

How to Access Genetic Testing

Genetic testing is typically ordered by a genetic counselor, medical geneticist or a health professional with specific expertise regarding hereditary conditions and genetic testing.

These specialists will know the most appropriate test to order, and which lab to use based on insurance and other factors. Most HHT Centers of Excellence and major medical centers have genetic counselors.

Genetic Counseling Resources



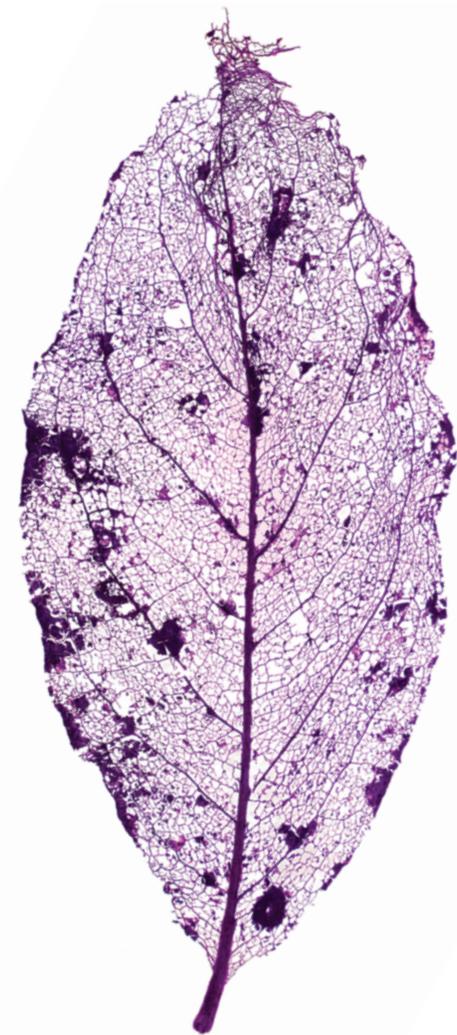
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[curehht.org/understanding-hht/
diagnosis-treatment/genetic-testing-hht](http://curehht.org/understanding-hht/diagnosis-treatment/genetic-testing-hht)



[impactgenetics.com/testing-services/
hereditary-hemorrhagic-telangiectasia/
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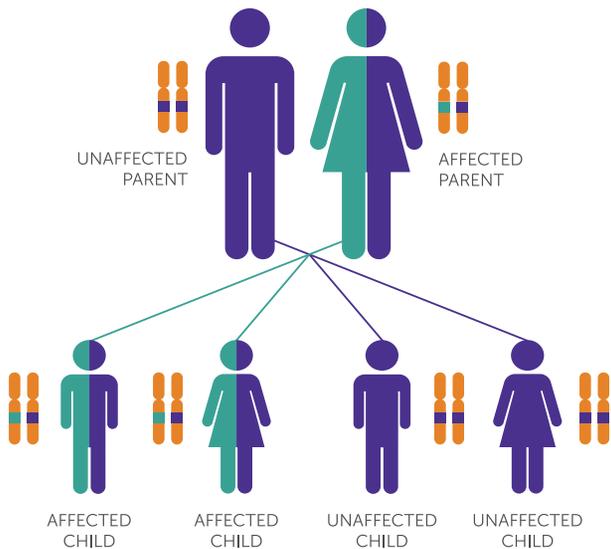
Genetic Testing & HHT

HEREDITARY HEMORRHAGIC
TELANGIECTASIA

What Causes HHT?

HHT is a relatively common inherited genetic condition affecting about 1 in 5000 people. It is caused by alterations in genes that help maintain normal blood vessel formation. These alterations lead to blood vessel abnormalities in the lung, brain, liver, gastrointestinal (GI) tract, skin and nose.

Men and women are equally affected. A person that has HHT has a 50% chance of passing this condition on to their children.



Forms of HHT

HHT is most commonly caused by alterations in the *ENG* or *ACVRL1* gene.

Alterations in a third gene, *SMAD4*, can lead to a condition called Juvenile Polyposis HHT syndrome (JP-HHT). Individuals with this condition have HHT symptoms as well as specific GI polyps called juvenile polyps and are at an increased risk for stomach and intestinal cancer.

Diagnosing HHT

Patients with HHT can be diagnosed on the basis of their clinical signs and symptoms, according to established clinical diagnostic criteria known as the Curaçao Criteria.

HHT can be difficult to diagnose since the symptoms vary among individuals, from very mild to severe, and can be age dependent. Children with HHT may not have any symptoms. There also tends to be a delay in diagnosis of 15-30 years from the onset of first symptoms due to lack of disease awareness in the medical community.

Why is Genetic Testing Important and How are Results Used?

Genetic testing is the only way to diagnose HHT for a person who is at risk for HHT but who does not meet the clinical diagnostic criteria.

The first step is to identify the family's HHT genetic alteration (variant). This is accomplished by testing a clinically diagnosed family member.

Once the HHT-causing genetic variant is found, any family member who is at risk for the condition, (even a distant blood relative), can have genetic testing for their family variant to determine whether or not they have HHT.

Only family members with the HHT causing variant need HHT screening for the blood vessel abnormalities associated with the condition.

Determining which gene is responsible for HHT in a family can affect medical management and care. For example, a patient with HHT caused by an alteration in the *SMAD4* gene is at risk for gastrointestinal GI polyps and should be managed accordingly.

The Importance of Genetic Counseling

Genetic counseling enables patients to make educated decisions regarding genetic testing.

Genetic counselors will:

- Obtain a complete family history, detailed medical history, and review the most appropriate genetic testing strategy for patients and families.
- Discuss the risks, benefits and limitations of testing as well as the implications of genetic results for the individual patient and their family members.

Genetic counselors are uniquely qualified to help patients explore the psychological impact and medical implications of genetic testing.

