

Metabolic disease too easily missed

Dutch researcher Terry Derks has demonstrated that the metabolic disease MCAD deficiency can be detected at an early stage. At present the disease is only found in half of the expected number of patients. With the help of a new screening method, all newborns can now be screened. Young children can develop complications due to MCAD deficiency if this is not diagnosed on time. Where early detection fails, it is too late to intervene in a quarter of cases.

A recently developed screening method makes it possible to screen newborns for several metabolic diseases, such as MCAD deficiency. Over the past three years researchers have tested the screening for MCAD deficiency in the north of the Netherlands by means of the heel prick. In this trial project, MCAD deficiency was found to be twice as prevalent than had been predicted. Moreover it was detected four times as frequently as is the case without screening.

How prevalent is the condition? The number of patients can be predicted based on the presence of changes in the DNA, so called gene mutations, which can cause the disease. From this the expected number of patients was 1:12,100. And how often was this disease found? Without the heel prick test 1 in 27,400 newborns in the Netherlands were diagnosed with MCAD deficiency. This happens after the patient shows complaints. Therefore the disease was not discovered in more than half of the patients. With the heel prick screening in the northern part of the country the researchers observed the disorder in 1 in 6600 newborns.

MCAD deficiency was found to be diagnosed more often in the north than in the south of the Netherlands. Interestingly, a north-south gradient for the prevalent MCAD gene mutation is present throughout Europe.

In patients with MCAD deficiency the MCAD enzyme does not function well enough. This enzyme is needed for the production of extra energy. Extra energy is particularly important under certain conditions, for example, during infections, fasting or major exertion. For young children, in particular, having the MCAD deficiency can be dangerous. Normally, they need more energy than adults, and during fever this demand increases. Young children with MCAD deficiency and fever can develop serious complications, probably due to the shortage of energy. As a result of this a simple infection can even become life threatening. About one-quarter of the clinically ascertained patients with MCAD deficiency who are admitted to hospital die before the diagnosis is known. A significant proportion of the children who survive the complications remain permanently disabled.

After an early diagnosis, a child can be helped with a simple treatment. With the new screening method, MCAD deficiency can be investigated for during the heel prick test neonatally. All newborn babies in the Netherlands receive the heel prick. At the recommendation of a Health Council of the Netherlands committee, the heel prick has been extended since 1 January 2007 to include, for example, screening for MCAD deficiency. The committee based its recommendations on a combination of literature research and scientific opinion. For MCAD deficiency, Derks and colleagues provided the scientific evidence that screening newborns is worthwhile.

Source: Netherlands Organization for Scientific Research

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