Genetic Counseling and Testing for HHT

FREQUENTLY ASKED QUESTIONS

How is HHT inherited?
HHT is an autosomal dominant genetic condition. That means if your parent has HHT, you have a one in two (50%) chance of also inheriting HHT. If you have HHT, each of your children has a one in two (50%) chance of inheriting HHT. Someone cannot pass on HHT if they do not have HHT themselves. Specific signs and symptoms for each person with HHT may vary.

Who should have genetic testing for HHT?
HHT is a clinical diagnosis and does not need genetic testing to confirm. However, it is caused by a genetic mutation – a spelling change in one of 5 genes. Screening for blood vessel differences (arteriovenous malformations – AVMs) in lungs, brain and liver is recommended for first-degree relatives of someone who has HHT. A sibling or child may want to know if they have the genetic mutation for HHT or not, through genetic testing.

To find the genetic mutation causing HHT in your family, it is best to order genetic testing on someone who is confirmed to have HHT. If we order testing on someone who does not have HHT, and it is negative, this does not tell us whether the person has HHT or not. HHT could be caused by an unknown gene in the family, in which case a negative result is uninformative.

Once the genetic mutation in HHT has been found, relatives may choose to have genetic testing to see if they are positive or negative. If positive, they have inherited the genetic mutation for HHT and will need to undergo routine lifelong screening for AVMs. If negative, they did not inherit the mutation for HHT, will not develop HHT, and does not need screening. They also cannot pass on HHT to their future children.

There are currently 5 genes that are known to cause HHT. However not all genes for HHT have been found. There are some people with HHT in whom a genetic mutation cannot be found. In this case it is not possible to test relatives that do not yet show symptoms, because there is no genetic mutation to test for. These families can be enrolled in gene-finding studies if they are interested, to find more genes for HHT.

What is genetic counseling?
The Johns Hopkins HHT Center for Excellence offers genetic counseling for families with HHT. The goal of genetic counseling is to help you better under the inheritance of HHT in your family, and to help you decide how to proceed with genetic testing. A genetic counseling session includes taking a family history, reviewing medical history, explaining how genetic conditions are passed down through families, and discussing your options for genetic testing, including the advantages and limitations of genetic testing. Coming in for a genetic counseling appointment does not mean that you must have genetic testing performed.

How do I prepare for a genetic counseling visit?
You should bring results from any genetic test you or your relatives have had. It is most helpful

to bring the actual genetic testing laboratory report rather than the results letter from the hospital. Also, because family history can help determine the right type of testing, it can be helpful gather information about health conditions in the family prior to the appointment. In particular, it is helpful to know if your relatives have signs or symptoms of the following: nosebleeds, AVMs and where these have been located, aneurysms, skin telangiectasia (red spots) and where they are on the skin, gastrointestinal or other internal bleeding, or colon cancer. It is also helpful to know about what age they were when they had symptoms.

What does genetic testing involve?
The majority of genetic tests require a blood sample. Sometimes this can be done on a saliva sample. Results of genetic testing for HHT usually take between one and two months.

What is the cost of genetic testing?
Genetic testing for HHT depends on whether the mutation has been identified in your family. If the gene mutation in the family needs to be identified, testing can cost from $475 to $3000. If the gene mutation in your family has already been found, testing is about $200 to $350. In most cases these costs are covered by insurance.

Is genetic testing covered by insurance?
While genetic testing is covered by many insurance plans, some insurance companies require pre-authorization for genetic testing. Our staff will work with you to verify whether your insurance plan includes benefits for genetic testing, and whether you have co-pays or deductibles. We will not send out any genetic tests without your permission.

The genetic counselor for the Johns Hopkins HHT Center for Excellence is Kelsey Stauff.

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To set up a new appointment, please call the main Genetics office at Johns Hopkins at **410-955-3071**. Ask for a genetic counseling appointment for HHT. The office will review your insurance information to check coverage for the genetic counseling and genetic testing. You will get a call back within 2 weeks to schedule the actual appointment. Genetic counseling appointments are usually on Wednesday afternoons or Thursday mornings, but if there is a time that does not work for you, please call Kelsey directly to discuss.

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