Hereditary Hemorrhagic Telangiectasia Encyclopedia Article

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Hereditary Hemorrhagic Telangiectasia

Hereditary hemorrhagic telangiectasia (HHT) is an inherited condition characterized by abnormal blood vessels which are delicate and prone to bleeding. Hereditary hemorrhagic telangiectasia is also known as Rendu-Osler-Weber disease. Hereditary hemorrhagic telangiectasia, most often occurring as an autosomal dominant inherited disorder, affects 1 in 50,000 people. Twenty percent of the time, however, HHT occurs in individuals who have no family history of bleeding disorders. Spontaneous **mutation** is the most probable explanation for these sporadic cases of HHT

The term telangiectasia refers to a spot formed, usually on the skin, by a dilated capillary or terminal artery. In hereditary hemorrhagic telangiectasia, these spots occur because the blood vessel is fragile and bleeds easily. The bleeding may appear as small, red or reddish-violet spots on the face, lips, inside the mouth and nose or the tips of the fingers and toes. Other small telangiectasias may occur in the digestive tract.

Unlike hemopjilia,where bleeding is caused by an ineffective clotting mechanism in the blood, bleeding in hereditary hemorrhagic telangiectasia is caused by fragile blood vessels. However, like **hemophilia**, bleeding may be extensive and can occur without warning.

Recurrent nosebleeds are a nearly universal symptom in this condition. Usually the nosebleeds begin in childhood and become worse with age. The skin changes begin at puberty, and the condition becomes progressively worse until about 40 years of age, when it usually stabilizes.

There are few no specific treatment for hereditary hemorrhagic telangiectasia. The bleeding resulting from the condition can be stopped by applying compresses or direct pressure to the area. If necessary, a laser can be used to ablate the vessel. In severe cases, as in stroke, a leaking arterial vessel can often be plugged or covered with a graft from normal tissue.

In most people, recurrent bleeding results in an iron deficiency and it is usually necessary to take iron supplements.