Newborn Screening

Information for parents concerning the early detection of congenital disorders in newborn babies program

Dear Parents,

your child is already born or you will give birth shortly.
You want it to grow up healthy.
This information leaflet informs you about newborn screening, a preventive medical examination performed a few days after your child is born.
Why are these early diagnostic checks performed?
Most children are born healthy, but there are exceptions. Approximately one out of 1,000 newborns suffers from a rare congenital metabolic or organic disorder (e.g. hearing disorder), which cannot be detected through external manifestations.
With many of these diseases, the chances to avoid or diminish disabilities or prevent fatalities when the right therapy is initiated as soon after birth as possible are excellent. The hearing test is important because a hearing defect delays or prevents speech development altogether.

By participating in this examination program, you help secure your child's health. For statutory health insurance patients, this examination is free of charge.

When and how does the examination take place?

The screening examinations take place on the second or third day of your child's life (36 to 72 hours after birth), possibly together with the second preventive medical examination (U2). A few drops of blood are taken for the metabolic screening (from a vein or the heel), placed on the filter paper card provided for this purpose and immediately sent to a screening lab after drying.
There the samples are examined right away with special and very sensitive methods.
For the hearing test, the inaudible signals emitted by the newborn's healthy inner ear are measured completely pain-free with a probe inserted in the auditory canal (so-called otoacoustic emissions, OAE).

Which diseases does the screening include?

Newborn screenings are performed for over 30 years now in Germany. These serve to detect congenital hypothyroidism, adrenogenital syndrom, biotinidase deficiency, galactosemia and phenylketonuria (PKU). The examinations were significantly improved in the past years and other treatable diseases and organic defects (hearing) were included in the screening.
This makes it possible to detect a lot more protein and fat metabolism disorders. The individual diseases are described on the back. The newborn screening is not a genetic test, meaning the genome is not examined.

Who is informed about the results?

The person/institute submitting the blood sample (maternity clinic, midwife, paediatrician) is informed in writing about the results of the hormone and metabolism screening within a few days. The parents are not informed directly.

For you this means: no news is good news.

In case of a clear indication of a diseases which has to be treated immediately, we will inform you directly right away or pass this information on to your midwife or paediatrician. Therefore, please indicate your telephone number and address where we can contact you in the first days after the birth of your child. The results of the hearing test are available right away and communicated to the parents.

You may also receive a written report right in the clinic, which you should place in the yellow examination booklet.

The parents are informed about institutes where the necessary checkups can be performed together with the diagnoses.

The results are sent to the screening center together with the screening card and the necessary measures are initiated there.

What does the test result mean?

The result of a screening test is not a medical diagnosis and the request for a repeat examination does not mean that your child is sick. Sometimes there was just not enough blood for all examinations, or the blood was taken too soon in order to detect all diseases, or the result is borderline and has to be checked. In the rare cases where the examination result indicates a disease, the diagnosis is not final and must be cleared up in additional medical check-ups.

Please react as soon as possible when you are asked for repeat metabolic screening. It is in your child's interest if the situation is cleared up as fast as possible. The hearing test should be repeated early (2^{nd} to 3^{rd} day), for the other tests, the organ should mature a while longer and the examination should take place some weeks later.

Is there a cure for these diseases?

All the above stated metabolic defects and endocrine disorders are congenital and can therefore not be cured. However, the effects of these congenital disorders are prevented or at least diminished if special treatment is started as early as possible. Metabolism and hormone specialists are available for consulting and care in case of a suspected or actual disease. If the second hearing test also does not provide reliable information about you child's hearing, the exact hearing threshold is determined at one of the stated specialist clinics or doctor's offices. This examination is also completely pain-free and performed while your child is asleep.

The corresponding treatment measures are initiated immediately if it turns out that you child's hearing is impaired. With early treatment, almost any hearing-impaired child has the chance of normal speech development.

Important general conditions:

Participation in the newborn screening examinations is voluntary. Your consent to the examination only covers the diseases listed here.

Your personal data is also protected: the filter paper cards with the blood and your and your child's personal details are separated from each other after three months. These can only be reallocated in special cases and with your written consent.

The examination results are of course treated confidentially. If examinations are required at different institutes, the screening laboratories will compare the respective data to ensure optimum treatment.

The statutory health insurance covers the costs for the blood test; the parents or the health insurance companies are not charged for the hearing test at present. Most private health insurance companies also refund the costs. For details, contact your insurance company or government benefit office.

If you have any further questions about this newborn screening program, please contact your maternity hospital, your paediatrician or midwife.
The newborn screening team wishes you and your child all the best for the future.

Diseases detected in newborn screening

Adrenogenital syndrome
Hormone disorder caused by a defect of the suprarenal cortex: virilization in females, possible lethal course with salt loss crises. Treatment with hormones (frequency: approx. 1/10,000 newborns).

Maple syrup disease
Defect in the breakdown of amino acids: mental handicap, coma, possible lethal course. Treatment with special diet (frequency: approx. 1/200.000 newborns).

Biotinidase deficiency
Metabolic biotin defect: skin changes, metabolic crises, mental handicap. Treatment with biotin (frequency: approx. 1/ 80.000 newborns).

Carnitine metabolism defects
Defects in the fatty acids metabolism: metabolic crises, coma, possible lethal course. Treatment with special diet (frequency: approx. 1/100.000 newborns).

Galactosemia
Defect in the metabolization of lactose: blindness, physical and mental handicap, liver failure, possible lethal course. Treatment with special diet (frequency: approx. 1/40.000 newborns).

Glutaric aciduria type I
Defect in the breakdown of amino acids: permanent locomotive disorder, sudden metabolic crises. treatment with special diet (frequency: approx. 1/80.000 newborns)

Hypothyroidism
Congenital hypofunction of the thyroid gland: severe impairment of the mental and physical development. Treatment with hormones (frequency: approx. 1/4.000 newborns)

Isovalericacidemia
Defect in the breakdown of amino acids: mental handicap, coma, treatment with special diet and amino acid (frequency: approx. 1:50.000 newborns).

LCHAD/VLCAD
Defect in the metabolism of long-chained fatty acids: metabolic crises, coma, muscular and cardiac insufficiency, possible lethal course. treatment with special diet, avoiding hunger phases (frequency: approx. 1/80.000 newborns).

Medium chain acyl-CoA dehydrogenase deficiency
Defect in the energy production from fatty acids: metabolic crises, possible lethal course. Treatment with carnitine, avoiding hunger phases (frequency: approx. 1:10.000 newborns).

Phenylketonuria
Defect in the metabolism of the amino acid phenylalanine: seizures, spasticity, mental handicap. Treatment with special diet (frequency: approx. 1/10.000 newborns)

Notice: early treatment cannot completely prevent the manifestations of the above diseases. With immediate treatment, children will develop normally in most cases.