HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)
Osler-Weber-Rendu Syndrome

HHT is an inherited, genetic disorder that causes malformed blood vessels in multiple organs of the body, and typically begins with nosebleeds during childhood.

Approximately 1 in 5,000 people are affected by HHT, totaling 1.4 million worldwide. However, HHT is often misdiagnosed and its symptoms are often mistaken for other conditions. The disease affects men, women and children at any age, as well as all ethnicities and races.

DIAGNOSIS

It’s important to recognize that HHT does not affect every individual in the same way. Some people have symptoms in several different parts of the body (detailed below), while some may show hardly any symptoms at all.

AFFECTED ORGANS

- **NOSEBLEEDS**: 90% of those with HHT develop nosebleeds
- **BRAIN AVMs**: are found in 5–20% of people with HHT
- **LUNG AVMs**: are found in 3–7% of people with HHT
- **LIVER AVMs**: are found in 3–7% of people with HHT
- **TELANGIECTASIAS**: 95% of those with HHT have telangiectasia on skin of hands, face and mouth
- **STOMACH INTESTINES**: 80% of those with HHT have telangiectasia on stomach or intestines

COMMON MISDIAGNOSES

- **SKIN**: Birthmarks or cherry angioma
- **NOSE**: Allergies, drug addiction, hemophilia, or Von Willebrand Disease
- **BRAIN**: Aneurysm, cerebral hemorrhage, epilepsy, abscess, migraine, or stroke
- **LUNGS**: Asthma, Alzheimer’s disease, lung disease, tumor, stroke, Polycythemia, Patent Foramen Ovale (PFO)
- **SPINE**: Scoliosis or stroke
- **LIVER**: Cirrhosis, heart failure, shortness of breath
- **GI TRACT**: Angiodysplasia, anemia, black stool or cancer

visit www.curehht.org for more info.

Cure HHT, P.O. Box 329, Monkton, MD 21111 | (410) 357-9932
**Diagnostic Criteria**

- Recurrent and spontaneous nosebleeds (epistaxis), which may be mild to severe.
- Multiple telangiectasias on the skin of the hands, lips or face, or inside the nose or mouth.
- Arteriovenous malformations (AVMs) or telangiectasias in the internal organs including the lungs, brain, liver, intestines, stomach, and spinal cord.
- A family history (i.e., first-degree relative such as brother, sister, parent or child) who meets these same criteria for definite HHT or has been genetically diagnosed.

*Definite HHT = 3+ criteria; Possible HHT = 2 criteria

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**ICD-10 / Insurance Codes**

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Code</th>
<th>Code (for complications)</th>
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<tbody>
<tr>
<td>HHT</td>
<td>178.0</td>
<td>D50.D50.0</td>
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<tr>
<td>Cerebral AVM</td>
<td>Q28.2</td>
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<td>Colon AVM</td>
<td>K55.21</td>
<td>537.83</td>
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<td>Epistaxis</td>
<td>R04.0</td>
<td>448.9,178.8</td>
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<tr>
<td>GI or Liver AVM</td>
<td>Q27.33</td>
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</tbody>
</table>

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**Complications & Statistics**

- Untreated AVMs can cause strokes, seizures, brain abscesses, numbness of the arms or legs, heart failure and in the worst of cases, death.
- 50% of HHT patients suffer from anemia and iron deficiency, which causes symptoms such as fatigue and migraines.
- Dental care: The direct connection of blood vessels and lack of capillary filtration in the lungs places patients at-risk to develop a brain abscess if bacteria enters the blood stream. 5-10% of patients with pulmonary AVMs develop a brain abscess. This may be preventable with the administration of a single dose of prophylactic antibiotics approx. one hour prior to dental work.

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**RESOURCES**

- Locate HHT Treatment Centers of Excellence (CoEs)  
  North America & Int'l: curehht.org/hht-treatment-centers
- Access the free Cure HHT Resource Library  
  Review Clinical Guidelines: curehht.org/resource-library
- Review Physician Directory  
  Add yourself or find a local doctor: directory.curehht.org