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. If you are privately insured the costs are normally also overtaken, however, for further details please contact your insurance company.

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

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 paediatrist.

Information for parents

For early detection of congenital disorders at newborns.

Inclusive - Screening for mucoviscidosis

Newborn hearing screening

Hearing disorders are the most frequent sensory disorders during infancy with multiple reasons, partly appearing in combination with other diseases (cardiac, mental and visual disorders). Early detection and therapy starting are condition for permanent damages. Therapy among others supports hearing devices supply and if necessary costs of a hearing implant. (Frequency approx. 1/3,000 newborns)

Which diseases can be detected by our Newborn Screening?

Is it possible to cure these diseases?

Screening for mucoviscidosis

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 take place only a few days after birth of your baby. At the same time with an enlarged newborn screening you are offered a mass screening for mucoviscidosis after birth of your baby.

All mentioned metabolic defects and endocrine disorders are congenital and cannot be cured. However, the consequences of such diseases did not appear before. The newborn screening is of great importance because concerned newborns often seem to a disease or illness. The screening examinations take place during the second or third day of life (36th - 72nd hour after birth), where a few drops of blood are taken. A few seconds on mucoviscidosis makes sense?

Why do we carry out early detection examinations?

The following applies for you: No News are Good News. The screening examinations are for your baby's benefit. We want your baby to be healthy and therefore we examine the baby for diseases that can be dangerous for health.

Why do we carry out early detection examinations?

The newborn screening is conducted against your wishes. You as parents do normally not receive 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.

Who is informed about the testing result?

The paediatrician (physician) receives within a few days a written finding about the testing result. In urgent cases you will be contacted immediately.

What does the testing result tell us?

The finding tells you about your baby's health status. A result is either positive or negative. A positive result means that the newborn has a disease or illness. However, the symptoms can be improved or relieved by different therapies, so that the life expectancy has increased continuously.

How can mucoviscidosis be treated?

Mucoviscidosis (also called cystic fibrosis) is a congenital disease. 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 pulmonary function can be significantly wasting of time in order to finish the testing results immediately and tests can be repeated if necessary. You are in the examiners' care. In the lab, the samples are examined instantly by very special, sensitive methods.