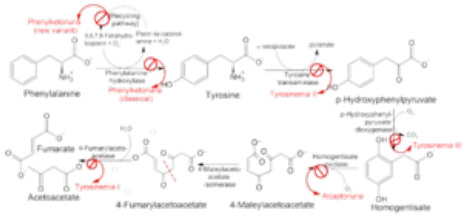
Tyrosinemia

This article has been translated from WikiSkripta; ready for the **editor's review**.

Tyrosinemia is a disease caused by a disorder in the metabolism of the amino acid tyrosine. Tyrosine is important for the synthesis of proteins (proteosynthesis), dopamine, adrenaline, norepinephrine, melanin and thyroxine. It is obtained partly from the diet and partly synthesized in the liver from phenylalanine.



Phenylalanine and tyrosine metabolism and its disorders

- Metabolic diseases with hyperthyrosinemia (hereditary AR, incidence 1: 50-100,000):
  - **tyrosinemia I**: fumarylacetoacetate hydroxylase (FAH) disorder,
  - **tyrosinemia II**: tyrosine aminotransferase disorder,
  - **tyrosinemia III**: 4-hydroxyphenylpyruvate dehydrogenase disorder (4-HPPD);
- Secondary hypertyrosinemia: manifestation of hepatopathy in neonates with congenital CMV infection;
- Transient hypertyrosinemia: in the first 2 weeks of life with a high protein content in the newborn's diet; benign; rapidly decreases after administration of vit. C and reduction of protein intake.name="KlinPed2012:138"></ref>

Tyrosinemia type I

Fumarylacetoacetate hydroxylase (FAH) disorder. Tyrosine is metabolized in the liver and kidneys by an alternative route to the tissue toxin **succinylacetone**, which causes progressive liver and kidney function. Symptoms of liver disease include anorexia, vomiting, hepatomegaly. Renal impairment can lead to metabolic acidosis. Some may develop a "porphyric crisis" with manifestations of peripheral neuropathy or paralytic ileus;

Diagnosis: acute disruption of the internal environment, hepatopathy, coagulopathy, high alpha-fetoprotein, increased tyrosine and methionine, increased serum succinylacetone concentration; urine: increased succinylacetone concentration; molecular-genetic diagnostics.

Therapy:

- comprehensive treatment of acute crisis, including hemodialysis;
- long-term low-protein diet + supplementation of essential amino acids without phenylalanine and tyrosine + pharmacological treatment (NTBC, 2- (2-nitro-4-trifluoromethylbenzoyl) -1,3-cyclohexanedione) - inhibition of tyrosine degradation at the 4-HPPD enzyme level → inhibition of succinylacetone formation;
- event. liver transplantation.

Prognosis: good with early diagnosis and treatment.[1]

Tyrosinemia type II

Autosomal recessive inherited **tyrosine-aminotransferase** enzyme deficiency. The eyes, skin and CNS are affected. Herpetiform corneal involvement is manifested by eye pain, tearing and photophobia. Hyperkeratosis of the palms and soles is noticeable. Mental retardation occurs in 50% of patients. We observe increased levels of **tyrosine** (it is also excreted in the urine). Treatment is a diet without phenylalanine and tyrosine.

Transient neonatal tyrosinemia

The transient increase in plasma tyrosine in the first two weeks of life is due to delayed maturation of tyrosine aminotransferase or 4-hydroxyphenylpyruvate dioxygenase enzymes in the liver[2]. Newborns have a positive Guthrie test, usually screened for phenylketonuria (vitamin C can be given; spontaneous adjustment). High levels of tyrosine in the blood occur in scurvy, hyperthyroidism and liver failure.

Links

Related articles

- Hereditary disorders of amino acid metabolism
- Tyrosine

Source

- BENEŠ, Jiří. *Studijní materiály* [online]. ©2007. [cit. 2010-04]. <http://www.jirben.wz.cz/>.

References

1. **Cite error: Invalid <ref> tag; no text was provided for refs named `KlinPed2012:138`**
2. MUNTAU, Ania Carolina. *Pediatrie.* 4. edition. Praha : Grada, 2009. pp. 102. ISBN 978-80-247-2525-3.

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