

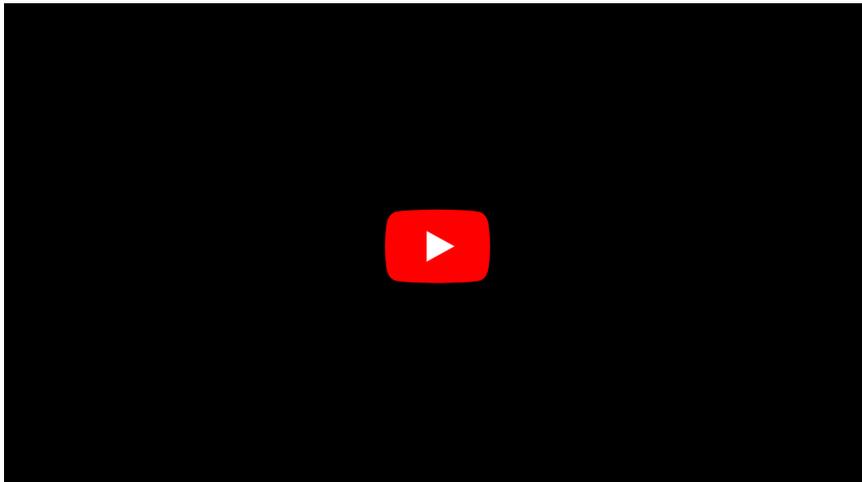
Sideroblastic anemia

Sideroblastic (also sideroblastic) anemias are disorders manifested by the finding of ring-shaped sideroblasts in hematopoietic tissue. Sideroblasts are erythrocyte precursors containing a ring formed by mitochondria filled with iron stores, which cannot be used for heme synthesis. We usually divide this disease into two groups: **congenital** and **acquired**. We classify it as **anemia from a disorder of erythrocyte hemoglobinization**, which also includes thalassemia and other hemoglobinopathies.

Sideroblastic anemia:



Sideroblastic anemia 2:



Congenital sideroblastic anemia

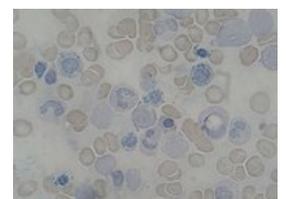
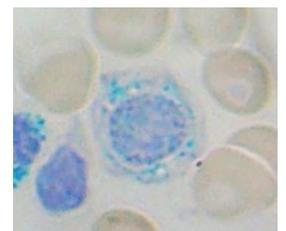
It is a rare disease with a **mutation of the ALAS2 gene**, i.e. the gene for *delta-aminolevulinic acid synthetase*, which is important for **heme synthesis**. It affects **men** due to the location of the gene on the X chromosome.

Main symptoms

The affected erythrocyte cell population is **hypochromic** and **microcytic**, but the erythrocytes produced by normal hematopoiesis are normocytic and normochromic. We find increased iron stores that increase **transferrin saturation** and **serum ferritin** values. **Ring-shaped sideroblasts** are found in the bone marrow.

Treatment

This type of anemia can sometimes be treated with **pyridoxine** at a dose of 200 to 300 mg/day, which is suitable for heme synthesis. Another variant is the administration of a **transfusion of erythrocytes**.



There is a high risk of iron overload when giving red blood cell transfusions because we are transfusing iron and already have pre-existing increased stores! We cannot exclude iron from the body.

Acquired sideroblastic anemia

The most common form of this disease includes **refractory anemia with proliferation of annular sideroblasts** (RARS) within the myelodysplastic syndrome. The cause of iron accumulation is a mutation in the enzyme system that utilizes iron in the mitochondria. It can rarely occur during **treatment with antituberculosis drugs** (*isoniazid*) or drugs such as *azathioprine*. They also occur in **lead poisoning**, when the enzyme ferrochetalase is inhibited - important for the synthesis of heme and also in rheumatoid arthritis and some tumors.

Summary video

<mediaplayer width='500' height='300'>https://www.youtube.com/watch?v=N_hXUkzppLY</mediaplayer>

Links

Related Articles

- Anemia from reduced erythrocyte production
- Anemia
- 5-aminolevulinic acid

Literature

- ČEŠKA, Richard. *Interna*. - edition. 2015. ISBN 9788073878955.
- NEČAS, Emanuel. *Patologická fyziologie orgánových systémů : Část I. 2.* vydání edition. Universita Karlova v Praze, 2013. 378s pp. ISBN 9788024617114.