
Hereditary Hemorrhagic Telangiectasia Testing Consent Form (Family Specific Mutation)

BACKGROUND

Hereditary hemorrhagic telangiectasia (HHT), also called Osler-Rendu-Weber disease, is a blood vessel disorder characterized by abnormal, direct connections between arteries and veins. Telangiectases are small abnormal blood vessels, which appear as red spots often found on the nose, lips, tongue, finger, and intestines. Larger abnormal blood vessels called arteriovenous malformations (AVMs) can occur in the internal organs, most commonly the lung, liver, and brain. Bleeding telangiectases in the nose or intestines can be either a minor annoyance or a major medical problem. Undetected and untreated lung and brain AVMs are a significant cause of life-threatening or disabling complications.

A large percentage of families with HHT have a mutation in either the endoglin gene (called HHT type 1) or the ACVRL1 (ALK 1) gene (called HHT type 2).

PURPOSE

I will be tested for a specific alteration in an HHT gene, previously identified in my family. The purpose of this genetic testing is to determine whether this altered HHT gene is present in me, which would predict the development of telangiectases and AVMs and confirm HHT.

Individuals with a close family history of HHT, regardless of symptoms, can be at-risk for this condition and may choose to be tested. Individuals with clinical features of HHT are also candidates for testing. Individuals identified as having HHT can benefit from consultation with medical practitioners who are knowledgeable about the particular clinical dilemmas unique to HHT. There is specific medical screening and management routinely recommended to all patients with HHT.

I have had the opportunity to discuss the purpose of HHT genetic testing for myself.

TESTING PROCEDURE

When testing has been completed, I understand that I will be contacted regarding my results by the office/center where I am being counseled and signing this consent form. I understand that ARUP Laboratories will report results to the physician or clinician requesting this test.

Several teaspoons (1 tube) of blood will be drawn to try to identify an HHT causing mutation. I agree to allow information to be provided to ARUP Laboratories about my medical and family history to aid in the interpretation of the test results. Testing is expected to take approximately four weeks to complete.

No information pertaining to my genetic test results will be provided to any of my relatives without my consent.

RISKS AND DISCOMFORTS

I understand that there is usually a minimal risk involved in drawing blood sample. This includes pain at the blood drawing site, bruising, bleeding, and infections.

The risks of information disclosure regarding my genetic diagnosis of HHT include depression, anxiety, anger, and fear of the future. This result could affect my relationships with family members and loved ones. Some individuals may experience feelings of guilt or other forms of anxiety even if they are found not to have an altered gene, while other family members have an altered gene.

I understand that if I learn that I have an altered HHT gene, my health and life insurance rates, my ability to obtain health, disability, or life insurance and my employability could be affected. Certain health, disability, and life insurance companies may consider HHT alteration or mutation to represent a "preexisting condition," and I may be responsible for disclosing this prior to obtaining new health or life insurance.

RESULTS

The results of this test are typically reported as either positive (predicts HHT) or negative (predicts no HHT). However, results cannot be used to predict disease if the variant for which testing is requested is considered to be of uncertain clinical significance.

The accuracy rate for this test is currently felt to exceed 99%. Rare errors can occur due to misincorporation of DNA bases by the enzyme used to perform the test, sample misidentification, sample contamination, primer site mutations, and general laboratory error.

BENEFITS OF RECEIVING RESULTS

If the family HHT gene mutation is not detected in me, I may experience a sense of relief as a result. If it is determined that I have the HHT gene mutation, I will be able to initiate a comprehensive surveillance plan for the variable and complex symptoms of HHT. It is the opinion of acknowledged experts in the field that enhanced surveillance will be of benefit. Also, I may choose to advise my relatives of this. They can have counseling and decide whether or not they wish to be tested to see if they inherited the same mutation.

LIMITATIONS OF DNA TESTING FOR HHT

I understand that there are limitations as to what these test results can tell me. This testing will not provide me with any information about my current health status. It does not predict which HHT symptoms I will have or their severity. I understand that it is possible to have HHT, but not have an HHT mutation detected by this testing.

USE OF SPECIMENS

My (or my child's or my fetus') DNA sample may be stored indefinitely to be used for test validation or education after personal identifiers are removed. No clinical tests other than the ones authorized will be performed. I may request disposal of my blood and DNA sample following completion of the test requested above by contacting the laboratory at (800) 242-2787 ext. 3301. For more information about ARUP, please refer to www.aruplab.com.

REQUEST FOR MORE INFORMATION

I understand that I may ask more questions about this testing and my results at any time. I will be given a copy of this "Consent Form" for my records.

CONSENT OF PATIENT

I have explained to _____ the purpose of this genetic testing, the procedure required, and the possible risks, benefits, and limitations.

Professional obtaining consent signature: _____

Print Name: _____ Date: _____

I have read and received a copy of this consent form. I understand the information provided in this document and I have had the opportunity to ask questions about the testing, the procedure, and the associated risks and limitations. I agree to have genetic testing and accept the risks and limitations.

Patient Signature: _____ Date: _____

Print Name: _____ Patient Date of Birth: _____