EARLY DETECTION SCREENING FOR NEWBORNS

A FACTSHEET FOR PARENTS

Universitätsklinikum Leipzig
Medizin ist unsere Berufung.

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DEAR PARENTS,

Most children come into the world healthy. However, there are some rare congenital disorders that are not apparent in newborns through external clinical signs. In order to detect these disorders early, an early detection screening is offered for your baby (extended newborn screening including cystic fibrosis). The disorders screened for occur statistically in approximately one in 1,500 newborns. If untreated, these disorders can lead to organ damage and irreversible physical or mental disability.

Why are early detection screenings carried out and what is the procedure?

Rare congenital disorders of the metabolism and of organ functions need to be detected in good time. Early treatment as soon as possible after birth will allow the effects of a congenital disorder in these children to be avoided or reduced. For this reason, appropriate blood tests have been performed on all newborns for over 30 years. These tests have been significantly improved since that time; other treatable conditions have meanwhile been included in the test. In the course of the second to third day of life (36th-72nd hour after birth), and at the latest together with the second preventive screening of your child, the U2, a few drops of blood are extracted (from the vein or the heel), dropped onto the filter paper card provided for the purpose and, after drying, sent directly to a screening laboratory. There, the samples are immediately examined with specific, very sensitive analysis methods.

What conditions are tested for?

Several disorders of the metabolism, hormone, blood and immune system as well as of the neuromuscular system are tested for (the conditions are described briefly in the info box). A congenital disorder is found in total in approximately one newborn in 1,500. In many of the affected families, such disorders have never occurred before. As the affected children may still appear to be perfectly healthy at birth, newborn screening is important in order to safeguard the children in good time against serious disorders and their effects, for example disorders of mental and physical development.

Who finds out about the tests result?

In any case, the sender of the blood sample receives a written finding from the screening center within a few days. In addition, parents are contacted directly in urgent cases. For this reason, please provide your phone number and the address at which you can be reached in the initial days after the birth on the test card. Early detection and treatment of affected newborns are only possible when all those involved – parents, hospital, paediatrician and screening laboratory – cooperate without delay, to allow the test findings to be collected and checked in good time.

What does the test result mean?

The finding of a screening test is not yet a medical diagnosis. The test result may either allow the disorders tested for to be largely ruled out, or make a further diagnostic test necessary, e.g. a repetition of the test, if there is suspicion of a condition. Repetition of a screening test may also be necessary, however, if for instance the time the blood sample was taken was not ideal.

Special feature of cystic fibrosis (mucoviscidosis)

What is the procedure for the cystic fibrosis test?

The cystic fibrosis test is a special case: If suspicion arises in the screening of cystic fibrosis on the basis of conspicuous laboratory parameters, a DNA test (genotyping) may be necessary. Thereby, solely genetic changes that can lead to cystic fibrosis are looked for. As the cystic fibrosis test, in contrast to the other tests, may be carried out up to 4 weeks after birth, the taking of a blood sample may also, if necessary, be done again in this period.

Who finds out about the finding of a DNA test?

A DNA test may become necessary in some circumstances to clarify the presence of cystic fibrosis. As is also the case with all other findings of the newborn screening, the finding of this DNA test is subject to medical confidentiality. The finding of the DNA test will only be communicated to you and the doctor who sent the sample if suspicion of cystic fibrosis arises from it.

HINWEIS: Zum Verbleib bei den Personensorgeberechtigten

What does a follow-up-needed cystic fibrosis test mean?

If a cystic fibrosis test requires follow-up, this does not necessarily mean that your child is actually suffering from cystic fibrosis. There could also exist a so-called predisposition. In this event, you will be referred to a specialized cystic fibrosis center for further evaluation.

Can these conditions be cured?

All the metabolic, neuromuscular and endocrinal disorders as well as blood and immune defects referred to are congenital and thus cannot be etiologically cured. The effects of these congenital disorders may, however, be avoided or at least reduced with appropriate early treatment. Treatment, depending on the diagnosed disorder, may consist of a special diet, administration of specific medicines, and/or specific preventive actions. Specialists are available for consultation and care in the event of suspicion of or presence of the condition.

You decide for your child!

Participation in newborn screening is voluntary; consent may be withdrawn at any time. In this event, any existing test cards will be destroyed and the test findings deleted.
The test costs are covered by your statutory health insurance. Private health insurers generally reimburse the costs. Please address any queries to your health insurance company.

