

REQUEST FOR HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT) TESTING

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS
Note that there is a 48-hour window for test cancellation. Cancellation requests after this period will be accommodated at the discretion of the lab.

PATIENT INFORMATION* Sample Type: _____ Collection Date & Time: _____

FIRST NAME _____ MI _____ LAST NAME _____ BIRTH DATE (MM/DD/YYYY) _____ ASSIGNED SEX _____

ANCESTRY European (non-Finnish) Amish European (Finnish) African

East Asian Jewish (Ashkenazi) Admixed American Middle Eastern South Asian

Specify countries: _____ Other: _____

CLINICAL INFORMATION

Patient currently has symptoms of HHT (please check the symptoms below):

Telangiectasias, Locations(s) _____ Nosebleeds, Frequency _____

PAVM CAVM Liver AVM GI Bleeding Positive family history of HHT

Juvenile polyps _____ Positive family history (Please include a 3 generation pedigree)

Patient is at risk for HHT or Juvenile Polyposis but is currently asymptomatic

If the test request is for site specific FAMILIAL ANALYSIS for a KNOWN VARIANT:

Name of person previously tested and relationship: _____

Was the previous testing performed at the Genetic Diagnostic Laboratory? Yes No

Result (Please include a copy of the result):* _____

ICD-10 CODE(S)* I78.0 Hereditary Hemorrhagic Telangiectasia I78.1 Other capillary disease (telangiectasia)
 R04.0 Epistaxis (nosebleeds) Z86.01 Personal history of colon polyp(s)
 Q07.9 Unspecified anomaly of brain, spinal cord, & nervous system Z84.81 Family history of carrier of genetic disease
 Other: _____

TEST REQUESTED*

STAT REQUEST (Check this box in addition to the test below if you intend to order STAT results in half (50%) of the listed TAT and a total cost of +20% of the base cost listed for the ordered test, this option excludes *Expedited* and *Prenatal* test requests.)

Sequence analysis of ENG / ACVRL1/ SMAD4 genes; if negative, reflex to deletion/duplication analysis

Site specific Sequencing analysis (familial) of ENG gene: _____ ACVRL1 gene: _____ SMAD4 gene: _____

Prenatal Site specific Sequencing analysis (familial) of ENG gene: _____ ACVRL1 gene: _____ SMAD4 gene: _____

* Required information

** Please call the laboratory prior to sending a prenatal or STAT sample. Please refer to the special requirements for prenatal samples on the Instructions for Sample Submission page.

PATIENT REGISTRATION FORM

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS

PATIENT INFORMATION

FIRST NAME	MI	LAST NAME	BIRTH DATE (MM/DD/YYYY)	ASSIGNED SEX
STREET ADDRESS				
CITY	STATE	ZIP	PHONE	

PHYSICIAN INFORMATION*

REFERRING PHYSICIAN	PHONE	FAX	
GENETIC COUNSELOR	PHONE	FAX	
EMAIL ADDRESS FOR COUNSELOR	EMAIL ADDRESS FOR PHYSICIAN		
INSTITUTION AND DEPARTMENT			
STREET ADDRESS	CITY	STATE	ZIP

PAYMENT OPTIONS* (must choose one) [a receipt will be mailed to the patient for self-pay options]

- I have enclosed a check payable to the "Genetic Diagnostic Laboratory" for \$ _____
- Please charge my credit card for the amount of \$ _____
- VISA
 Master Card
 Discover
 American Express
- Card Number: _____ Exp date: _____
- Name of cardholder as it appears on card: _____
- I have Pennsylvania Medicaid. A copy of my Medicaid card is attached.
- INSTITUTIONAL BILLING: The Institution where my testing originated has agreed to pay all charges for the testing.
INCLUDE Billing Address, Person Authorizing Payment, Telephone, and Fax below:

BILLING ADDRESS		
BILLING ADDRESS		
NAME OF INDIVIDUAL AUTHORIZING PAYMENT	PHONE	FAX

INFORMED CONSENT: GENETIC TESTING FOR HEREDITARY HEMORRHAGIC TELANGECTASIA (HHT)

Background: Hereditary hemorrhagic telangiectasia (HHT), also called Osler-Weber-Rendu 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 lips, tongue, fingers, intestines, or nose. 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 minor or a major medical problem, sometimes requiring transfusions. Undetected and untreated lung and brain AVMs are a significant cause of life-threatening or complications in individuals with HHT. Many individuals with a clinical diagnosis of HHT will carry a variant in either the *ENG* or *ACVRL1* genes. Some individuals with HHT may also have clinical features for familial juvenile polyposis (JP) – combined features of HHT and JP can be related to a variant in the *SMAD4* gene.

Purpose: The diagnostic samples will be used for the purpose of attempting to determine if I (or my child/fetus) am/is a carrier of an altered gene known to cause hereditary hemorrhagic telangiectasia. This information may help establish appropriate medical management.

Results: I understand that there are four possible results to this testing:

PATHOGENIC VARIANT: A clinically significant variant is detected in gene(s) analyzed. This may explain my personal or family history of HHT. My or my child's healthcare provider will make medical management recommendations based on this information.

