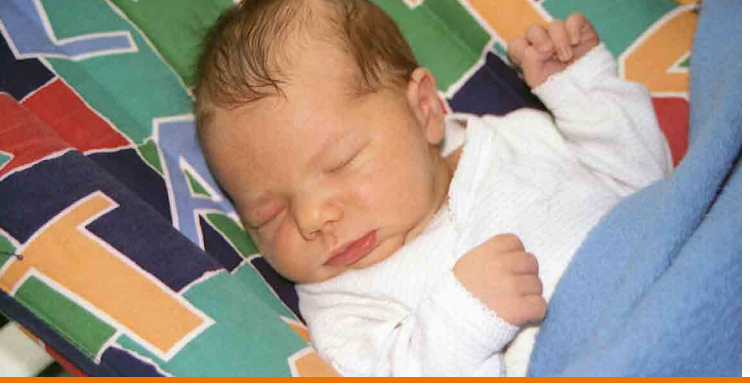

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Information for Parents

Newborn Screening

Note

Early treatment cannot completely prevent the effects of illness in all the disorders listed. Immediate treatment will allow the affected child to develop normally in most cases.

Only in the event of a clear indication of a disorder, which must be treated without delay, will you be informed by us or by the sender of the sample. Therefore, please provide a telephone number and address details for the test card, where you can be reached during the first few days after the birth. If the results are not clear for any other reason, the screening laboratory will contact you – by letter or via the sender – to ask you to attend further testing. The results of the hearing screening are available straight away and you will be informed immediately of the results, which will be recorded in the yellow book. Information about establishments where control tests can be carried out is provided with the report. The results are recorded on the screening card and passed on to the screening centre, which coordinates and follows up any necessary measures.

Important terms and conditions

- Participation in newborn screening tests is voluntary. The decision for or against newborn screening should be made based on reliable information. Your consent to testing applies only to listed disorders.
- Provisions are made for data protection: The results of the tests are of course subject to medical confidentiality and may not be passed on to third parties without your consent. You have the right to withdraw your consent at any time.
- The costs of the blood tests and hearing test are covered by statutory health insurance. For patients with private healthcare, the costs are usually covered – ask your health insurance company or benefit office for details.

If you have any other questions about the newborn screening programme, please ask your maternity clinic, paediatrician or midwife. You can always discuss your questions with your doctor.

The team at the newborn screening programme wish you and your child all the best for the future.

Disorders included in newborn screening tests

Congenital adrenal hyperplasia
Hormone imbalance caused by a defect in the adrenal cortex: Masculinisation of girls, may cause death due to loss of salt. Treatment through hormone supplements (incidence: approx. 1/10,000 births).

Biotinidase deficiency
Lack of the vitamin biotin in the metabolism: Skin changes, hearing and vision disorders, physical and intellectual development disorders. Treatment through administering biotinidase (incidence: approx. 1/80,000 births).

Metabolic carnitine deficiency
A defect in the metabolism of fatty acids: Metabolic crises, coma, possibly resulting in death. Treatment through a special diet (incidence: approx. 1/100,000 births).

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

Glutaric acidemia type 1
Inability to break down amino acids: Metabolic crises, permanently impaired movement. Treatment through a special diet (incidence: approx. 1/80,000 births).

Hypothyroidism
Congenital disorder of the thyroid: Severe impairment of intellectual and physical development. Treatment through administering hormones (incidence: approx. 1/4,000 births).

Isovaleric acidemia
Inability to break down amino acids: Metabolic crises, coma, intellectual disability. Treatment through a special diet (incidence: approx. 1/50,000 births).

MCAD/LCHAD/VLCAD deficiencies
Impaired ability to produce energy from fatty acids: Metabolic crises, coma, muscle and cardiac weakness, possibly resulting in death. Treatment by avoiding periods of hunger, special diet where applicable (incidences: approx. 1/10,000 (MCAD-D) to 1/80,000 births).