Screeningzentrum | Sachsen

Saxony Screening Centre
www.screeningzentrum-sachsen.de

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Disorders tested for in newborn screening:

Cystic fibrosis (mucoviscidosis)
Condition in which viscous mucus accumulates for example in the lungs and pancreas. It can lead to irreversible organ damage up to and including organ failure. Therapy by special nutrition, remedial physiotherapy, and medication can slow and mitigate the course of the disease. (Incidence 1:3,300)

Hypothyroidism
Congenital subfunction of the thyroid. After several months, feeding problems, growth failure and mental disability appear. Therapy by administration of hormones. (Incidence 1:3,500)

Sickle cell disease (SCD)
Misformation of red blood cells ("sickle cells") leads to anaemia, increased blood viscosity and impaired organ oxygen supply, causing long term organ damage . Acute complications may be cerebral infarction, kidney failure, splenic infarction, blood poisoning and anaemia. Treatment involves education, infection prophylaxis (e.g. vaccinations), medication with hydroxycarbamide, blood transfusions and in some cases stem cell transplantation. Untreated, symptoms usually commence in the third month of life. (Incidence 1:3,950)

Phenylketonuria
Disorder in the breakdown of the amino acid phenylalanine. Causes serious mental and motor disorders. Therapy by special diet. (Incidence 1:5,000)

Spinal muscular atrophy (SMA)
Lack of the SMN (survival motor neuron) protein leads to increasing muscular weakness with decreasing motor skills and impaired lung function. Therapy involves medication as well as physiotherapy, rehabilitation, orthopaedics and psychotherapy. Early symptoms of infantile SMA (most common and severe form of SMA) appear in the first 6 months of life. Untreated, affected kids die within the first 2 years of life. (Incidence: 1:6,000 to 1:11,000)

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
Disorder in the breakdown of fatty acids. Early crisis or crisis arising after some months with hypoglycaemia, coma, sudden infant death. Therapy by avoidance of hunger periods and administration of carnitine. (Incidence 1:10,000)

Congenital adrenal hyperplasia (adrenogenital syndrome)
Disorder of the formation of adrenal hormones. May lead to false gender determination in girls and to life-threatening salt-wasting crises. Treatment by administration of hormones. (Incidence 1:13,000)

Biotinidase deficiency
Disorder in the metabolism of the vitamin biotin. Leads to metabolic crises, skin and hair changes as well as to mental disability. Therapy by administration of biotin. (Incidence 1:22,000)

Severe combined immunodeficiencies (SCID)
Affected children are born without acquired immune system, which leads to increased susceptibility to infections and severe complications. Strict hygienic precautionary measures are necessary. Therapy is based on bone marrow or stem cell transplantation and enzyme replacement therapy. No breast feeding, live vaccines or transfusion of untreated blood products is allowed. Without therapy, most affected children pass away within the first 2 years of life. (Incidence 1:32,500)

Long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) and Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
Disorder in the metabolism of long-chain fatty acids. Metabolic crises, coma, muscle weakness and cardiac insufficiency, can be fatal. Treatment by special diet, avoidance of hunger periods. (Incidence 1:55,000)

Galactosaemia
Disorder in the breakdown of milk sugar (galactose). Leads to serious liver and kidney damage, death in the first months, mental and physical disability. Therapy by galactose-free nutrition. (Incidence 1:70,000)

Isovaleric acidemia
Disorder in the breakdown of amino acids. Can lead to premature vomiting, coma and mental disability. Therapy by special diet. (Incidence 1:97,000)

Glutaric acidemia, type I
Disorder in the breakdown of amino acids. After initially unremarkable development a serious neurological crisis arises with apraxia and seizures. Therapy by special diet. (Incidence 1:132,000)

Tyrosinemia, type I
Disorder in breakdown of amino acids. Leads to serious liver and kidney damage; can be fatal. Therapy based on a combination of medication and specialized diet. (Incidence: 1:135,000)

Maple syrup urine disease
Disorder in the breakdown of amino acids. Leads to impaired consciousness, coma, seizures, mental retardation. Therapy by special diet. (Incidence 1:150,000)

Carnitine metabolic defects
Disorder in the metabolism of fatty acids. Leads to metabolic crises, coma, can be fatal. Treatment by special diet. (Incidence 1:500,000)

Note:
Prompt treatment cannot completely prevent all the consequences of all the conditions mentioned above. Immediate treatment allows the affected child to have a normal development in most cases.

