# Hereditary disorders of fat metabolism

**Hereditary disorders of fat metabolism** in a broader context include:

- fatty acid transport and oxidation disorders;
- sphingolipidoses (lipidoses, lysosomal fat storage diseases): Gaucher disease, Niemann-Pick disease, Krabbe disease, metachromatick leukodystrophy, Fabry disease, abd gangliosidoses;
- disorders of lipoprotein metabolism;
- disorders of peroxisome metabolism – involved, among other things, in the metabolism of very long chain fatty acids and the synthesis of ether phospholipids (plasmalogens) [1].

The neonatal laboratory screening in the Czech Republic includes:

- **beta-oxidation disorders**:
  - medium chain fatty acid acyl-CoA dehydrogenase deficiency (MCAD deficiency),
  - long chain fatty acid 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency),
  - very long chain fatty acid acyl-CoA dehydrogenase deficiency (VLCAD deficiency);
- **beta-oxidation transport disorders**:
  - carnitine palmitoyltransferase I (CPT I) deficiency,
  - carnitine palmitoyltransferase II (CPT II) deficiency,
  - carnitine acylcarnitine translocase (CACT) deficiency.[2]

## Disorders of β-oxidation of fatty acids

- Beta-oxidation: oxidation of fatty acids to acetyl-CoA, takes place in mitochondria;
  - contributes significantly to meeting energy needs during the starvation period;
  - a direct source of energy for heart and muscle tissue and a source of ketone bodies for the CNS;
- more than 20 disorders are known, AR hereditary; occurrence of 1: 5000;
- most common disorders: **MCAD** (Medium-Chain-Acyl-CoA Dehydrogenase) and **LCHAD** (Long Chain-3-OH-Acyl-CoA Dehydrogenase).[1]

**MCAD a LCHAD**

- **clinical picture** (MCAD+LCHAD): in infant and toddler age **hypoketotic hypoglycemia** → convulsions;
  - or **Reye-like syndrome** attack in impaired consciousness and hepatomegaly;
  - sudden death syndrome;
  - LCHAD: may also begin in neonates with cardiomyopathy with acute heart failure or in older children with muscle weakness, rhabdomyolysis attacks with myoglobinuria, neuropathy, progressive cardiomyopathy and retinitis pigmentosa;
- **diagnosis**: neonatal screening - coupled mass spectrometry; examination of MK β-oxidation parameters in lymphocytes; enzymatic and molecular examination;
  - hypoketotic hypoglycemia, decreased carnitine;
  - LCHAD: increased creatine kinase and myoglobin in the blood after increased physical activity;
  - urine: dicarboxylic aciduria (MCAD), 3-OH-dicarboxylic aciduria + exercise-induced myoglobinuria (LCHAD);
  - liver steatosis;
- **therapy**: prevention of starvation → frequent diet with fat reduction (for LCHAD + substitution of MCT oil, especially before any physical exertion); nutrition also during the night (infants and toddlers 2x / night meal with maltodextrins, older children meal 1x / night with uncooked corn starch);
  - in case of fever increase the intake of sweet drinks, in case of vomiting and diarrhea early administration of glucose;
- **prognosis**: without early diagnosis, the risk of death under the picture of sudden death syndrome or Reye-like syndrome.[1]

## Links

### Related articles

- Mitochondrial diseases / Disorders of beta oxidation and ketogenesis
- Lipidosis
- Disorders of lipoprotein metabolism: Famlilial hypercholesterolemia • Dyslipidemia
- Lysosomal disorders
- Disorders of peroxisome metabolism
- Mitochondrial disorders of energy metabolism
- Hereditary disorders of amino acid metabolism • Disorders of urea cycle • Organic acidura
- Hereditary disorders of carbohydrate metabolism • Glycogenosis
- Disorders of protein glycosylation

- Hereditary disorders of purine and pyrimidine metabolism
- Porphyrias

## References

- ws:Dědičné poruchy metabolismu tuků

1.
2.

Template:Pahýl

Kategorie:Pediatrie Kategorie:Patobiochemie Kategorie:Endokrinologie