Cystic Fibrosis (Mucoviscidosis)
A genetic disorder, which affects the transfer of salt in gland cells and leads to the build-up of thick mucus in the airways and other organs, with persistent inflammation. Varying degrees of severity depending on genetics. Those affected are underweight and often have impaired growth. Restricted function of lungs and pancreas. Treatment through nutritional therapy, physiotherapy and medication (incidence: approx. 1/3,300 births).

Phenylketonuria (PKU) and maple syrup urine disease (MSUD)
Disorders in the metabolism of fatty acids: Seizures, severe impairment of intellectual and physical development. Treatment through a special diet (incidence: approx. 1/10,000 (PKU) or 1/200,000 births (MSUD)).

Hearing problems
The most common sensory impairment in children, various causes, can be in combination with other illnesses (heart, kidney, visual disorders). Early detection and treatment are vital for optimum speech and intellectual development. Late detection leads to lasting damage. Treatment includes early support, provision of hearing aids and, where necessary, cochlea implants (incidence: approx. 1-3/1000 births).

A hearing impairment may only occur as the child develops and it is therefore important that, even in the event of a normal test result, you as parents continue to observe your child's hearing ability.

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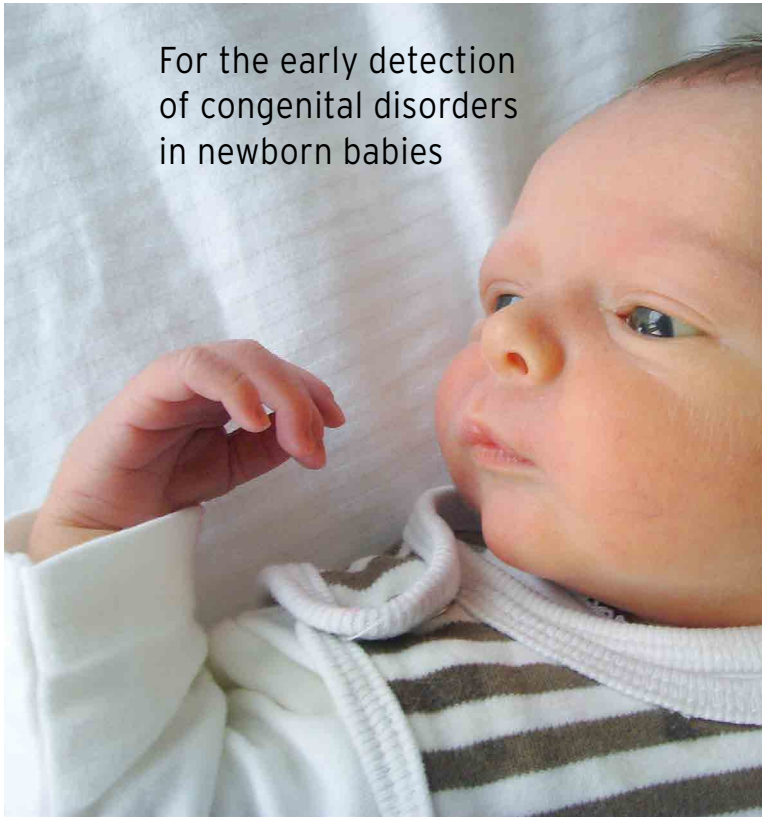

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Dear Parents,

The birth of your child is coming up very soon or has recently taken place. You would like your child to grow up in good health. This information sheet will tell you all about the early detection tests that are part of the newborn screening programme, which takes place a few days after the birth of your child. Hearing tests are important because any hearing impairment will lead to delays in speech development or may completely prevent speech from developing.

Why are early detection tests carried out?

Most children are born healthy but there are some exceptions. The health of about one in 1000 newborn babies is endangered by a rare congenital metabolic or organ function disorder (e.g. hearing impairment), which may not display any external signs. With many of these disorders, if treatment is provided as soon as possible after birth, there are very good chances of preventing deaths and avoiding or alleviating any lasting damage. Mass screenings, which are regulated by law, are therefore carried out in Germany for these kinds of disorders and this includes the screening of newborn babies.

