

# Diagnosis and misdiagnosis of HHT

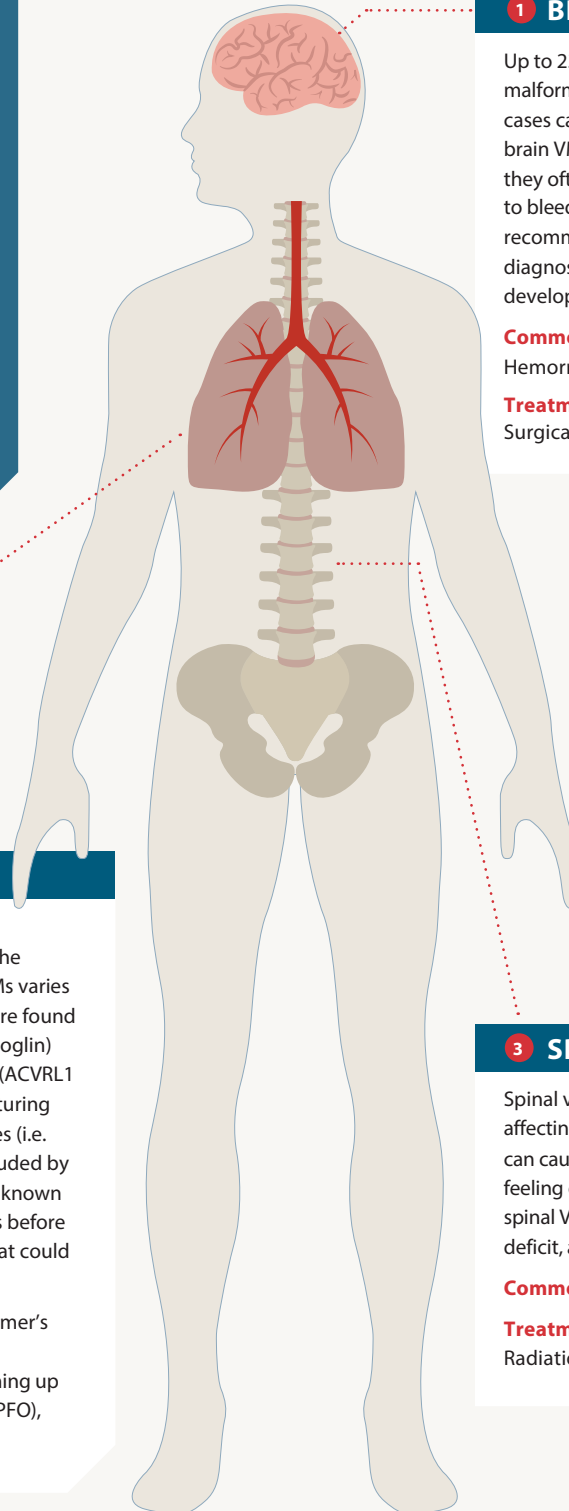
Hereditary Hemorrhagic Telangiectasia (HHT), also sometimes referred to as Osler-Weber-Rendu (OWR), is a genetic disorder of the blood vessels affecting approximately 1 in 5,000 people totaling 1.4 million worldwide. While HHT affects specific areas or organs of the body, it's important to understand the potential impact on other aspects of life as well. To ensure the best possible health for you or your loved ones, remember to consider how HHT might impact or cause the following: aging, pregnancy, iron deficiency, genetic testing, anemia.

As HHT symptoms are often mistaken for other conditions, knowing the common misdiagnoses can lead to faster identification and significant improvements in quality of life.

## AFFECTED ORGANS

HHT does not affect every individual in the same way. Some people may have symptoms in several different parts of the body, while some may show hardly any symptoms at all.

- 1 BRAIN
- 2 LUNGS
- 3 SPINE
- 4 NOSE
- 5 GASTROINTESTINAL (GI)
- 6 LIVER
- 7 SKIN



## 1 BRAIN

Up to 23% of people with HHT will harbor vascular malformations (VMs) in the brain, and in most cases can be successfully treated. If left untreated, brain VMs can be disabling or deadly, and because they often have no symptoms prior to bleeding, the HHT International Guidelines recommend brain screening for anyone diagnosed with HHT, even infants and during developmental growth periods until age 21.

