Patient information

NEWBORN SCREENING FOR CONGENITAL DISEASES ~~IN NEWBORNS~~

Information for parents

All ~~newborns~~ babies born in Estonia are provided screening for ~~twenty~~ twenty-one different congenital diseases. The screening is available free of charge for the babies who are insured by the Estonian Health Insurance Fund.

Why is it necessary to screen newborns?

The purpose of the screening is to detect as early as possible certain hereditary or congenital diseases which may have a detrimental effect on your child's health or development~~,~~ if left without attention. The likelihood of a newborn baby having a hereditary or congenital disease is 3–4%. ~~One p~~Some~~art~~ of those ~~diseases~~conditions ~~is~~are formed by hereditary metabolic diseases in the case of which the production, breaking down or function of a certain protein, sugar, fatty acid, or chemical substance in the body is disturbed due to a genetic defect ~~which~~ that causes various ~~different~~ health or development issues for the child.

Those diseases cannot be diagnosed ~~in a timely manner~~on time without screening, as there are often no symptoms or the symptoms are barely detectable. By the time the child's underdevelopment or another health problem becomes evident, based on which the doctor may suspect the disease in question, the child's health will have already been damaged in a manner ~~which~~ that often cannot be reversed by treatment. Thus, health issues arising from diseases can only be prevented if we test all children and find those who require treatment before they develop symptoms of the disease. With timely treatment, most problems can be prevented and the child can develop equally with other children of their age.

How is the screening conducted?

The procedure is quick and simple. A midwife or a nurse will collect ~~on a special test slide~~ a blood sample from the heel or toe of a newborn baby 48–120 hours after birth on a special card. The sample~~. The test slide~~ is then sent to the laboratory ~~of~~ of the Department of Clinical Genetics ~~centre of clinical genetics of~~at the Tartu University Hospital which will conduct the required analyses and retain the test ~~slides~~ cards as required. If the blood samples are collected too soon or when the child is older than seven days, the test will become less accurate and the disease may be diagnosed with ~~a~~ delay.

The data are regularly anonymously analysed ~~anonymously in order~~ to check and improve the quality of the screening.

Is my baby still at ~~a~~ risk of developing those diseases if there have been no cases in our family?

Yes. As a rule, babies with congenital diseases ~~**the diseases screened for**~~ are born to ~~the~~ families with no previous history of those diseases. The risk remains even if the same parents already have healthy children together. Thus, the birth of a child with a congenital disease is often unexpected.

On average, thirteen to fifteen children are diagnosed with one of the diseases screened for in Estonia every year.

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But my child is healthy...

The diseases screened for cannot be associated with issues during pregnancy or labour and most of the babies affected appear fully healthy at birth and ~~in~~ during the first weeks after birth. The screening of newborns is designed to diagnose those diseases before the symptoms appear ~~in order~~ to start with treatment as soon as possible.

How will I receive the results of the screening?

If there are no abnormalities, the families or family physicians are not notified of the results but all results will be retained in the database of the laboratory of the Department of Clinical Genetics at the Tartu University Hospital ~~of the centre of clinical genetics of the Tartu University Hospital~~ and are also sent to the Patient Portal (www.digilugu.ee).

If a disease-related abnormality is found in the screening results, the parents and/or the family physician are contacted as soon as possible in order to quickly organise repeat tests. Thus, it is very important to accurately specify the contact details of the parents and the family physician on the test card.

Does a positive screening result mean that the baby has the disease which they were screened for?

No. The purpose of the screening is to identify those children who are at the highest risk of developing the diseases screened for, thus, being invited to repeat testing does not automatically mean that your baby has the disease.

Is the screening of newborns mandatory?

The screening of newborns is very important, but it is conducted ~~on a voluntary basis~~voluntarily and ~~it~~ is up to the parents to decide for their child. If you refuse to take part in the screening, you must notify your family physician of your decision.~~this.~~

The laboratory which is conducting the screening of newborns in Estonia is internationally acknowledged and is successfully participating in international quality programmes. However, the screening may sometimes provide false positive or false negative results, which means that further testing may be offered to a healthy child based on the screening result or, vice versa, some babies with a congenital hereditary disease~~s~~ may be deemed healthy by the screening. Thus, if your baby's physician has any reason to suspect any of the diseases screened for based on the symptoms of your child, carrying out new analyses is completely justified.

The test cards used for screening newborns are archived at least for 25 years.

Which diseases are screened for?

- Congenital hypothyroidism
- Classic galactosemia
- Amino acid metabolism disorders
    - Phenylketonuria
    - Maple syrup urine disease
    - Tyrosinemia type I
    - Homocystinuria
    - Argininemia
    - Citrullinemia type I
- Vitamin B12 deficiency

- Organic acid disorders
  - Isovaleric acidemia
  - Methylmalonic acidemia
  - Propionic acidemia
  - Glutaric acidemia type I
  - Glutaric acidemia type II
- Fatty acid metabolism disorders
  - Medium chain acyl-coA dehydrogenase deficiency
  - Long-chain fatty acid oxidation disorders
  - Very long-chain acyl-CoA dehydrogenase deficiency
  - Carnitine deficiency
  - Carnitine-acylcarnitine translocase deficiency
  - Carnitine palmitoyltransferase I deficiency
  - Carnitine palmitoyltransferase II deficiency

Further information about the diseases screened for is available at the following address:
https://www.kliinikum.ee/geneetikakeskus~~/vastsuendinute-skriining~~

Drawn up by the Department of Clinical Genetics ~~centre of clinical genetics~~

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