

## Newborn Screening Information



### Information for parents

### For early detection of congenital disorders at newborns.



#### 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 send 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/HPA), maple syrup urine disease (MSUD), fatty acid metabolism defects (MCAD-, LCHAD-, VLCAD deficiency), carnitine cycle deficiencies, glutaric aciduria type 1, isovaleric acidaemia (IVA), tyrosinemia typ 1, severe combined immunodeficiency (SCID), cystic fibrosis (CF), 5q-associated spinal muscular atrophy (SMA) and sickle cell disease (SCD), (diseases exactly described in the following). 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, paediatrist) 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.

Early detection and treatment of concerned newborns is only possible when all parties such as parents, maternity clinic, paediatrist 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 unsuspecting testing results upon personal request at your maternity clinic or paediatrist.

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.

### Which diseases are included in the newborn screening?

<p><b>Biotinidase deficiency</b> Metabolic defect of the vitamin biotin: Skin alteration, metabolic crisis, mentally handicapped. Treatment by biotin intake. (Frequency: 1/80,000 newborns)</p> <p><b>Congenital adrenal hyperplasia</b> Endocrine disorder due to defect of the adrenal cortex: androgenisation at girls, possible lethal outcome at salt wasting crisis. Treatment by hormone intake. (Frequency: 1/10,000 newborns)</p> <p><b>Carnitine metabolic disorder (CPT I, CPT II, CACT)</b> Metabolic disorder of fatty acids: metabolic crisis, coma, possible lethal outcome. Treatment by special diet. (Frequency: 1/100,000 newborns)</p> <p><b>Galactosemia</b> Impaired metabolism of lactose sugars: Blindness, physical and intellectual disability, liver failure, possibly resulting in death. Treatment through a special diet. (Frequency: 1/40.000 newborns).</p> <p><b>Glutaric acidemia type 1 (GA 1)</b> Degradation defect of amino acids: permanent movement disorder, sudden metabolic crisis. Treatment by special diet. (Frequency: 1/80.000 newborns)</p> <p><b>Hypothyreosis</b> Congenital hypothyreosis: Severe mentally and physical development disorder. Treatment by hormone intake. (Frequency: 1/4.000 newborns)</p> <p><b>Isovaleric acidaemia (IVA)</b> Degradation defect of amino acids: mentally handicapped, coma. Treatment by special diet and intake of amino acids. (Frequency: 1/50.000 newborns)</p> <p><b>LCHAD/VLCAD</b> 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: 1/80.000 newborns)</p> <p><b>Maple syrup urine disease (MSUD)</b> Degradation defect of amino acids: mentally handicapped, coma, possible lethal outcome. Treatment by special diet. (Frequency: 1/200.000 newborns)</p>	<p><b>MCAD-deficiency</b> Energy production defect from fatty acids: metabolic crisis, coma, possible lethal outcome. Treatment by intake of carnitine, prevention of starvation periods. (Frequency: 1/10.000 newborns)</p> <p><b>Cystic fibrosis (CF)</b> Defect of a transport protein (CFTR). Causes a salt exchange disorder in the gland cells. Viscous mucous formation in the respiratory tract and organs which become permanently inflamed. Treatment: inhalation, physiotherapy, special diet, drugs. (Frequency: 1/3.300 newborns)</p> <p><b>Phenylketonuria (PKU/HPA)</b> Metabolic defect of the amino acid phenylalanin: seizures, spasticity, mentally handicapped. Treatment by special diet. (Frequency: 1/10.000 newborns)</p> <p><b>Severe combined immunodeficiency (SCID)</b> Complete lack of immune defense: in infancy high susceptibility to infections coupled with infectious complications. Untreated, most affected children die within 1 to 2 years. Therapy: bone marrow or stem cell transplantation, enzyme replacement therapy (Frequency: 1/35.000 newborns).</p> <p><b>Sickle cell disease (SCD)</b> Inherited condition of the red blood cells (crescent or sickle shape) leads to anemia and increased blood viscosity: shortage of red blood cells, organ damage, brain infarct or even early death. Treatment: medication, blood transfusion, stem cell transplantation.</p> <p><b>Spinal Muscular Atrophy (5-q SMA)</b> Life-threatening genetic neuromuscular disorder that affects the nerve cells controlling the muscles. People with SMA may have difficulty walking, eating, and breathing because of muscle weakness. The condition is serious and can lead to early death. Treatment: medication, physical therapy. (Frequency: 1/10.000 newborns).</p> <p><b>Tyrosinemia Typ 1</b> Degradation defect of the amino acid tyrosine: formation of harmful metabolic products can lead to severe damage to the liver, kidneys, brain and nerves. Treatment by special diet and medication (Frequency: 1/135.000 newborns).</p>
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## 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.

### 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.