CONDITION

Hereditary hemorrhagic telangiectasia

An inherited condition that causes fragile blood vessels and abnormal connections between arteries and veins.

WHAT IT IS
Inherited condition causing fragile vessels and abnormal vessel connections

BODY SYSTEM
Blood vessels throughout the body

MAIN SYMPTOMS
Nosebleeds, red skin spots, anemia, shortness of breath

WHAT CAUSES IT
Inherited gene changes affecting blood vessel development

HOW IT IS DIAGNOSED
Clinical criteria, genetic testing, and scans for abnormal vessels

MAIN TREATMENT
Bleeding control, iron replacement, and abnormal vessel treatment

CAN IT BE CURED?
No - blood vessel changes remain lifelong

IS IT AN EMERGENCY?
Sometimes - severe bleeding, stroke signs, or breathing trouble

LONG-TERM OUTLOOK
Often manageable with screening and targeted treatment

Overview

Hereditary hemorrhagic telangiectasia, usually called HHT, is an inherited condition affecting how blood vessels develop. It is also known as Osler-Weber-Rendu syndrome.

HHT can cause small, fragile blood vessels called telangiectasias. These may appear as tiny red or purple spots on the lips, tongue, face, hands, or inside the nose and digestive tract. They can bleed easily.

HHT may also cause larger abnormal connections between arteries and veins. These are called arteriovenous malformations, or AVMs. An AVM allows blood to pass directly from an artery into a vein without moving through the usual network of tiny vessels.

AVMs can occur in the lungs, brain, liver, digestive tract, and other areas. Some never cause

symptoms. Others can affect oxygen levels, strain the heart, or allow a blood clot or infection to reach the brain.

HHT does not mean that your blood cannot clot normally. Bleeding happens mainly because affected blood vessels are fragile or formed abnormally.

The condition lasts for life, but its effects differ greatly between people, including relatives with the same gene change. Screening matters because treatable AVMs may be present before symptoms appear.

Symptoms

Frequent nosebleeds are the most common sign. They often begin in childhood or adolescence but may start later. Nosebleeds can range from occasional spotting to repeated bleeding that causes iron deficiency.

Telangiectasias usually become more noticeable with age. They may look like pinpoint red marks or fine, branching blood vessels. Common locations include the lips, tongue, inside the mouth, fingertips, face, and nose. Pressing on a spot may briefly make its colour fade.

Blood loss from the nose or digestive tract can lead to iron-deficiency anemia, meaning there are not enough healthy red blood cells because the body lacks iron. Symptoms may include:

Lung AVMs may cause shortness of breath, low blood oxygen, chest discomfort, or coughing up blood. Many cause no obvious symptoms. Even a silent lung AVM can allow a clot or bacteria to bypass the lungs' filtering system and reach the brain.

Brain AVMs are also often silent. When symptoms occur, they may include seizures, persistent headaches, weakness, problems with speech, or bleeding in the brain.

Liver AVMs frequently cause no symptoms. Larger or more extensive changes can make the heart work harder and may contribute to breathlessness, swelling, abnormal liver tests, or problems with bile flow.

- Tiredness, weakness, or reduced exercise tolerance
- Shortness of breath, dizziness, or headaches
- Pale skin, a racing heartbeat, or difficulty concentrating
- Dark or bloody stools from digestive tract bleeding

Causes and risk factors

HHT is caused by a harmful change in a gene involved in blood vessel development. Changes in the ENG and ACVRL1 genes cause most identified cases. Changes in SMAD4 and some other genes account for a smaller number.

HHT is usually inherited in an autosomal dominant pattern. This means that a person with HHT has a 1 in 2 chance of passing the gene change to each child. The chance is the same for every pregnancy.

A child can be affected even if the parent has mild symptoms. Likewise, one person may have frequent bleeding while another relative with the same gene change has few problems.

A person occasionally has HHT because of a new gene change rather than inheriting it. Having no known family history therefore does not completely rule it out.

Some people with a SMAD4 gene change have both HHT and juvenile polyposis syndrome. Juvenile polyposis causes growths called polyps in the digestive tract and requires additional cancer surveillance.

