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Hereditary hemorrhagic telangiectasia

Hereditary hemorrhagic telangiectasia (HHT) is an inherited disorder of the blood vessels that can cause excessive bleeding.

Causes

HHT is passed down through families in an autosomal dominant pattern. This means the abnormal gene is needed from only one parent in order to inherit the disease.

Scientists have identified four genes involved in this condition. All of these genes appear to be important for blood vessels to develop properly. A mutation in any one of these genes is responsible for HHT.

Symptoms

People with HHT can develop abnormal blood vessels in several areas of the body. These vessels are called arteriovenous malformations (AVMs).

If they are on the skin, they are called telangiectasias. The most common sites include the lips, tongue, ears, and fingers. The abnormal blood vessels can also develop in the brain, lungs, liver, intestines, or other areas.

Symptoms of this syndrome include:

- Frequent nosebleeds in children
- Bleeding in the gastrointestinal tract (GI), including loss of blood in the stool, or dark or black stools
- Seizures or unexplained, small strokes (from bleeding into the brain)
- Shortness of breath
- Enlarged liver
- Heart failure
- Anemia caused by low iron

Exams and Tests

Your health care provider will perform a physical examination and ask about your symptoms. An experienced provider can detect telangiectases during a physical examination. There is often a family history of this condition.

Tests include:

- Blood gas tests
- Blood tests
- Imaging test of the heart called an echocardiogram
- Endoscopy, which uses a tiny camera attached to a thin tube to look inside your body
- MRI to detect AVMs in the brain
- CT or ultrasound scans to detect AVMs in the liver

Genetic testing is available to look for changes in genes associated with this syndrome.

Treatment

Treatments may include:

- Surgery to treat bleeding in some areas
- Electrocautery (heating tissue with electricity) or laser surgery to treat frequent or heavy nosebleeds
- Endovascular embolization (injecting a substance through a thin tube) to treat abnormal blood vessels in the brain and other parts of the body

Some people respond to estrogen therapy, which can reduce bleeding episodes. Iron may also be given if there is a lot of blood loss, leading to anemia. Avoid taking blood-thinning medicines. Some medicines that affect blood vessel development are being studied as possible future treatments.

Some people may need to take antibiotics before having dental work or surgery. People with lung AVMs should avoid scuba diving to prevent decompression sickness (the bends). Ask your provider what other precautions you should take.

Support Groups

More information and support for people with HHT disorder and their families can be found at:

- Cure HHT -- curehht.org/ [<https://curehht.org/>]
- National Organization for Rare Disorders -- rarediseases.org/rare-diseases/hereditary-hemorrhagic-telangiectasia/ [<https://rarediseases.org/rare-diseases/hereditary-hemorrhagic-telangiectasia/>]

Outlook (Prognosis)

People with this syndrome can live a completely normal lifespan, depending on where in the body the AVMs are located.

Possible Complications

These complications can occur:

- Heart failure
- High blood pressure in the lungs (pulmonary hypertension)
- Internal bleeding
- Shortness of breath

- Stroke

When to Contact a Medical Professional

Contact your provider if you or your child has frequent nose bleeds or other signs of this disease.

Prevention

Genetic counseling is recommended for couples who want to have children and who have a family history of HHT. If you have this condition, medical treatments can prevent certain types of strokes and heart failure.

Alternative Names

HHT; Osler-Weber-Rendu syndrome; Osler-Weber-Rendu disease; Rendu-Osler-Weber syndrome

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