Screening for rare congenital diseases in newborns

(VasSeu2 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 disorder that can be discovered through screening. Permanent damage as a result of the disease can be avoided by early treatment. Without screening, it is often impossible to diagnose these disorders in time. Newborn screening has been introduced in most Western countries years ago.

Children who are born with serious congenital disorders may appear completely healthy as newborns. Screening tests are used to check 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 permanent damage or even the death of the child.

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

Newborn screening helps to prevent the harmful effects of certain treatable congenital disorders. For the few infants born with these disorders, screening and early diagnosis can be life-saving.

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

IMPORTANT!

The screening sample is usually taken in the maternity ward when the infant is 2–5 days old. If you go home when the baby is 36–48 hours old, the sample can be taken when leaving the hospital. If you go home before the baby is 36 hours old, the sample is taken in the laboratory at the age of 2–5 days. You can get more information about screening from paediatricians at the maternity hospital.

Information about the diseases screened for:

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 disorder, a child could die if the salt and water balance is dramatically affected. The disorder also increases the secretion of male hormones. The incidence of congenital adrenal hyperplasia in newborns is c. 1:10 000 – 1:20 000. The CAH disorder is treated by hormone replacement therapy.

Metabolic amino acid and fatty acid disorders and disorders causing the accumulation of organic acids (such as PKU, LCHAD, MCAD, GA1, MSUD and tyrosinemia) that are included in the screening are rare. There are no precise data concerning the incidence of these diseases in Finland, but it has been estimated that about a dozen children are born annually who are affected by this group of disorders. These diseases are usually treated with special diets and nutritional preparations.

The disorders 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 death syndrome cases are caused by congenital metabolic diseases. The majority of the disorders screened for can be treated effectively. The prognosis is essentially dependent upon how much damage has been done prior to the start of treatment.

Most of the disorders 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.