

How do we carry out the mucoviscidosis screening?

Normally, no additional blood taking is necessary for this examination. The mucoviscidosis screening takes place at the same time and from the same blood sample which was taken from your baby for the extended newborn screening.

The laboratory first determines the so called enzyme immunoreactive trypsin (IRT). In case of a heightened factor, a second test for the pancreatitis associated protein (PAP) takes places from the same blood sample. If the second factor is also heightened, a DNA test (genetic examination) takes place with the aim to detect one of the 31 most frequently appearing genetic alterations which appear at presence of mucoviscidosis. If the first test is very heightened or at least one genetic alteration was detected, the mucoviscidosis screening is declared to be suspicious.

A combination of the different test steps guarantees a maximum precision and reliability of the test results. However, in very rare cases it is also possible that a child falls ill with mucoviscidosis and did not have a suspicious test result during this early detection examination.

What happens at presence of a suspicious screening result?

In case of a result which has to be controlled again, you and your baby are referred to a specialised mucoviscidosis centre where a sweat test takes place as confirmation examination. Please consider that a result which needs a follow up examination does not mean that your newborn has mucoviscidosis. Only one out of five children with a suspicious result and who needed a follow up examination actually has mucoviscidosis. Furthermore, the sweat test is harmless and painless and does not stress your baby.

You decide for your baby!

The participation in the mass screening is voluntary. Also the costs for this examination are taken if you are insured in a statutory health insurance. The results of this examination underlie the medical confidentiality and may not be transmitted to other persons without your agreement. The screening laboratory transmits the results directly to the responsible person who contacts you in case of a positive result. You have the right to revoke your agreement to participate in the mucoviscidosis mass screening at any time.



Notice:

Also an early treatment cannot prevent consequences of the mentioned diseases completely in all cases. However, an immediate treatment makes a normal development possible for concerned children in the most cases.

Important FRAMEWORK conditions

- The participation in the newborn screening examinations is voluntary. Your agreement to participate includes a test for the here mentioned diseases.
- Data protection: The filter paper card and the part with personal data of you and your baby are separated after the analyses have been finished. The examined dried blood sample is destroyed after finishing of all analyses (at the latest after 3 months).
- The results of the examinations underlie the medical confidentiality and may not be transmitted to other persons without your agreement.
- The costs for this examination are overtaken if you are insured in a statutory health insurance. If you are privately insured the costs are normally also overtaken, however, for further details please contact your insurance company.

In case you have any further questions about the topic of the newborn screening, please contact your maternity clinic or your paediatricist.

Our team of the newborn screening laboratory wishes you and your baby all the best for your future.

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Which diseases are included in the newborn screening?

Congenital adrenal hyperplasia

Endocrine disorder due to defect of the adrenal cortex; androgenisation at girls, possible lethal outcome at salt wasting crisis. Treatment by hormone intake. (Frequency: approx. 1/10,000 newborns)

Maple syrup urine disease

Degradation defect of amino acids; mentally handicapped, coma, possible lethal outcome. Treatment by special diet. (Frequency: approx. 1/200,000 newborns)

Biotinidase deficiency

Metabolic defect of the vitamin biotin; Skin alteration, metabolic crisis, mentally handicapped. Treatment by biotin intake. (Frequency: 1/80,000 newborns)

Carnitine metabolic disorder

Metabolic disorder of fatty acids; metabolic crisis, coma, possible lethal outcome. Treatment by special diet. (Frequency: approx. 1/100,000 newborns)

Galactosemia

Impaired metabolism of lactose sugars; Blindness, physical and intellectual disability, liver failure, possibly resulting in death. Treatment through a special diet (Frequency: approx. 1/40,000 newborns)

Glutaric acidemia type 1

Degradation defect of amino acids; permanent movement disorder, sudden metabolic crisis. Treatment by special diet. (Frequency: approx. 1/80,000 newborns)

Hypothyreosis

Congenital hypothyreosis; Severe mentally and physical development disorder. Treatment by hormone intake. (Frequency: approx. 1/4,000 newborns)

Isovaleric acidemia:

Degradation defect of amino acids; mentally handicapped, coma. Treatment by special diet and intake of amino acids. (Frequency: approx. 1/50,000 newborns)

LCHAD/VLCAD

Metabolic defect of long-chain fatty acids; metabolic crisis, coma, myasthenia, myocardial insufficiency, possible lethal outcome. Treatment by special diet, prevention of starvation periods. (Frequency: approx. 1/80,000 newborns)

MCAD-deficiency

Energy production defect from fatty acids; metabolic crisis, coma, possible lethal outcome. Treatment by intake of carnitine. Prevention of starvation periods. (Frequency: approx. 1/10,000 newborns)

Phenylketonuria

Metabolic defect of the amino acid phenylalanin; seizures, spasticity, mentally handicapped. Treatment by special diet. (Frequency: approx. 1/20,000 newborns)

Mucoviscidosis (since 09/2016)

Also called cystic fibrosis. Defect of a transport protein (CFTR). Causes a salt exchange disorder in the gland cells. Viscous mucus formation in the respiratory tract and organs which become permanently inflamed. Relief and improvement of symptoms by following treatment: inhalation, physiotherapy, special diet, drugs. (Frequency: 1/3,300 newborns)

Newborn hearing screening

Hearing disorders are the most frequent sensory disorders during infancy with multiple reasons, partly appearing in combination with other diseases (cardiac, renal, and visual disorders). Early detection and therapy starting are condition for an optimal linguistic and mental development. Too late detection causes permanent damages. Therapy among others support, hearing device supply and if necessary cochlea implant. (Frequency approx. 1-3/1000 newborns)

NEWBORN SCREENING



Information for parents

For early detection of congenital disorders at newborns.

