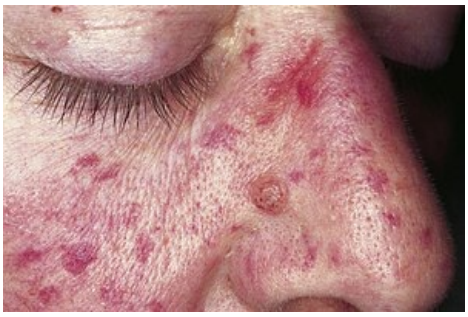
# Hereditary hemorrhagic telangiectasia

**Hereditary hemorrhagic telangiectasia** (lat. Telangiectasis hereditaria haemorrhagica; Rendu-Osler-Weber disease) is an autosomal dominant disease. It is one of the congenital vascular malformations and vascular hemorrhagic diathesis. The disease is characterized by an inherited structural abnormality of the walls of small vessels with subsequent dilatation, i.e. telangiectasia. Bleeding symptoms are caused by increased fragility of small vessels weakened in places, as well as the inability of vasoconstriction at the site of the bulge.



Morbus Osler - Hereditary hemorrhagic telangiectasia

## Etiopathogenesis

The basic deviation is the reduction of smooth muscle with a significant thinning of the vascular wall, sometimes extending to the endothelial layer. The pathogenesis is not precisely elucidated, a link with the HLAA2Bw17 haplotype is assumed.

## Manifestation

The disease can occur in both sexes and can affect all organs. Manifestations usually begin after puberty on the skin in the form of dark red nodules measuring 0.5 to 3 mm, which increase with age and also in number. They occur on the nasal mucosa, in the oral cavity, on the tongue, skin of the face, torso and limbs. Lesions of the internal organs are most common on the mucous membrane of the digestive system, in the respiratory tract, in the bladder, but they can also occur in the brain.

Bleeding is caused by mechanical damage to the elastic tube. The most common manifestation is *epistaxis*(90%), in which 50% of patients bleed into the gastrointestinal tract, microscopic hematuria is present in 18% of patients, and pulmonary arteriovenous shunts form in approximately 15% of patients, leading to hypoxemia. Bleeding into the brain is a serious complication. Repeated bleeding leads to posthemorrhagic anemia.

## Diagnostics

The disease is evidenced by the familial occurrence of telangiectasias on the skin and mucous membranes with recurrent bleeding. In the laboratory picture, the examination of hemocoagulation parameters is normal, a gradual accentuation of hypochromic microcytic sideropenic anemia with red-line hyperplasia in the bone marrow is detected.

## Treatment

Treatment is supportive (treatment of anemia by transfusion of erythrocytes, substitution treatment with iron preparations). Bleeding can be prevented by compression or tamponade. Skin lesions can be removed with a laser

## Prognosis

It depends on the location and size of the lesions and the intensity of the bleeding. As a rule, the course worsens with increasing age.

## Odkazy

### Zdroj


- DOBROTOVÁ, Miroslava. *Hematológia a transfuziológia : učebnica.* 1. edition. Grada ; Bratislava : Grada Slovakia, 2006. ISBN 80-8090-000-0.