About Hereditary Hemorrhagic Telangiectasia (HHT)

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KEY POINTS

- HHT is a genetic disorder in which blood vessels do not develop normally leading to bleeding that can be serious or life threatening.
- Frequent nosebleeds are the most common sign.
- Complication can vary widely, even among affected family members.
- Treatment to control bleeding and prevent complications may be needed.

What it is

HHT is a disorder in which some blood vessels do not develop properly. A person with HHT may form abnormal capillaries or abnormal capillary connections between the arteries and veins. Capillaries are tiny blood vessels that pass blood from arteries to veins. The abnormal blood vessels formed in HHT are often fragile and can burst, causing bleeding. Men, women, and children from all racial and ethnic groups can be affected by HHT and experience the problems associated with this disorder, some of which are serious and potentially life-threatening. Fortunately, if HHT is discovered early, effective treatments are available. However, there is no cure for HHT.

Signs

Nosebleeds are the most common sign of HHT, resulting from small abnormal blood vessels within the inside layer of the nose. Abnormal blood vessels in the skin can appear on the hands, fingertips, face, lips, lining of the mouth, and nose as delicate red or purplish spots that lighten briefly when touched. Bleeding within the stomach or intestines is another possible indicator of HHT that occurs because of abnormal blood vessels lining the digestive tract. Additional signs of HHT include abnormal artery-vein connections within the brain, lungs, and liver, which often do not display any warning signs before rupturing.

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Causes of HHT

HHT is a genetic disorder. Each person with HHT has a gene that is altered (mutated), which causes HHT. It takes only one gene with a mutation to cause HHT. Hundreds of possible mutations in several different genes have been linked to HHT.

Diagnosis

HHT can be diagnosed by performing genetic testing. Most people with HHT are first diagnosed by using clinical criteria (presence of signs and a history of signs in a parent, sibling, or child).

Complications and Treatments

The complications of HHT can vary widely, even among people affected by HHT in the same family. Complications and treatment of HHT depend on the parts of the body that are affected by this disorder. Treatment may include controlling bleeding and anemia and preventing complications from abnormal artery-vein connections in the lungs and brain.

Resources

- Cure HHT
- More than a Nosebleed: HHT
- Resource Library
- International HHT Guidelines

SOURCES

CONTENT SOURCE:
National Center on Birth Defects and Developmental Disabilities