LIKELY PATHOGENIC VARIANT: A variant is detected in the gene(s) analyzed which is the likely deleterious. This may explain my personal or family history of HHT. My or my child's healthcare provider will make medical management recommendations based on this information.

VARIANT OF UNCERTAIN SIGNIFICANCE: The laboratory may detect an alteration in the gene(s) analyzed which is currently of unknown significance, called a "variant of unknown significance (VUS)". The laboratory will work with my physician to help determine if the VUS can be further classified as to whether it is associated with HHT.

NEGATIVE: No clinically significant variants were identified in the gene(s) analyzed. This result reduces the likelihood that I, or my child, have a clinically significant variant in the gene(s) tested. Methods currently in use are unable to detect all variants and therefore I may still carry a variant that was not detected by the current technology.

Disclosure Policy: The Genetic Diagnostic Laboratory will release my test results to the ordering healthcare provider or genetic counselor, and otherwise only as permitted by law. The results will be kept confidential to the extent allowed by law. If I provide separate written consent, the lab will release my test results to other medical professionals or third persons I want to receive my results.

Limitations: While genetic testing is highly accurate for detection of the majority of disease causing variants, a small fraction of variants may be missed by the current technology. Due to the nature of the testing, there is a small possibility that the test will not work properly or that an error will occur. Occasionally, testing may reveal a variant of unknown significance that is unable to be definitively interpreted as positive or negative for disease-association based on the current knowledge of the variant. The DNA analysis performed at the University of Pennsylvania

Genetic Diagnostic Laboratory is specific only for the gene(s) analyzed and in no way guarantees my health. There are federal laws in place that prohibit health insurers and employers from discriminating based on genetic information, such as test results. There currently are no federal laws prohibiting discrimination based on genetic information by life insurance, long term care, or disability insurance companies, but state laws may restrict this. I understand I can ask my ordering provider or genetic counselor for more information about how insurers might use genetic information.

Initials _____

Genetic testing may identify information about biological family relationships that may be unexpected. Genetic testing may identify a medically relevant finding that is not related to the indication for testing. If genetic testing suggests that biological relationships between family members are not as reported to the Genetic Diagnostic Laboratory and/or another incidentally identified genetic result, I understand that these findings may be disclosed to the healthcare provider(s) who ordered the genetic testing.

Use of Specimens After Clinical Test Performed: I understand my blood or tissue specimen will not be returned to me or the ordering healthcare provider, and becomes the property of the lab upon receipt. The laboratory is not a DNA banking facility; therefore this is no guarantee that samples will be available or usable for additional or future testing. Samples from New York residents will be disposed of 60 days after clinical testing is complete.

After the laboratory completes the ordered clinical test, the lab may retain and preserve the specimen to validate the development of future genetic tests or for future research or education purposes. The laboratory is committed to continuous improvement and therefore I understand my coded sample may be used to validate a new assay. If testing reveals a clinically significant result during the validation process of a new assay related to the original indication for testing, my health provider may be contacted. If the lab uses the specimen for future research or education purposes, the specimen will be de-identified by removing my personally identifying information. My name, address and other personal identifying information will not be linked to the samples, or the results of the research, and I will not be identified in any research results or publications. I will not receive a copy of the research results. I can decline for my sample to be retained at the lab by filling out "Research Opt Out" form found on the following website: <https://genetics.med.upenn.edu/genetic-diagnostic-laboratory>

I understand the lab may wish to contact me, or my ordering healthcare provider, for additional information. The additional information may include, but would not be limited to, information about health and family history that might be relevant to the research. I understand I can decline future contact from the lab by filling out "Research Opt Out" form found on the following website: <https://genetics.med.upenn.edu/genetic-diagnostic-laboratory>

Genetic Counseling provided by a qualified specialist (i.e. genetic counselor/ medical geneticist) is a recommendation for individuals proceeding with genetic testing. This service is available before and after genetic testing. Additionally, other testing or further physician consults may be warranted.

The Genetic Diagnostic Laboratory is also an available resource to ask more questions about this testing. The laboratory genetic counselor can be reached at 215-573-9161 and Arupa Ganguly, PhD, FACMG can be reached at 215-898-3122. I will be given a copy of this consent form to keep.

HEALTHCARE PROVIDER STATEMENT:

I have explained to _____ the purpose of this genetic testing, the procedures required and the possible risks and benefits to the best of my ability.

Printed Name of Professional Obtaining Consent

Signature of Professional Obtaining Consent

Date

CONSENT OF PATIENT:

I have read and received a copy of this consent form. I agree to have genetic testing performed for myself, child or my fetus, and accept the risks. I understand the information provided in this document and I have had the opportunity to ask questions I have about the testing, the procedure, the associated risks and the alternatives.

Patient's Printed Name: _____

DOB: _____

Patient's Signature: _____
(or Parent/Guardian if patient is a minor)

Date: _____

Name and Relationship: _____
(Parent/Guardian if patient is a minor)

Initials _____