

# FAMILY TIES: GENETIC TESTING



Cure HHT recommends all individuals and families who meet the diagnostic criteria for HHT to visit an HHT Center of Excellence for genetic testing by experts who understand the complexities and limitations of the process.

FACTSHEET  
**FS**

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## SOME IMPORTANT FACTS TO REMEMBER ABOUT HHT ARE:

As a hereditary disease, HHT has the potential to devastate families if not identified and treated.

Some of the most easily identifiable symptoms of HHT do not manifest until adolescence.

Throughout the diagnosis process, it is important to remember that HHT does not define you.

With Cure HHT's help, you can empower yourself with the knowledge and resources required to advocate for yourself and seize control of your life.



The Cornerstone of  
the HHT Community

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## WHAT ARE THE DIAGNOSTIC CRITERIA FOR HHT?

Individuals with definite HHT will exhibit three or more of the following characteristics.

- › Spontaneous and recurrent nosebleeds
- › Telangiectasias at characteristic sites
- › Organ manifestations: pulmonary, gastrointestinal, cerebral, hepatic AVM
- › Affected first degree relative

## WHY SHOULD I VISIT AN HHT CENTER FOR GENETIC TESTING?

As a hereditary disease, HHT has the potential to devastate families if not identified and treated. Centers of Excellence provide an integrated and multidisciplinary approach to HHT. Experts can provide accurate diagnosis as well as screenings for AVMs related to HHT, and you and your family will receive state of the art treatment. Centers of Excellence also offer the opportunity to participate in clinical research studies and provide patients with genetic counseling and DNA testing.

Some of the most easily identifiable symptoms of HHT do not manifest until adolescence. As a result, it may appear in childhood as though an individual does not have HHT. Anyone with a parent or sibling with HHT is considered “at-risk” and screening for brain and lung AVMs is strongly recommended.

## WHAT IS GENETIC TESTING?

There are three types of genetic testing used to screen for HHT.

- › Sequencing of genes, which involves looking at the precise sequence of building blocks in the DNA sample to check for abnormalities.
- › Targeted Sequencing examines whether one particular mutation previously identified within another family member is present or absent.
- › Deletion of Duplication testing checks to see whether a portion of a gene is missing or duplicated.

## WHAT ARE THE POSSIBLE RESULTS OF GENETIC TESTING FOR HHT?

A full gene analysis for the HHT genes is generally done for the first person tested in the family, while others subsequently tested will receive a “single mutation” analysis.

There are three potential results of the full gene analysis.

- › Positive for a “deleterious” or HHT causing mutation. This means that a lab test revealed a mutation in one or more of the HHT associated genes, and that the mutation is thought to cause HHT. This result would confirm that the person tested has HHT, and would make it possible to test other people in the same family for this precise mutation and determine whether or not they have HHT.
- › A negative result means the lab test did not find a mutation in an HHT-associated genes. A negative result could be indicative of a number of different things. First, the person tested may not have HHT. Second, the person tested does have HHT but carries the mutation in an HHT-associated gene that has yet to be identified. Third, a person might have a mutation in one of the tested genes, but the current testing techniques were not able to pick up the mutation.
- › If a test result reveals a variant of uncertain significance, then the lab found a change in an HHT-associated gene but cannot predict whether or not it causes HHT.

There are two possible results when single-mutation analysis for the HHT-associated gene has been identified in the family.

- › Positive. This person carries the HHT gene mutation present in the family, and thus has HHT.
- › Negative. This person does not carry the HHT gene mutation present in the family and did not inherit HHT.

HHT requires your attention and treatment, but it does not have to take over. It is crucial to remember throughout the process of diagnosis that this disease will not define you. The goal of Cure HHT is to empower you with the knowledge and resources required to advocate for yourself and seize control of your life.

**DISCLAIMER:** Even if you have already received genetic testing through a counselor or geneticist, we strongly encourage you to visit an HHT Center of Excellence. Our Centers have all been approved by the Foundation to provide integrated and comprehensive care by a handpicked team of experts.



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