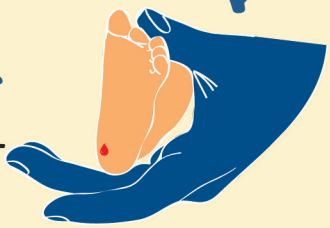
How is newborn screening done?

The probability that your baby will have any of the diseases screened for in newborns is very small, but newborn screening can considerably help the one out of 1,100 newborns who need help, or in some cases can even save a life.

All newborns born in the Czech Republic undergo newborn screening

The screening card contains:

- Dried blood spots
- Contact details of the mother and paediatrician



Sample collection 48-72 hours after birth

The cards are sent to two different laboratories, where they are safely kept for 5 years after they are analysed, and then are discarded.

Test results are usually available within 7 days (only for cystic fibrosis within 4-6 weeks).

NEGATIVE RESULT

None of the screened diseases is suspected. This result is not notified in writing.

UNCLEAR RESULT

A blood sample needs to be collected again, usually because the first sample was not collected properly, or was taken too soon, or as a result of medicines given to the newborn.

POSITIVE RESULT

There is a suspicion that the baby could have one of the diseases, other tests should follow.

The result of newborn screening may also be FALSE POSITIVE. This means that the first result indicates a certain disease which is not proven in the baby by subsequent tests.

For more details see the reverse side or www.novorozeneckyscreening.cz

Information about newborn laboratory screening for parents and legal representatives

What is newborn laboratory screening?
Newborn screening is used to search for diseases in their early stage so that these diseases in newborns can be diagnosed and treated before they manifest and cause irreversible damage to health. You will find more information about newborn laboratory screening at www. novorozeneckyscreening.cz.

What diseases does newborn laboratory screening look for?
18 diseases are screened for as part of newborn screening

- congenial severe deficiency of thyroid function (congenital hypothyroidism),
- congenial adrenal gland disorder (congenital adrenal hyperplasia),
- congenital disorder of viscosity of respiratory tract mucus (cystic fibrosis),
- 15 inherited metabolic diseases (phenylketonuria, selected organic acidurias and disorders of amino acid metabolism, fatty acid metabolism disorders, and biotinidase deficiency).

How is newborn laboratory screening done?
In the period between 48 and 72 hours after birth, a few drops of blood are taken from the newborn's heel and put on two filter paper screening cards (hereinafter referred to as the "screening cards"). One screening card is sent to the laboratory of the Královské Vinohrady University Hospital, or the University Hospital in Brno, where the first three congenital diseases (congenital hypothyroidism, congenital adrenal hyperplasia, and cystic fibrosis) are tested. The second screening card is sent to the laboratory of the General University Hospital in Prague, or the University Hospital in Olomouc, where inherited metabolic diseases are tested.
The diseases are looked for on the basis of a changed quantity of certain substances in the newborn's blood (proteins and enzymes, hormones, amino acids, and acylcarnitines). As part of newborn screening of cystic fibrosis, the level of a substance produced by the pancreas (so-called immunoreactive trypsinogen – IRT) is determined in the first stage. This test is sufficiently sensitive and should detect most patients suffering from cystic fibrosis, but an increased level of IRT may occur in many newborns who do not suffer from cystic fibrosis. Therefore, population-specific inherited changes in the gene for cystic fibrosis (*CFTR*) are tested in 1% of newborns with the highest IRT level in the second stage to distinguish whether the newborn could really have cystic fibrosis, or whether this is an unspecific diagnosis. The result of this second-level screening 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. there is a suspicion that the newborn could have cystic fibrosis, which must be confirmed or excluded by further diagnostic testing). This test is performed by specialised laboratories at the University Hospital in Motol (Prague) or the University Hospital in Brno directly in a dried blood spot of the original sample in which a high IRT level was discovered. If inherited changes in the gene for cystic fibrosis are tested, DNA isolated from the screening card is kept for 2 months, because the test needs to be repeated. After expiry of this period, the DNA is discarded.

How is the newborn screening result notified?
In view of the fact that the probability of disease is low (only approx. 1 out of 1,150 newborns suffers from any of the above diseases), health service providers that are specialised in newborn laboratory screening – screening laboratories (hereinafter referred to as the "provider") do not notify normal (negative) results. If the provider does not contact the newborn's legal representatives, it means that there is no suspicion that any of the tested diseases is present. On the contrary, if there is a suspicion that a tested disease may be present, the provider will actively contact the newborn's legal representatives or registering paediatrician (usually within 1 week of collecting a sample; within 4-6 weeks for cystic fibrosis). Therefore, when the sample is taken, it is necessary to give the contact details (full address, telephone number) of the legal representative and paediatrician with whom the baby is to be registered.

If the screening laboratory contacts me, does it mean that my baby is ill?
If the provider contacts you or the registering paediatrician, there is a suspicion that your baby could suffer from a disease. A screening test cannot give a definite diagnosis, and a suspicion needs to be confirmed or excluded by another detail test; approximately three-quarters of abnormal results are not confirmed by subsequent tests (so-called false positive results). Further steps will depend on the urgency and type of disease. If the probability of a disease is low, another blood drop from the heel may be required, if the probability of a disease is higher, a special test of a venous blood sample will follow, or exceptionally fast admission of the newborn to a hospital.

Is it possible that a disease is not diagnosed by laboratory newborn screening?
Newborn laboratory screening can only diagnose the diseases stated above. It may very rarely occur that one of the tested diseases is not diagnosed (so-called false negative results). In such cases, a mild form of the disease is usually present.

Is it possible to refuse the collection of blood drops for laboratory newborn screening?
Such a decision should be considered seriously by the legal representatives – whether such personal approach is adequate to the risk of potential permanent damage to the newborn's health caused by a disease which was not diagnosed. If a baby suffers from any of the tested diseases and you refuse screening, the baby will lose the chance of prompt treatment. Late treatment can lead to severe and irreparable damage to health. There is no alternative procedure enabling screening for the listed diseases.
If the collection of a sample is refused, but newborn laboratory screening is performed, it needs to be properly documented in the baby's medical records and signed by the legal representative. Should the legal representative refuse to sign, disagreement with completed screening will be signed by a witness who will confirm that the legal representative was notified of the possible consequences if newborn laboratory screening is not done.

What happens to the screening card after testing?
According to Regulation No. 98/2012 Coll., on medical records, screening cards – requests – are kept by the provider of health services that performed laboratory testing for a period of five years, and are protected against misuse.