

Wilson's disease

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__BEZOBSAHU__ Template:Infobox - genetická choroba

Wilson's disease, *hepatolenticular degeneration* is an autosomal recessively inherited metabolic disease with a prevalence of approximately 1:25 000–30 000 and a frequency of heterozygotes in the population of approximately 1:90. It is characterized by abnormal accumulation of copper in the liver, which causes liver cell damage, CNS dysfunction and hemolytic anemia.^[1]

Etiopatogenesis

The disease is caused by a mutation in the *ATP7B* gene on chromosome 13 (13q14.3–q21.1). This gene encodes a copper-transporting ATPase.

A defect in this protein results in impaired biliary excretion of copper and incorporation of copper into apoceruloplasmin in hepatocytes. As a result of impaired copper excretion into the bile, this metal accumulates in the liver, brain and other organs and leads to an excess of free radicals causing damage to these organs.

File:Hepatocop.jpg
Copper transport in hepatocyte

Clinical picture

- Neurological manigestations – tremor, deterioration of school performance, deterioration of handwriting (motor disorders), mental changes, progression to severe extrapyramidal syndrome.
- Anemia,coagulation disorders due to portal hypertension.
- Gradual progression of liver fibrosis to cirrhosis – jaundice, arachnid hemangiomas, portal hypertension, liver failure^[1].
- In 5% of those affected, the disease manifests as fulminant liver failure.
- Other possible symptoms: renal acidosis, bone disease, hormonal disorders, growth disorders.

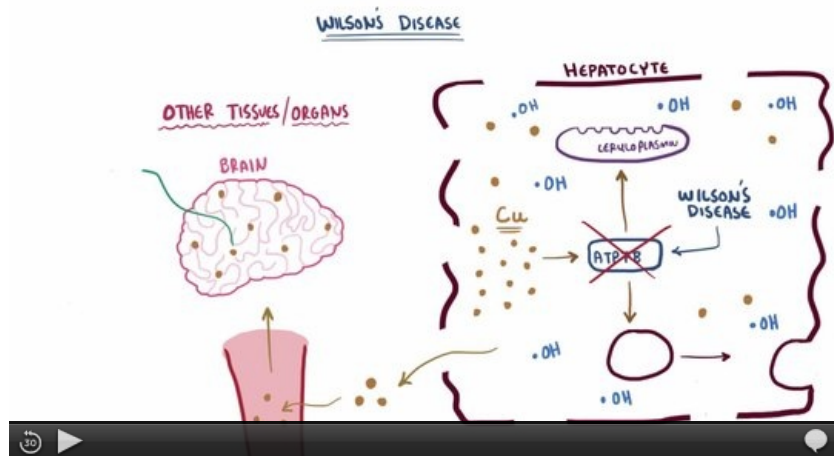
Diagnostics

- decreased serum ceruloplasmin levels;
- increased urinary excretion of copper (in 24 hours);
- Kayser-Fleischer ring at the edge of the cornea^[2];
- hemolysis;
- increased copper content in the liver;^[1]
- the diagnosis can be confirmed by molecular genetic testing of the ATP7B gene (it is also available in the Czech Republic).

Therapy

- lifelong treatment is the prevention of liver and CNS damage;
- reduction of copper-rich foods (sea fish, chocolate, cocoa);
- administration of copper chelating drugs (Penicillamine 1000 mg / day);
- zinc – reduces the resorption of copper by the intestine;
- monitoring urinary excretion of copper^[1];
- liver transplantation.

Summary video



Video in English, definition, pathogenesis, symptoms, complications, treatment.

Odkazy

Související články

- Gen ATP7B

Externí odkazy

- Wilsonova choroba – text na stefajir.cz (<http://www.stefajir.cz/?q=wilsonova-nemoc>)

Reference

- KLIEGMAN, Robert M. – MARCDANTE, Karen J. – JENSON, Hal B.. *Nelson Essentials of Pediatrics*. 5. edition. China : Elsevier Saunders, 2006. pp. 619-620. ISBN 978-0-8089-2325-1.
- VOKURKA, Martin – HUGO, Jan. *Velký lékařský slovník* [online] . 8. edition. Maxdorf, 2009. 1144 pp. Available from <<http://lekarske.slovniky.cz/>>. ISBN 978-80-7345-166-0.

Použitá literatura

- MAREČEK, Zdeněk. *Diagnostika a léčba Wilsonovy choroby* [online]. [cit. 2009-12-14]. <<http://web.archive.org/web/20100805235116/http://www.ceska-hepatologie.cz:80/index.php?node=44#content-left>>.
- ČEŠKA, Richard, et al. *Interna*. 1. edition. Praque : Triton, 2010. 855 pp. ISBN 978-80-7387-423-0.

Doporučená literatura

- Lutschenko laboratory – stránka výzkumné laboratoře, která se věnuje metabolismu mědi (http://www.ohsu.edu/xd/research/centers-institutes/octri/funding/pilot-project-lutsenko-sveta.cfm?WT_rank=1)
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Template:Navbox - monogenně dědičné choroby