Hereditary Hemorrhagic Telangiectasia

What is hereditary hemorrhagic telangiectasia?

Hereditary hemorrhagic telangiectasia (HHT; HEM-or-RAJ-ick tell-AN-jee-eck-TAY-zhuh) is a genetic disorder of the blood vessels. It is also called Osler-Weber-Rendu disease. People with HHT have abnormal blood vessels that tend to burst open and bleed.

Who gets it?

HHT affects men and women from all racial and ethnic groups. The abnormal gene comes from one parent who has HHT. If a parent has HHT, his or her children have a 50 percent chance of inheriting the gene and developing HHT. It is possible to have HHT with symptoms so mild that you don't notice them.

How can I tell if I have it?

HHT is diagnosed based on the signs and symptoms caused by abnormal vessels. These vessels have a greater chance of rupturing and bleeding compared with normal blood vessels. They can occur in the nose, skin, gastrointestinal tract, lungs, and brain.

HHT can be diagnosed with genetic tests. If the gene is found in one family member who is known to have HHT, other family members can be tested to see if they have HHT.

What are the symptoms?

About nine out of 10 people with HHT have nosebleeds that keep coming back. These can happen every day or only once in a while. The bleeding may be light or heavy.

Abnormal vessels in the stomach or intestines can cause black or bloody stools or anemia. In the lungs, they can cause shortness of breath and possibly strokes if they become large. There is a higher risk of bleeding during pregnancy. In the brain, abnormal vessels can be dangerous if they bleed. Often they don't cause warning symptoms, so anyone with known or suspected HHT should be screened. Some people with HHT have abnormal vessels in the liver, but these don't usually cause problems in most people.

No one with HHT has all of the signs and symptoms. Most symptoms vary, even within a family. A parent may have bad nosebleeds, but no abnormal vessels in an internal organ. Yet, his or her child may have a nosebleed only rarely but abnormal vessels in one or more internal organs. Doctors can't predict how likely someone is to have one of the hidden, internal abnormal vessels based on how many nosebleeds or skin telangiectasias (small red to purplish spots on the skin) they have.

How is it treated?

There is no way to stop the abnormal blood vessels from forming, but most can be treated if they cause problems. Using a humidifier or ointments in the nose can help nosebleeds. Laser therapy or other surgeries can also help. Abnormal blood vessels in the lungs or brain

Hereditary Hemorrhagic Telangiectasia *(continued)*

are treated by embolization, which closes off the blood vessel to keep it from rupturing. Telangiectasias of the skin can be treated with laser therapy if they bleed often.

Who should be screened for HHT?

Anyone with a family history of HHT should be checked for the disease. If someone in a family has the disease, other family members should see their doctor, who can help arrange for genetic testing. The symptoms of HHT can be mild, so even family members without symptoms should be screened.

Where can I get more information?

The HHT Foundation International
Telephone: 1-800-448-6389 or 1-410-357-9932
(outside the United States)
Web site: http://www.hht.org
E-mail: hhtinfo@hht.org

October 2010

Notes: