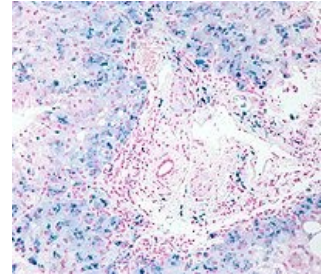


Hemochromatosis

Hereditary hemochromatosis (also **genetic hemochromatosis**, abbreviated **HH** or **GH**) is the most common monogenically transmitted disease, which manifests itself after a long period of time diseases (40 years and over). It occurs with an incidence of 2-5/1,000 inhabitants, when the incidence of heterozygotes for this disease is 1:10.

Hemochromatosis:



Liver biopsy stained with Berlin blue. Iron deposits in hepatocytes are shown as blue granules.

The essence of hemochromatosis is a **disruption of the HFE gene** on the 6th chromosome, which leads to **excessive storage of iron** in hepatocytes. Iron is excessively reabsorbed in the duodenum. The disease is autosomal recessive with incomplete penetrance. An excess of iron is of course **toxic**, because on the basis of Fenton's reaction, there is a higher production of the **hydroxyl radical**, which destroys cells and cellular structures. Typically, iron is deposited first in hepatocytes, then bile ducts, Kupffer cells and macrophages, where the main and most serious manifestation of the disease is the development of liver cirrhosis with the possible emergence and the development of hepatocellular carcinoma. Other organs damaged in hemochromatosis are the myocardium, pancreas, joints and testicles. Known by definition is the so-called **bronze diabetes**, which arises from the destruction of the pancreas, or the islets of Langerhans, and the inability to produce insulin.

Diagnostics

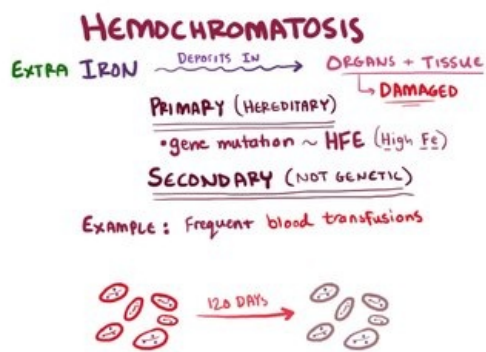
- The patient will typically have elevated iron, serum ferritin, and increased transferrin saturation (80-90%).
- Increased values of ALT, AST.
- Prolongation of prothrombin time.

Investigation

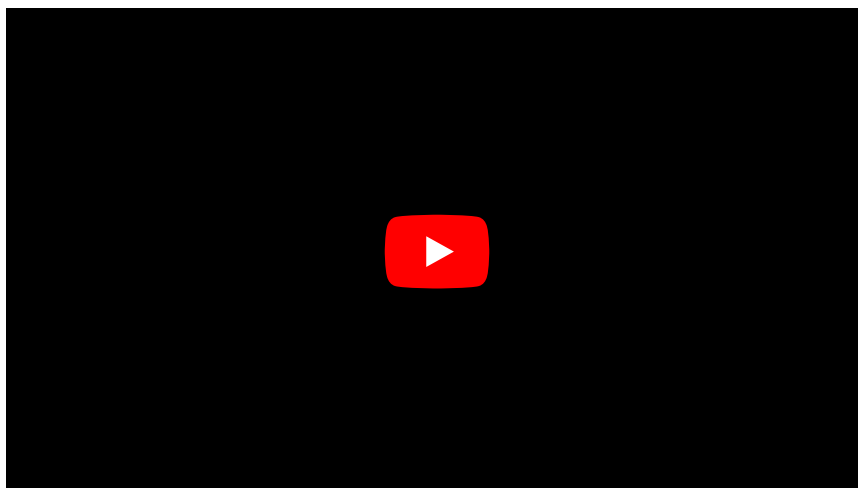
- USG liver;
- liver biopsy;
- genetic examination.

Clinical features

- Hepatomegaly (due to a large amount of stored ferritin);
- manifestations of liver cirrhosis;
- joint pain;
- cardiomegaly;
- insulin resistance (DM);
- hepatocellular carcinoma.



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



Treatment

The best and so far the only treatment is **venipuncture** (injection through a vein), which at least slows down the progression of the disease. Women with this disease are slightly protected by menstrual blood loss, however, they do not have a better prognosis in the long term.

Links

Related Articles

- Disorders of iron metabolism
- Iron

External links

- Recommended CHS procedure for diagnosis and treatment of genetic (hereditary) hemochromatosis (<http://www.ces-hep.cz/file/326/doporuceny-postup-chs-hemochromatoza.pdf>)
- Hemochromatosis genetic on medicabaze.cz (http://www.medicabaze.cz/index.php?sec=term_detail&catId=6&termId=1234&tname=Hemochromat%C3%B3za+genetick%C3%A1)
- Hemochromatosis - youtube.com video (<https://www.youtube.com/watch?v=iUga3oM65Bc>)

Source

- Hemochromatosis on Stefajir.cz (<http://www.stefajir.cz/?q=hemochromatoza>)
- Hemochromatoza na vitalion.cz (<https://nemoci.vitalion.cz/hemochromatoza/>)

Recommended reading

- SOBOTKA, Paul, et al. *Pathological physiology : practicum*. 4. edition. Prague : Karolinum, 2012. ISBN 9788024621289.
- POWELL, Lawrie W - SECKINGTON, Rebecca C - DEUGNIER, Yves. Haemochromatosis. *The Lancet*. 2016, y. 10045, vol. 388, p. 706-716, ISSN 0140-6736. DOI: 10.1016/s0140-6736(15)01315-x (<http://dx.doi.org/10.1016%2Fs0140-6736%2815%2901315-x>).

