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Information for the child's legal representative about neonatal laboratory screening

What is neonatal laboratory screening?
Neonatal screening is used to screen for diseases in their early stages to enable early diagnosis and treatment of the newborn before the disease can manifest and cause irreversible damage to health.

Which diseases are screened for in neonatal laboratory screening?
The neonatal laboratory screening includes 18 diseases:

- congenital thyroid failure (congenital hypothyroidism)
- congenital adrenal failure (congenital adrenal hyperplasia)
- congenital disorder of airway mucus viscosity (cystic fibrosis)
- 15 hereditary metabolic diseases (phenylketonuria, etc.)
- spinal muscular atrophy (SMA) and severe combined immunodeficiency (SCID)

How is the neonatal laboratory screening performed?
A few drops of blood are collected from the baby's foot 48-72 hours after the birth on two screening cards (hereinafter referred to as screening cards). One screening card is sent to a laboratory at the University Hospital Brno, where the first three congenital diseases (congenital hypothyroidism, congenital adrenal hyperplasia and cystic fibrosis) are examined. The second screening card is sent to the laboratory of the University Hospital Olomouc, which performs tests for hereditary metabolic diseases.

Diseases are screened based on an increased amount of certain substances in the newborn's blood (e.g. proteins, hormones, amino acids, and acylcarnitines). The neonatal screening of cystic fibrosis detects the level of a substance produced by the pancreas (called immunoreactive trypsinogen - IRT) in the first phase. This test is sufficiently sensitive to detect most patients with cystic fibrosis, but elevated IRT may occur even if the newborn does not suffer from cystic fibrosis. Therefore, 1% of newborns with the highest IRT level undergo testing of hereditary changes in the cystic fibrosis gene in the second stage to differentiate truly positive screening results from false positive ones.

This test is provided by a specialised laboratory at the University Hospital Brno. The test is carried out directly from the dry drop of blood from the original sample, where the high level of IRT was detected. The result of this specific screening test is only used to conclude the screening result for cystic fibrosis as negative (i.e. the newborn does not suffer from cystic fibrosis) or positive (i.e. the newborn is suspected of suffering from cystic fibrosis and must be confirmed by further diagnostic examinations).
In the case of testing for hereditary changes in the cystic fibrosis gene, the DNA isolated from the screening card is kept for 2 months as it may be necessary to repeat the tests. The DNA is discarded after this time.

How is the outcome of neonatal screening reported?
Diseases investigated in the neonatal screening are serious congenital innate or hereditary diseases. Early detection of the disease is the first prerequisite for early start of effective treatment. The sooner the treatment is started, the more successful it is and the development of various complications may be prevented – some of these complications might also lead to irreversible damage to health.

Approximately 1 in 1,150 newborns suffer from any of the above-mentioned diseases and screening laboratories do not issue reports of normal (negative) findings due to the very low probability of the disease. If the laboratory does not contact the legal representative of the child, it means that none of the diseases under investigation are suspected. On the other hand, if there is a suspicion of any of the diseases, the laboratory will actively contact the legal representatives of the child (most often within 1 week of the sampling, usually within 4 - 6 weeks for cystic fibrosis screening). Hence, it is necessary to provide the healthcare facility with detailed contact details (full address, telephone contact) for the legal representative and the general practitioner for children and adolescents where the child will be registered.

If I am contacted by the screening centre, does it mean that the child is ill?
The screening examination is not a definitive diagnosis, it only provides a suspicion of a hereditary disorder. This must be confirmed or refuted by further detailed examination. The next steps depend on the urgency and the type of disease. In general, it can be only further collection of a blood drop from the heel or the collection of venous blood, exceptionally a quick admission to hospital.

Is it possible that any diseases are not detected by laboratory neonatal screening?
Only the diseases listed above can be detected by neonatal laboratory screening. Some of these diseases may rarely be missed in the diagnostic process (the so-called false negative finding). These are usually mild forms of the disease.

What happens to the screening paper after testing?
Screening cards - request forms are kept for five years with the health service provider that performed the laboratory examination and are protected against misuse according to the Decree No. 98/2012 Coll., on medical records.

Is it possible to refuse the collection of blood drop for laboratory neonatal screening?
In accordance with the guidelines of Czech medical societies, it is recommended consider such a step responsibly - whether this personal attitude is proportional to the risk of possible permanent damage to health due to an undetected disease. Should a child have any of the above-mentioned diseases, he or she will lose the chance of early treatment if the screening is refused. Health damages due to late initiation of treatment are serious and irreversible. There is no alternative procedure for screening relevant diseases.

What steps should I take if I do not wish my child to undergo a neonatal laboratory screening?
You will be informed about the scheduled sample collection by the nurse who will be taking caring for your child. If you do not wish your child to undergo this examination, notify us before the 48th hour of life of your child or at the latest before the screening itself. Your physician will go through the "Record of refusal of neonatal laboratory screening" with you and both you and your physician will sign it.

More information about neonatal screening and a list of healthcare facilities where screening is performed is available at www.novorozeneckyscreening.cz.