Hemophilia

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Hemophilia is a disease with gonosomal recessive inheritance manifested by a disorder of blood clotting. The essence of the disease is either no or insufficient production of coagulation factors VIII, IX or XI.

The Gens for clotting factors VIII and IX are sex-linked chromosome X. The disorder occurs mostly in males, males (karyotype 46,XY) are hemizygousi. Women (karyotype 46,XX) – heterozygotes are carriers, the disease is rarely manifested in them (effect of lyonization). Of course, the disease manifests itself in recessive homozygotes females.

Types of Hemophilia

We distinguish two basic forms of hemophilia. The inheritance and manifestations of both types are the same. There is also a third type of hemophilia with autosomal inheritance.

- **Hemophilia A** (OMIM (https://www.omim.org/) 306700 (https://omim.org/entry/306700)) – the cause is a lack of coagulation factor VIII or a decrease in its functional activity below 10%. This can occur, for example, as a result of mutations in the gene for F8C (location Xq28). Mutation includes deletion, inversion, insertion or point mutation. Incidence: 1/10,000 boys.
- **Hemophilia B** (OMIM (https://www.omim.org/) 306900 (https://omim.org/entry/306900)) – this is a lack or reduction of the functional activity of coagulation factor IX, which is caused by a mutation of the F9 gene (location Xq27.1-q27.2). Incidence: 1/70,000 boys.
- **Hemophilia C** (OMIM (https://www.omim.org/) 612416 (https://omim.org/entry/612416)) – caused by deficiency of coagulation factor XI (gene F11, localization 4q35). Inheritance is autosomal recessive.

Disease Manifestations

- spontaneous bleeding (e.g. epistaxis)
- bleeding after minor trauma (formation of ecchymoses, hematomas)
- bleeding usually does not stop after compression
- bleeding into the joints = hemarthrosis (joints deformed, blood irritates the synovium * remodeling; tile appearance)
- bleeding into the GIT (manifestation of melena), URO (hematuria)

Treatment

The basis of the treatment of hemophilia is the injection of a clotting factor concentrate, which is absent in the blood of people with hemophilia. For each patient, the treatment is individual, depending on the severity of the disease and the current level of the clotting factor in the blood. Injection application can be performed at home.

In children with a severe form of hemophilia, prophylactic treatment is used, where the patient with hemophilia regularly applies a certain amount of clotting factor even when there are no signs of bleeding. Prophylactic treatment helps to prevent spontaneous bleeding and protects above all from permanent damage to the joints. The precipitation factor is usually applied 2x to 3x a week.

Spontaneous bleeding (bleeding episode) may occur in people with hemophilia who do not apply clotting factor regularly. This is most often manifested by severe pain in the affected joint. Then it is important to administer a dose of clotting factor as soon as possible to prevent permanent damage to the joint. That's why it's important to have clotting factor concentrate at home. The treatment of the bleeding episode continues in cooperation with the hematology center under the guidance of a specialist doctor.

Dose calculation: 1 unit/kg will increase fVIII by 2% and fIX by 1%

antifibrinolytics are used as supportive therapy (for bleeding from the oral cavity), and *desmopressin* is also used.

Links

Related Articles

- Hemostasis disorders: Hereditary coagulopathies • Acquired coagulopathies • Bleeding conditions (pediatrics) • Hemorrhagic diatheses (pathology)
- Hemostasis • Hemocoagulation • Blood coagulation test • Bleeding test
- X-linked inheritance

External links

- http://www.hemofilici.cz/cs/o-hemofilii
- https://www.hemofilie.cz/ – website for the public and people with hemophilia

References

- INDRÁK, Karel, et al. *Hematologie : VII. Vnitřní lékařství.* 1. edition. 2006. ISBN 80-7254-868-9.