By participating in the newborn screening programme, you will be helping to ensure the health of your child.

Which diseases are tested?

Newborn screening tests have been available in Germany for more than 30 years. These tests successfully detect congenital hypothyroidism, congenital adrenal hyperplasia, biotinidase deficiency, galactosemia and phenylketonuria (PKU). These tests have improved significantly over the past few years, covering further treatable protein and fat metabolism disorders and including hearing impairments. Some disorders are inherited and so the screening programme tests for genetic (inherited) features (genetic screening). Since 2016, there has also been a test for mucoviscidosis / cystic fibrosis (CF), which also involves (in about 1 in 1000 newborns) analyses of the genetic material (DNA). The individual diseases are described in detail overleaf.

What do the test results mean?

The results of a screening test are not a medical diagnosis and if you are called for repeat testing, this does not mean that your child is ill. Sometimes, it just means that the blood sample taken was insufficient to allow all the tests to be carried out or that the sample was taken too soon to safely detect all diseases or the results were borderline and need to be checked. Even in the rare cases for which the test results suggest the existence of a disorder, the diagnosis is not definitive and must be clarified by further medical tests. Please respond quickly if you are called for repeat testing in the newborn screening programme. The diagnosis and treatment centres will make appointments available at short notice. It is in your child's interest to clarify the situation quickly. A poor hearing test result does not always mean that your child has impaired hearing, but that the screening result needs to be checked. The first control measurement should take place in the maternity clinic wherever possible. You can find details of outpatient check-up centres and specialised clinics at the screening centre. In the event of irregular results from CF screening, the child should be taken to a specialised cystic fibrosis centre, since only one in 5 children with irregular results will actually be suffering from the disease. Here a "sweat test" will generally be carried out to confirm the diagnosis. This is safe, painless and the results are rapidly available. Other tests may also be necessary.

Is there a cure for these diseases?

All the diseases referred to here are genetic diseases and therefore no cure is available. For all the disorders that are tested, treatments are however available, which will help to prevent or reduce the effects of the disorder or improve quality of life, provided that the special course of treatment is started early enough. Specialist doctors are available to provide care and advice in the event of an illness being detected or suspected. For the hearing test, if there is still insufficient evidence about the hearing ability of your child even after the second test, a precise auditory threshold test will take place at a specialised facility. This test is painless and is carried out while the child is asleep. If the results show that hearing is impaired, the relevant treatment will be introduced immediately. If support is provided early enough, almost every child with impaired hearing will have good prospects for normal speech development.

When and how are the tests carried out?

The screening takes place two to three days (36 – 72 hours) after birth, usually at the same time as the second medical check up (U 2). For the hormone and metabolism screening, a few drops of blood are taken (from a vein or the baby's heel), dropped onto a special filter paper card and, once dry, sent to the screening laboratory. The samples are then tested for the various disorders using very sensitive methods. The screening for cystic fibrosis involves a three-stage process including two biochemical tests (IRT and pancreatitis associated protein) and a genetic analysis, in order to keep the number of control tests as low as possible. The hearing test involves measuring the inaudible signals generated by a healthy inner ear in a newborn child, using a completely painless process involving introducing a probe into the ear (otoacoustic emissions or OAE). Alternatively, the Automated Audiometry Brainstem Response (AABR) test is carried out. This involves testing the function of the inner ear to see whether sound waves are transmitted and processed as electrical impulses from the inner ear to the brain.

Who receives the test results?

Within a few days, a written report of the results of the hormone and metabolism screening will be sent to the address (maternity clinic, paediatrician) from which the blood sample was sent. The results of the cystic fibrosis screening may take up to 14 days as the process involves several stages. As a rule, parents do not receive any results.

This means that no news is good news.