Hereditary Hemorrhagic Telangiectasia

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What is HHT?

Hereditary hemorrhagic telangiectasia (HHT) or Osler-Weber-Rendu syndrome is an inherited disorder characterized by malformations of blood vessels which potentially results in bleeding (hemorrhaging). The most common locations affected are in the nose, lungs, brain and liver. These malformations include *arteriovenous malformations* (AVMs) which are larger abnormal connections between arteries and veins, as well as *telangiectasias* which are small vascular malformations which cause small red lesions. HHT is categorized into 5 types depending on the genes implicated.

What are the signs and symptoms of HHT?

Signs and symptoms of HHT vary from person to person and depends on the specific gene that is altered. There is also variation in the age that symptoms begin to appear. The first symptom that patients normally experience are recurring nosebleeds or epistaxis. Nosebleeds normally occur because of the formation of telangiectasias in the mucous membranes that line the inside of the nose. These telangiectasias can also occur in the gastrointestinal tract which can cause bleeding resulting in bloody stools or vomit, but it may also not occur in some patients. Bleeding episodes may become more severe as patients age which may lead to low levels of iron in red blood cells (anemia). Anemia may cause chest pain, shortness of breath, or fatigue. Arteriovenous malformations occur typically in larger organs such as the lungs, liver, and brain. Symptoms of these depend on the size and location of the AVM, for example, if a person's lung is affected, they may experience shortness of breath.

What are the causes of HHT?

HHT is usually inherited as an autosomal dominant trait. This means that if one parent is affected, they will pass some form of the disease to all their children. There are 5 genes that have been identified as causing HHT.

How many people are affected by HHT?

The reported prevalence ranges between 1in 5000-8000 depending on the region. However, because some patients only experience mild symptoms, HHT is considered to be underdiagnosed and the true amount is higher.

How is HHT diagnosed?

HHT is often diagnosed by your doctor in the clinic using the Curaçao criteria, which includes identifying the presence of:

1. recurrent nosebleeds (epistaxis);
2. visible blood vessel malformations in areas such as the fingertips, lips, mouth and/or tongue (telangiectasia);
3. evidence of blood vessel malformations in internal organs (visceral lesions);
4. family history of HHT

Patients are diagnosed with HHT if they have 3 or more of the above features. Patients may also receive genetic testing, however this is only possible for 3 of the 5 causative genes, so typically the Curaçao criteria is used.

Imaging to locate AVMs in internal organs such as lungs, brain, and liver include:

- Ultrasound imaging: this can detect AVMs in the liver
- Magnetic Resonance Imaging (MRI): this is used to detect blood vessel abnormalities in the brain
- Bubble Study: this is used to detect abnormal blood flow in the lung
- CT scan: this may be used if there were abnormal results from the bubble study

Are there treatments for HHT?

As HHT is genetic, it cannot be cured however the symptoms can be managed. Management focuses on reducing chronic bleeding as well as screening and targeted treatments for internal organs that may be affected. Nosebleeds can be relieved with lubrication of the nostrils and application of pressure.

Medications that may be used to manage HHT include hormone-relate drugs, drugs that block blood vessel growth, drugs that slow the disintegration of clots (such as tranexamic acid). Chronic bleeding is usually from the nose and GI tract, and some patients may experience iron-deficiency anemia from this which may have to be treated depending on the severity.

There are surgical procedures that can be used to help manage symptoms in the nose by an ENT surgeon. These include:

- Ablation: This is a procedure that uses energy from lasers or a high-frequency electrical current to seal the abnormal vessels that cause nosebleeds. This is a temporary measure and often has to be repeated
- Skin graft: This involves taking a skin graft from another part of your body and implanting it in the inside of your nose

There are other surgical procedures for symptoms affecting the lungs, brain, and liver.

Where can I learn more?
Rare Diseases (.org)
Mayo Clinic

NOTE: The SPSC has created a registry for all patients in British Columbia who have HHT in order to better understand HHT and provide optimal care for patients. If you or someone you know is interested in participating, please contact mailto:spschht@gmail.com.