Diagnosis

HHT may be diagnosed using a combination of medical history, physical findings, family history, genetic testing, and imaging.

Clinicians often use the Curaçao criteria. These look for four features: repeated spontaneous nosebleeds, characteristic telangiectasias, AVMs in internal organs, and a first-degree relative with HHT. Meeting three or four criteria makes HHT clinically definite. Two suggests possible HHT. These criteria may be less reliable in children because signs often appear gradually.

A genetic blood or saliva test can look for a gene change linked to HHT. If a specific change is already known in your family, testing can usually show whether you inherited it. When no gene change is found, HHT may still be possible because current tests do not identify every cause.

Screening looks for internal AVMs before they cause complications. Tests may include:

Digestive tract tests, such as endoscopy, may be used when anemia is not explained by nosebleeds or when there are signs of digestive bleeding.

- Contrast echocardiography, followed by a chest CT when needed, for lung AVMs
- MRI of the brain for brain blood vessel abnormalities
- Ultrasound, CT, or MRI when liver involvement needs assessment
- Blood counts and ferritin tests for anemia and low iron stores

Treatment and management

There is no treatment that removes the inherited tendency to develop abnormal blood vessels. Care focuses on limiting bleeding, replacing lost iron, treating important AVMs, and monitoring for complications.

Simple nose care may reduce mild nosebleeds. This can include saline sprays or gels and humidifying dry air. If bleeding remains troublesome, treatment may involve laser therapy, cautery, or other procedures that seal fragile vessels. Severe bleeding may require more extensive nasal surgery.

Doctors may consider medicines that help stabilise clots or reduce the growth of abnormal vessels. These include antifibrinolytic medicines and, in selected severe cases, medicines that block blood vessel growth, such as bevacizumab. Their benefits and risks require specialist assessment.

Iron deficiency is treated with iron taken by mouth or given into a vein. Blood transfusions may be needed when anemia is severe or bleeding is heavy. Ongoing blood tests help show whether treatment is replacing iron as quickly as it is being lost.

A significant lung AVM is commonly treated by embolization. During this procedure, a specialist passes a thin tube through a blood vessel and places a plug or coil to close the abnormal connection. Follow-up imaging is needed because treated vessels can reopen and new AVMs can appear.

Treatment of a brain AVM is individualised. Options may include observation, surgery, embolization, or focused radiation treatment. The safest choice depends on its size, position, structure, and bleeding risk.

Liver AVMs that cause no problems usually do not need direct treatment. Complications may be managed with heart or liver treatments. Liver transplantation is an option for a small number of people with severe, otherwise unmanageable complications.

Blood-thinning medicines are not automatically forbidden in HHT. If one is needed for another condition, the bleeding risk and expected benefit should be assessed carefully rather than stopping or avoiding it without medical advice.

Complications

Repeated bleeding may cause long-term iron deficiency and anemia. This can affect energy, concentration, exercise, sleep, and quality of life.

An untreated lung AVM can allow a blood clot or bacteria to pass into the arteries and reach

the brain. Possible complications include stroke and brain abscess, which is a pocket of infection in the brain. Lung AVMs can also bleed, although this is less common.

Brain AVMs can sometimes bleed into or around the brain. The risk is not the same for every AVM, which is why assessment by an experienced team is important.

Extensive liver involvement can force the heart to pump much more blood than usual. Over time, this may lead to high-output heart failure. HHT-related liver vessel changes can also cause abnormal pressure in liver blood vessels or problems with the bile ducts.

Pregnancy increases blood flow and may increase the risk of complications from untreated lung AVMs. Planning before pregnancy allows time for screening and treatment where appropriate.

Prevention

HHT itself cannot currently be prevented because it is inherited. Lifestyle choices do not cause the underlying blood vessel changes.

Genetic counselling can help you understand inheritance, testing, and reproductive options. It can also help identify relatives who may benefit from screening, including those who feel well.

Some complications can be prevented or found early. Regular iron checks can detect blood loss before anemia becomes severe. Screening and treating suitable lung AVMs can reduce the risk of stroke, brain infection, and bleeding.