Inclusive - Screening for mucoviscidosis



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Dear parents,



you expect your baby to be born soon or it just came into the world. You deeply wish that it will grow up healthy. For this reason our handout informs you about the early detection examinations of the Newborn Screening, which takes place only a few days after birth of your baby.

Why do we carry out early detection examinations?

Most children are born healthy and also stay healthy during their life. However, some newborns are affected by rarely appearing congenital diseases which are not detectable by outer signs. Approximately, one out of 1000 newborns suffers from such a rarely appearing congenital disorder of the metabolism or of the organ function and its health is thereby in danger. An early treatment after birth can avoid consequences of congenital diseases in the most cases. Therefore, every newborn in Germany is examined already for more than 30 years. The metabolic tests are improved steadily and further treatable diseases have been included into the examinations.

When participating in this investigation program you help to ensure the healthiness of your baby. If you are insured under a statutory insurance plan, no further costs arise.

When and how do we carry out the examinations?

The screening examinations take place during the second or third day of life (36th - 72nd hour after birth), where appropriate also in connection with the second early detection examination of the newborn (so-called U2). A few drops of blood are taken from vein or heel of the newborn and dropped on the corresponding filter paper card with the purpose to be sent immediately after drying to the screening laboratory. There, the samples are examined instantly by very special, sensitive methods.

Which diseases can be detected by our Newborn Screening?

Congenital hypothyreosis, congenital adrenal hypoplasia (CAH), biotinidase deficiency, galactosemia, phenylketonuria (PKU)/hyperphenylalaninemia (HPA), maple syrup urine disease (MSUD), fatty acid metabolism defects (MCAD-, LCHAD-, VLCAD deficiency), carnitine cycle deficiencies, glutaric aciduria type I and isovaleric acidemia (IVA) (diseases exactly described in the following). A test for mucoviscidosis (cystic fibrosis) forms part of the screening since 2016. In most of the concerned families such diseases did not appear before. The newborn screening is of great importance because concerned newborns often seem to be completely healthy at birth. We want to protect them from severe diseases and their consequences such as severe disorders in mental and physical development. Some of the mentioned diseases are inherited. For this reason the newborn screening examines genetic traits and is therefore of course subjected to the regulations of the Gene Diagnostics Act.



What does the testing result tell us?

The result of a screening test is no medical diagnosis and a request for a follow up examination does not automatically mean that your baby is ill. By means of the testing result it is possible rather to exclude the concerned investigated disorders or to require another diagnostic examination upon suspicion of a disease. However, a repetition of a test can also be necessary in case the point of time of blood taking was not optimal or the amount of blood was not sufficient for all tests. We kindly ask you to react quickly when you are asked to participate in a follow up examination. It is in the interest of your baby when the situation is clarified rapidly.

Is it possible to cure these diseases?

All mentioned metabolic defects and endocrine disorders are congenital and cannot be cured. However, the consequences of these congenital disorders can be prevented or at least reduced by an early treatment. Treatment does mean either a special diet and/or the intake of certain drugs. In case of mucoviscidosis there are new, improved therapeutic approaches, so that the life expectancy of concerned patients has increased continuously. Specialists for metabolism, hormones and mucoviscidosis are at your disposal for consultation and support in case of suspicion of a disease or illness.

Who is informed about the testing result?

In every case the sender of the blood sample (maternity clinic, paediatricist) receives within a few days a written finding about the result of the hormone, metabolism and mucoviscidosis screening. The result underlies the medical confidentiality and may not be transmitted to any other person without your declaration of consent. You as parents do normally not receive the finding. In urgent cases you will be contacted immediately. Therefore, we kindly ask you to indicate your telephone number and address on the testing card, where you can be contacted during the first days after birth of your baby.

The following applies for you: No News are Good News

Early detection and treatment of concerned newborns is only possible when all parties such as parents, maternity clinic, paediatricist and screening laboratory work together without wasting of time in order to finish the testing results immediately and tests can be repeated if necessary. You are only informed about unsuspected testing results upon personal request at your maternity clinic or paediatricist.



Screening for mucoviscidosis

At the same time with an enlarged newborn screening you are offered a mass screening for mucoviscidosis after birth of your baby. Aim of this screening is an early detection of mucoviscidosis so that a therapy can start as soon as possible and the quality of life and life expectancy of children with mucoviscidosis can be improved.

What is mucoviscidosis?

Mucoviscidosis (also called cystic fibrosis) is a congenital disease which appears approximately at one out of 3300 newborns. Here, a gene alteration of the so called CFTR gene causes a salt exchange disorder in the gland cells. This causes the development of viscous mucus in the respiratory tract and other organs which therefore become chronically inflamed. The severity of symptoms can vary due to different gene alterations. Also the pancreatic function is limited. Therefore concerned children are often underweight and do not grow very good. In severe cases and in case of frequently appearing lung inflammations the pulmonary function can be significantly impaired.

Why does a mass screening for mucoviscidosis makes sense?

A mass screening for mucoviscidosis enables an early diagnosis. And an early therapy starting can improve the physical development of concerned children. Thereby the chance of a longer and more healthy life increases.

How can mucoviscidosis be treated?

At the moment there is no healing therapy for mucoviscidosis. However, the symptoms can be improved or relieved by different therapies, so that the life expectancy has increased continuously during the last decades. The treatment of mucoviscidosis consists in inhalations and physiotherapy, a diet which is extremely rich in calories and the intake of drugs. Furthermore, it is useful to attend constantly certain control examinations in specialised mucoviscidosis centres to detect and treat early changes of the disease.