NEWBORN SCREENING FOR CONGENITAL DISORDERS

Information for parents

All babies born in Estonia are provided screening for twenty-one different congenital disorders. 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 congenital disorders, which may have a detrimental effect on your child's health or development if left without diagnosis. The likelihood of a newborn baby having a congenital disorder is 3–4%. Some of those conditions are inherited metabolic disorders when the production, breaking or function of a certain protein, sugar or fatty acid in the body is disturbed due to a genetic defect; and the disease may cause various health or development issues.

Those disorders usually cannot be diagnosed in time without screening, as there are often no symptoms or the symptoms are not specific ones. By the time, the child's problems as developmental delay or another health problem becomes evident, and the doctor will suspect the disease in question, the child's health is already damaged in a manner that often cannot be reversed by treatment. Thus, health issues arising from disorders can only be prevented if we test all newborns and find those who have the disease. Adequate treatment prevents most problems and the child develops equally with the peers.

How is the screening conducted?

The procedure is quick and simple. A midwife or a nurse will collect a blood sample to the special test card from the heel or toe of a newborn baby, at the age of 48–120 hours after birth. The timing for sampling is important. If the blood is drawn too early or too late (at second week of life), there could be a life-threatening delay in providing care to baby and there might be false negative results due to delayed blood sampling.

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

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

Yes. Most babies with the disorders screened for are born to families with no previous history of these diseases. The risk remains even if the same parents already have healthy children. Therefore, the birth of a child with a congenital disorder is often unexpected.

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

But my child is healthy...

The disorders screened for cannot be associated with issues during pregnancy or labour and most of the babies affected, appear healthy at birth and during the first weeks of life. The screening of newborns is designed to diagnose disorders 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 informed 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 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 having the disorders screened for. Thus, being invited to repeat testing does not automatically mean that your baby has the disease.

Is the newborn screening mandatory?

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

The laboratory, which is conducting the screening of newborns in Estonia is internationally accredited 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 disorder may be deemed healthy by the screening. Thus, if your baby's physician has any reason to suspect any of the disorders screened for based on the symptoms of your child, carrying out new analyses is highly indicated.

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

Which disorders are screened for?

- Congenital hypothyroidism
- Classic galactosaemia
- Aminoacidurias
  - Phenylketonuria
  - Maple syrup urine disease
  - Tyrosinaemia type I
  - Homocystinuria
  - Argininaemia
  - Citrullinaemia type I
- Vitamin B12 deficiency
- Organic acidurias
  - Isovaleric aciduria
  - Methylmalonic aciduria
  - Propionic aciduria
  - Glutaric aciduria type I
  - Glutaric aciduria type II
- Fatty acid oxidation defects
  - Medium chain acyl-CoA dehydrogenase deficiency
  - Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
  - Very long-chain acyl-CoA dehydrogenase deficiency
  - Carnitine transporter deficiency
  - Carnitine-acylcarnitine translocase deficiency
  - Carnitine palmitoyltransferase I deficiency
  - Carnitine palmitoyltransferase II deficiency
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Further information about the disorders screened for, is available at the following address: *https://www.kliinikum.ee/geneetikakeskus*

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