People with lung AVMs may need antibiotics like amoxicillin or doxycycline before certain dental or medical procedures that can place bacteria in the bloodstream. They should also take care to prevent air from entering intravenous lines. Scuba diving may carry extra risk. These precautions should be based on advice from an HHT or respiratory specialist.

Living with it and what to expect

HHT changes over time. Nosebleeds and visible telangiectasias often become more frequent with age, while internal AVMs may remain stable, enlarge, or occasionally develop later. A symptom-free period does not always mean that screening is no longer needed.

Many people live active lives with planned monitoring and treatment. Day-to-day effects are often linked to nosebleeds, iron deficiency, appointments, or recovery from procedures. Keeping track of bleeding patterns and blood test results can help show when management needs to change.

Follow-up may include blood counts, iron tests, checks after AVM treatment, and repeat screening at intervals based on age and previous findings. Children with HHT or a known family gene change need age-appropriate assessment because they may have internal AVMs before developing typical nosebleeds or skin spots.

Pregnancy planning is particularly important. Screening for lung AVMs is ideally completed before pregnancy. If pregnancy occurs before screening, an obstetric and HHT team can decide which assessments are appropriate.

The condition can affect relatives differently. Family members should not assume that mild symptoms in one person mean everyone has a low risk of internal AVMs.

When to get medical help

Arrange routine medical follow-up if you have repeated nosebleeds, visible telangiectasias, unexplained low iron, or a close relative with HHT. Medical review is also appropriate if established symptoms become more frequent or begin affecting sleep, work, school, or exercise.

Seek prompt medical advice for black or bloody stools, worsening breathlessness, repeated dizziness, new swelling, coughing up a small amount of blood, or symptoms suggesting anemia.

Get emergency help for:

These symptoms do not always result from HHT, but they require urgent assessment.

- A nosebleed or other bleeding that is heavy or will not stop
- Coughing or vomiting a large amount of blood
- Sudden weakness, facial drooping, speech difficulty, or loss of vision
- A first seizure, fainting, confusion, or a sudden severe headache
- Severe breathing difficulty, chest pain, or blue-grey lips
- Fever with severe headache, neurological symptoms, or marked drowsiness

Common questions

Is HHT cancer?

No. HHT is a condition affecting blood vessel development, not a cancer. People with a SMAD4 gene change may also have juvenile polyposis syndrome, which carries separate digestive tract cancer risks.

Can I have HHT without nosebleeds?

Yes. Nosebleeds are common, but they may not begin until later, especially in children. Some people are identified through family genetic testing or screening for an internal AVM.

Will my child inherit HHT?

If you have a disease-causing HHT gene change, each child has a 1 in 2 chance of inheriting it. A genetic counsellor can explain testing and reproductive options without assuming what choice is right for your family.

Are the red spots on my skin dangerous?

Skin and mouth telangiectasias are usually more troublesome than dangerous. They may bleed after minor injury. Their number does not reliably show whether you have AVMs in the lungs, brain, or liver.

Can HHT make me tired all the time?

It can. Repeated blood loss may lower iron stores and eventually cause anemia. Iron deficiency can cause fatigue even before the red blood cell count becomes clearly abnormal, so ferritin testing may be useful.

Can I exercise if I have HHT?

Many people can exercise normally. Safe activity may be affected by anemia, low oxygen, an untreated AVM, or heart complications. New breathlessness, chest pain, fainting, or a major decline in exercise ability needs medical assessment.

Sources

- Faughnan ME, Mager JJ, Hetts SW, et al. Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. Annals of Internal Medicine . 2020;173(12):989-1001.
- McDonald J, Pyeritz RE. Hereditary Hemorrhagic Telangiectasia. GeneReviews . University of Washington, Seattle; updated November 24, 2021.
- Shovlin CL. Pulmonary arteriovenous malformations. American Journal of Respiratory and Critical Care Medicine . 2014;190(11):1217-1228.
- National Health Service. Hereditary haemorrhagic telangiectasia.
- Cure HHT. International HHT Guidelines: patient and clinician guidance on diagnosis, bleeding, anemia, and organ screening.