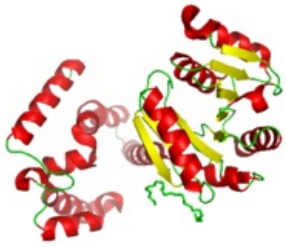
LCHAD deficiency

LCHAD deficiency is an autosomal recessive hereditary disorder (OMIM 609016 (https://omim.org/entry/609016)). LCHAD is a **long-chain 3-hydroxyacyl-CoA dehydrogenase** of the long-chain fatty acids. This enzyme is responsible for the transformation of fats into substitutive sources of energy for the human body.



3-hydroxyacyl-CoA dehydrogenase

Clinical signs

LCHAD deficiency can endanger the patient mainly during starvation, increased energy consumption or physical activity. In this case, the patient is not able to obtain energy from the fats of his own body. LCHAD symptoms commonly manifest during **early childhood**. These signs are for example faintness, hypoglycemia (low level of sugar in the blood), hypotonia (reduced muscle tone) and problems with the liver. In the latest stadium of the childhood, patients can have muscle pains, muscle tissue disorders, loss of the sensitivity in limbs, myocardial infarction or problems with breathing.

Diagnostics

LCHAD deficiency is diagnosed with help of the neonatal screening. The important thing is the long-chain hydroxyacylcarnitine level: increased values most likely show that the patient has LCHAD deficiency.

Treatment

- **Increased** amounts of sugars in food.
- **Reduced** amounts of fats.
- Usage of MCT fats, which means medium-chain triglycerides. Here belong for example breast milk, coconut milk, cow and sheep milk.
- In early childhood apply the diet every 4 hours.

Cause of the LCHAD deficiency

LCHAD deficiency is caused by mutation of the **HADHA** gene. This gene arranges instructions for the creation of the part of the enzyme complex which is called **mitochondrial trifunctional protein**. This protein contains 3 enzymes, each of which has different functions. The common task of all these enzymes is **the dissociation of long-chain fats**.

Mutations in HADHA the gene cause the dysfunction of one of these three enzymes. As a result, fatty acids cannot be transformed into energy, and their increased amounts in the body can cause life-threatening conditions.

Links

Bibliography

- MURRAY, Robert Kincaid – BENDER, David A – BOTHAM, Kathleen M, et al. *Harperova ilustrovaná biochemie.* 5. edition. Praha : Galén, 2012. 730 pp. ISBN 978-80-7262-907-7.

External links in Czech

- Problematika deficitu LCHAD (anglický jazyk) (https://ghr.nlm.nih.gov/condition/long-chain-3-hydroxyacyl-coa-dehydrogenase-deficiency)
- Stránky o screeningu (http://www.novorozeneckyscreening.cz/deficit-lchad)