HINWEIS: Zum Verbleib bei den Personensorgeberechtigten

DECLARATION OF CONSENT / EINVERSTÄNDNISERKLÄRUNG

Name of child: _____ Date of birth: _____
Name des Kindes Geburtsdatum

Street: _____ Postcode, City: _____
Straße Postleitzahl, Ort

I have received the information sheet and have been told in a consultation about the extended newborn screening including cystic fibrosis. I have been informed of the described diagnostic risks, of further tests of my child which may be necessary as a result of the screening, as well as of the possible negative consequences of a refusal and was able to ask questions. **I desire an extended newborn screening of my child including cystic fibrosis to be carried out.** I consent to the required taking of a blood sample and the laboratory testing for the conditions referred to in the information sheet.
I consent to the transmission of relevant personal data to the Saxony screening center for the purpose of medical reporting. The Saxony screening center may contact me directly in the event of abnormal findings. The result of the test may be relayed to attending and referred physicians. Medical confidentiality and data protection requirements will thereby be strictly observed. The sample material will be destroyed after a period of 3 months or after tests have been completed. Participation in these early detection screenings is voluntary; consent may be withdrawn at any time. In the event of withdrawal of consent, all samples will be destroyed and the test findings deleted.

Ich wurde in einem Gespräch über das Neugeborenenscreening (NGS) aufgeklärt, auf Risiken und Alternativen hingewiesen und konnte Fragen stellen. **Ich wünsche die Durchführung des Neugeborenenscreenings bei meinem Kind.** Ich willige in die erforderliche Blutentnahme und in die Laboruntersuchung auf die oben genannten Erkrankungen ein. Ich stimme einer Übermittlung personenbezogener Daten an das Screeningzentrum Sachsen zu. Das Screeningzentrum Sachsen darf mich bei auffälligem Befund direkt informieren. Das Ergebnis der Untersuchung darf nicht ohne meine Einwilligung an Dritte weitergegeben werden. Die Vorgaben der ärztlichen Schweigepflicht und des Datenschutzes werden dabei strikt eingehalten. Das Probenmaterial wird nach einem Zeitraum von 3 Monaten vernichtet. Die Teilnahme am NGS ist freiwillig. Die Kosten werden von der gesetzlichen Krankenkasse übernommen.

_____
Date, Name in block letters, Signature of parents* / of legal representative
Datum, Name in Druckschrift, Unterschrift der Eltern* / des gesetzlichen Vertreters

_____
Date, Name in block letters, Signature of informing doctor
Datum, Name in Druckschrift, Unterschrift des aufklärenden Arztes

REFUSAL OF THE EXTENDED NEWBORN SCREENING/ ABLEHNUNG ERWEITERTES NEUGEBORENENSCREENING

☐ I do **NOT** consent to an extended newborn screening of my child being carried out. I have been informed in a consultation of possible negative consequences of this decision (undetected disorders that may lead to permanent disability or early death).

Ich stimme der Durchführung des Erweiterten Neugeborenenscreenings bei meinem Kind **NICHT** zu. Ich wurde in einem Gespräch auf mögliche negative Folgen dieser Entscheidung hingewiesen (unentdeckte Erkrankungen, die zu dauerhafter Behinderung oder auch zum frühzeitigen Tode führen können)

_____
Date, Name in block letters, Signature of parents* / of legal representative
Datum, Name in Druckschrift, Unterschrift der Eltern* / des gesetzlichen Vertreters

*If only one parent signs, s/he affirms at the same time that s/he is acting with the other parent's consent or has sole custody of the child.
*Mit der Unterschrift nur eines Elternteils versichert dieser gleichzeitig, dass er im Einvernehmen mit dem anderen Elternteil handelt bzw. das alleinige Sorgerecht für das Kind hat.

REFUSAL OF NEWBORN SCREENING FOR CYSTIC FIBROSIS/ ABLEHNUNG MUKOVISZIDOSESCREENING

☐ I do **NOT** consent to a newborn screening for cystic fibrosis of my child being carried out. I have been informed in a consultation of possible negative consequences of this decision (undetected disorder that may lead to permanent disability or early death).

Ich stimme der Durchführung des Mukoviszidosescreenings bei meinem Kind **NICHT** zu. Ich wurde in einem Gespräch auf mögliche negative Folgen dieser Entscheidung hingewiesen (unentdeckte Erkrankung, die zu dauerhafter Behinderung oder auch zum frühzeitigen Tode führen kann).

_____
Date, Name in block letters, Signature of parents* / of legal representative
Datum, Name in Druckschrift, Unterschrift der Eltern* / des gesetzlichen Vertreters

*If only one parent signs, s/he affirms at the same time that s/he is acting with the other parent's consent or has sole custody of the child.
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HINWEIS: Für den Verbleib in der Patientenakte!