Screening for rare congenital diseases in newborns

(VasSeu screening sample)

Screening for rare congenital diseases is recommended for all newborn babies (STM = Ministry of Social Affairs and Health 7.4.2014). It is estimated that one in three thousand infants is born with a disease that can be discovered through screening. Permanent damage because of the disease can be avoided by early treatment. Without screening, it is often impossible to diagnose these diseases in time. Newborn screening has been introduced in most Western countries years ago.

Children born with serious congenital disorders may appear perfectly healthy as newborns. Screening tests are used to look for early signs of these rare disorders so that they can be detected, and the necessary treatment can be started in time. Early treatment can prevent severe permanent damage or even the child's death.

The screening sample is taken by pricking the baby's heel and drawing a few drops of blood into a piece of absorbent paper when the baby is 2–5 days old. If the screening result is abnormal, the family will be contacted immediately. Further testing will be done, and the doctor will perform a health examination on the child. Upon further testing, most of these children are also shown to be healthy. If the screening result is normal, the family will not be contacted but the screening result can be seen in My Kanta.

Newborn screening helps to prevent the harm of certain treatable congenital diseases. For the few infants born with these diseases, screening and early diagnosis can be lifesaving.

Below you will find more detailed information about the diseases screened for. More information can also be obtained at www.saske.fi.

IMPORTANT!

The screening sample is usually taken in the maternity ward at 2–5 days, i.e., 48–120 hours of age. If you are discharged from the maternity hospital when your baby is 36–48 hours old, the sample can be taken on discharge. If you are discharged before your baby is 36 hours old, the sample will be taken at a separate laboratory visit at 48–120 hours of age.

For more information on sampling and screening, please contact the staff at the maternity hospital.

Information about the diseases screened for:

Congenital hypothyroidism (CH) causes a deficiency in thyroid hormone. This hormone is essential for a child's growth and brain development. Thyroid hormone deficiency within the early years of life leads to severe growth failure and permanent intellectual disability. If the hormone deficiency is detected early and the treatment with hormone supplements is begun within the first weeks of life, the child will grow and develop normally. In Finland, the incidence of congenital hypothyroidism in newborns is c. 1:2000–1:3500. The disease is rarely hereditary.

Congenital adrenal hyperplasia (CAH) is caused by a defect in steroid hormone production in the adrenal cortex. These steroid hormones regulate vital functions, such as the blood sugar balance and the balance between salt and water. Left untreated, in the most severe form of the disease, a child could die if the salt and water balance is severely affected. The disease also increases the secretion of male hormones. In Finland, the incidence of congenital adrenal hyperplasia in newborns is c. 1:15 000–1:20 000. The disease is treated by hormone replacement therapy, which allows the patient to lead a normal life.

Severe combined immunodeficiency (SCID) is a rare inherited condition in which the body's immune system does not work properly. As a result of the defect, the child's body cannot fight off serious and life-threatening infections. Therefore, left untreated, the disease leads to death usually during the first two years of the child's life. Most children can be cured by stem cell transplantation if the disease is detected and treated before the onset of infections. The incidence of SCID in Finland is estimated to be 1:50 000–1:100 000.

The inborn diseases of amino acid and fatty acid metabolism that are included in the screening (such as PKU, LCHADD, MCADD, and GA1) are rare. There are no precise data concerning the incidence of all these diseases in Finland, but it is estimated that about a dozen children are born annually with some of these disorders. The inborn metabolic diseases are usually treated with special diets and nutritional preparations. There is also a specific drug for some of the disorders.

The inborn metabolic diseases included in the screening program often cause serious malfunctions in metabolism. There can be disruptions in energy production, or the body can accumulate toxic substances. The symptoms may include vomiting, poor growth, intellectual disability, and even death. It is estimated that 5 % of sudden infant deaths are caused by inborn metabolic disorders. The diseases screened for can usually be treated effectively if the treatment is started early. The prognosis is essentially dependent upon how much damage has been done prior to the start of treatment.

Most of the diseases screened for are inherited in an autosomal recessive manner. This means that a gene mutation is passed on via healthy carriers from one generation to the next, and the disease only manifests if both parents are carriers of the mutated gene and they pass the mutation on to their child. In each of these pregnancies, there is a 25 % chance that the child will have the disease.