rd day of life (36th – 72nd hour after birth). If your child is transferred or discharged from the clinic before 36 hours, the guideline requires an initial and later a prompt additional blood sample (taken, for example, by the midwife or the paediatrician), as some diseases cannot yet reliably uncovered in the first few hours of life.

**What are we looking for?**

- *Adrenogenital Syndrome (AGS)*
  A hormonal disorder of the adrenal cortex, may be fatal due to salt loss crisis
- *Maple Syrup Disease (MSUD)*
  Defect in amino acid breakdown, may be fatal
- *Biotinidase deficiency*
  Metabolic defect of the vitamin biotin. Mental handicap, may be fatal
- *Carnitine cycle defects*
  Metabolic fatty acid defect. Metabolic crises, coma, may be fatal
- *Galactosaemia*
  Lactose metabolism defect. Blindness, physical and mental handicap, fatal outcome possible
- *Glutaric aciduria Type I (GA I)*
  Amino acid breakdown defect. Persistent movement disorders, sudden metabolic crises
- *Hypothyroidism*
  Congenital underactive thyroid gland. Severe mental and physical developmental disorders
- *Isovaleric acidaemia (IVA)*
  Amino acid breakdown defect. Metal handicap, coma
- *LCHAD, VLCAD deficiency*
  Metabolic long-chain fatty acid defect. Metabolic crises, coma, muscle and heart muscle weakness, may be fatal
- *MCAD deficiency*
  Defective energy production from fatty acids. Metabolic crises, coma, may be fatal
- *Phenylketonuria (PKU/HPA)*
  Metabolic defect of the amino acid phenylalanine. Spasticity, seizures, intellectual disability
- *Tyrosinemia Type I (Hypertyrosinemia)*
  Tyrosine metabolism breakdown disturbance. Impaired liver function, liver cancer, icterus (jaundice), bleeding, anaemia, may be severe or fatal
- *Severe Combined Immunodeficiency (SCID)*
  Complete lack of defensive immune response: high susceptibility to infection paired with infection-related complications even in infancy
- *Spinal muscular atrophy (SMA)*
  Genetic defect. Increasing muscle weakness, decrease in motor ability, impaired lung function
- *Sickle cell diseases (HbS)*
  Disease of the red blood cells. Anaemia, oxygen deficiency, blockage of the blood vessels, pain, infections, organ damage

Approximately 1 in 1,000 children suffer from one of these diseases. The children may not show any signs of illness right after birth. Timely treatment can save them from serious effects of their disease.

**What happens if the screening test indicates an illness?**
First of all, a detailed examination of the child at the paediatrician's office or in a specialised children's hospital is required. Blood or urine are often also examined there.

All of the mentioned metabolic defects, endocrine disorders and immunodeficiencies are congenital, and sometimes therefore cannot be cured. However, with early treatment the effects of these congenital disorders can be avoided or at least reduced. The treatment consists of a special diet and/or taking certain medications, for example.

**Who will receive the results of the laboratory tests?**
The analysis results are strictly **confidential**. The results of the test are subject to **medical confidentiality** and may not be shared with third parties without your consent. The screening laboratory transmits the findings to the sender (for example, a maternity clinic, children's hospital or medical practice). Your paediatrician can also request a copy of the results if you consent to this.

**What happens to leftover blood from the sample?**
The filter paper with the drops of your child's blood will be destroyed after the tests, or after any necessary follow-up tests are performed.

**Cost of the test**
Newborn screening tests are services provided by statutory health insurance. Hospital patients entitled to optional services ("treatment by a senior consultant"), private outpatients and self-payers receive an invoice for the individual items according to the official schedule of doctor's fees (GOÄ). The costs are generally at least partially reimbursed by the insurance companies and/or the aid agency, in accordance with the insured fee schedules.

For performing newborn screening, screening for cystic fibrosis and other target diseases (study)
(Please cross out what does not apply) for:

Name: _____

Date of birth: _____

I have been informed about the test and have had sufficient time to think it over.

I have read the "Information for Parents on Newborn and Cystic Fibrosis Screening" (text adjacent and on back). I had the opportunity to ask questions about all of the tests and procedures described there.
I have received a **copy of the Information for Parents.** I can withdraw permission for the **tests at any time**. In that case, the tests will not be performed or will be cancelled.

I consent to have a specialised centre contact me directly if an illness is suspected. I consent to the transfer of personal data required for this purpose. I consent to the results of further tests being sent to the screening laboratory if the results are abnormal.

**I am aware that if I reject the newborn screening, a disease that may be present will only be able to be identified and treated at a later point in time.**

_____
**Date, signature of the person having legal custody**

**Medical information briefing conducted by:**

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# Information for Parents on Cystic Fibrosis Screening

Early detection of mucoviscidosis / cystic fibrosis (CF)

**Dear Parents,**
At the same time as the expanded newborn screening, you will be offered an early diagnostic test for cystic fibrosis for your child.