**Common Misdiagnosis:** Aneurysm, Cerebral Hemorrhage, Epilepsy, Abscess, Migraine, Stroke

**Treatment:** Embolization, Radiation, Surgical Removal

## 2 LUNGS

At least 40% of people with HHT have arteriovenous malformations (AVMs) in the lungs. The prevalence of pulmonary AVMs varies with the type of HHT: pulmonary AVMs are found in about 50% of patients with HHT1 (Endoglin) and in about 10% of patients with HHT2 (ACVRL1 or ALK1). PAVMs are at higher risk of rupturing when blood volume in the body increases (i.e. during pregnancy). Until PAVMs are excluded by testing, a person over the age of 10 with known or suspected HHT should take antibiotics before all dental work or medical procedures that could introduce bacteria to the blood stream.

**Common Misdiagnosis:** Asthma, Alzheimer's Disease, Lung Disease, Tumor, Stroke, Polycythemia, shortness of breath, coughing up blood, Infertility, Patent Foramen Ovale (PFO), Migraines, Brain Abscess, Stroke

**Treatment:** Endovascular Embolization

## 3 SPINE

Spinal vascular malformations (VMs) are rare, affecting only about 0.5% of HHT patients. They can cause back pain and sometimes lead to loss of feeling or mobility in an arm or leg. If untreated, spinal VMs can lead to further motor or sensory deficit, aneurysm, and high blood pressure.

**Common Misdiagnosis:** Scoliosis, Stroke

**Treatment:** Endovascular Embolization, surgery, Radiation Therapy

#### 4 NOSE

Recurring nosebleeds (Epistaxis) affect about 90% of people with HHT and are the most common symptom of the disease. Epistaxis is caused when telangiectasias in the nose rupture, leading to nosebleeds of varying severity, length, and frequency. The average age of onset for epistaxis is 12 years with nearly 100% affected by age 40. 10% of HHT patients will not have nose bleeds even though they may have 'silent manifestations' that could be potentially dangerous.

**Common Misdiagnosis:** Allergies, Drug Addiction, Hemophilia, Von Willebrand Disease

**Treatment:** Over the counter sprays/gels, moisture & humidification, Drug Therapy, Sclerotherapy, Surgery, Coblation

#### 6 LIVER

Vascular malformations (VMs) occur in the liver of 41-74% of people with HHT, but it is estimated that only about 8-14% of those affected with liver VMs are symptomatic at baseline. Untreated liver VMs can lead to heart failure, typically in combination with aging, when the heart has been overworked for years by pumping extra blood through the low-resistance pathway of the VM.

**Common Misdiagnosis:** Cirrhosis, Heart Failure, shortness of breath

**Treatment:** Intravenous blood vessel inhibitor drugs, additional therapeutic management, transplant

#### 5 GI TRACT

GI bleeding develops in approximately 30% of HHT patients. Most symptomatic patients have GI telangiectasias in the stomach (46-75%), small bowel (56-91%) and up to 30% also have telangiectasias in the colon. Bleeding can range from mild to severe and can occur anywhere in the GI system including the esophagus and large intestine. The most common sites are the stomach and upper portion of the small intestine.

**Common Misdiagnosis:** Black stool, Angiodysplasia, unexplained Anemia, Cancer

**Treatment:** Heater Probe, Bicap, Argon Photocoagulation, Laser, Intravenous blood vessel inhibitor drugs, Oral Medication

#### 7 SKIN

95% of people with HHT have telangiectasias on the skin of the hands, face, and mouth, although most patients report the appearance of telangiectasia of the mouth, face or hands 5-30 years after the onset of nose bleeds, most commonly during the third decade of life. Telangiectasias appear as tiny red or purple spots between the size of a pinpoint and pinhead. Rupture and bleeding of telangiectasias is common in the nose but less so in the mouth, face, or hands.

**Common Misdiagnosis:** Birthmarks, Cherry Angioma

**Treatment:** Laser



Cure HHT offers resources for managing these potential aspects of an HHT diagnosis. Fact Sheets and other resources for understanding and managing the different manifestations of HHT are available at: [curehht.org/resource-library/](http://curehht.org/resource-library/)

Visit us at [CureHHT.org](http://CureHHT.org), call us at (410) 357-9932, or find us on