**What is cystic fibrosis?**
Mucoviscidosis (also called cystic fibrosis (CF)) is an inherited disease that affects approximately 1 in 3,300 children. A genetic change leads to disruption of the salt exchange in gland cells. This in turn causes the formation of viscous mucus in the airways and other organs, which become permanently inflamed as a result. Pancreatic function is often impaired. As a result, affected children are often underweight and have poor growth. In severe cases, lung function can be significantly impaired due to repeated severe pneumonia.

**How can you treat cystic fibrosis?**
There is currently no cure for cystic fibrosis. However, as the symptoms of the disease can be improved or alleviated through various therapeutic approaches, the life expectancy of cystic fibrosis patients has continuously increased. Cystic fibrosis treatment consists of inhalations and physiotherapy, an especially high-calorie diet, and medication. In addition, it makes sense to seek out care in specialized cystic fibrosis facilities, so that treat pathological changes can be treated in good time.

**Why is health screening for cystic fibrosis a good idea?**
Early diagnosis and prompt treatment can improve the physical development of affected children. This also increases the chance of living a longer and healthier life.

**How is cystic fibrosis screening performed?**
Generally speaking, no additional blood draws are needed for cystic fibrosis screening. A molecular genetic analysis is also performed in about one in 1000 tests.
In accordance with legal requirements, a doctor must explain the process to you before cystic fibrosis screening can be performed. If the birth was attended by a midwife, the screening for cystic fibrosis can still be done later by a paediatrician, up to the regular preventive medical examination that children usually get at 3 weeks of age.

**How will you be informed of the screening test results and what will happen afterwards?**
The laboratory will inform the person who sent in the blood sample of the results within 14 days. If the result is normal, you will only be informed by the sender if you have expressly requested this. If the result is abnormal, you will be informed directly by a specialised centre or by the sender, and the next steps will be discussed with you.

**Cost of the test**
As with the expanded newborn screening, see overleaf.

# Information for Parents on Other Target Diseases (Study)

Taking part in this study is voluntary. You can withdraw your consent at any time without giving reasons, and without this causing any disadvantage for you or your child. Please inform the laboratory of your withdrawal in writing. By taking part, you agree that the obtained data may be evaluated for scientific purposes.

**Carnitine transporter defect**
Disorders of carnitine uptake with slowly progressing heart muscle and general muscle weakness, hypoglycaemia, liver failure. Treatment: Administration of Carnitine

**Citrullinemia**
Defect in protein breakdown, seizures, poor fluid intake, life-threatening crises (coma). Treatment: low-protein diet, medication

**3-Hydroxy-3-methylglutaryl-CoA-Lyase-Deficiency**
Disorders in ketone formation as a source of energy, poor fluid intake, vomiting, hypoglycaemia, acidification of the blood, clouded consciousness, life-threatening coma. Treatment: Avoidance of catabolic (starvation) phases

**Propionic acidaemia**
Disruption of the breakdown of amino acids leads to poor fluid intake, vomiting, over-acidification of the blood, seizures, and life-threatening coma. Treatment: low-protein diet, medication

**Multiple acyl-CoA dehydrogenase deficiency (glutaric aciduria type II)**
Amino acid and fatty acid breakdown disorders, poor fluid intake, vomiting, over-acidification of the blood, heart function disorders, life-threatening coma. Treatment: Diet, medication

**Methylmalonic aciduria and disorders of Vitamin B12 metabolism**
Congenital disorder of protein breakdown or due to a vitamin B12 deficiency in the mother (vitamin B12 absorption disorder, vegetarian/vegan diet), rapidly worsening fluid intake, vomiting, over-acidification of the blood, seizures and life-threatening coma. Treatment: low-protein diet, vitamin B12

**Remethylation disorders (MTHFR, CblD, CblE, CblG)**
Defects in the formation of the amino acid methionine, neurological crises, some of which occur later, severe intellectual disability, blood formation disorders. Treatment: Vitamins, medication

**Hypermethioninaemia, Homocystinuria**
Protein metabolism disruptions, affecting various tissues, eyes, CNS, skeletal and connective tissue, vessels, sometimes with life-threatening occlusions of the blood vessels (thrombi). Treatment: Diet, medication, vitamins

**Argininosuccinate-lyase deficiency**
Rare disruption of amino acid metabolism, leads to an increase in ammonia and arginine deficiency, symptoms such as low muscle tone, growth stagnation, vomiting, behavioural disorders and life-threatening coma. Treatment: low-protein diet, arginine, medication

**Aromatic L-amino acid decarboxylase (AADC)**
Enzyme defect, dopamine, serotonin, adrenaline and noradrenaline deficiencies lead to motor movement disorders, low muscle tone, dystonia and global developmental delays

**Cost of the test**
Free of charge as part of the study