

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform HHT molecular genetic testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT) TESTING

Patient Name _____ **Date of Birth** ____/____/____ **Gender** F M

Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)
 African American Ashkenazi Jewish Asian Caucasian
 Hispanic Middle Eastern Native American Other _____

Does the patient have SYMPTOMS? No Yes; please check all that apply
 Nosebleeds (frequency): _____ Telangiectasia (locations): _____
 Brain AVM Liver AVM Lung AVM Spinal AVM
 Juvenile polyps Migraines Stroke (age): _____ Other: _____

Does the patient have a FAMILY HISTORY of HHT? No Yes Unknown
 If yes, attach the pedigree or specify the RELATIONSHIP of the family member(s) to the patient and detail the symptoms/age of onset in each symptomatic/affected relative: _____

Please list the gene and familial mutation if known: _____

Has the patient undergone previous DNA testing for HHT? No Yes
 If yes, please describe test(s) and results: _____

Circle the HHT test you intend to order.

- 0051382 Hereditary Hemorrhagic Telangiectasia (ACVRL1 and ENG) Sequencing and Deletion/Duplication**
 Sequencing of *ACVRL1* and *ENG* and deletion/duplication analysis. Clinical sensitivity ~85%.
- 2001961 Familial Mutation, Targeted Sequencing-** A copy of a relative's DNA laboratory result is **REQUIRED**.
- 0051381 Hereditary Hemorrhagic Telangiectasia (ACVRL1 and ENG) Sequencing** - Clinical sensitivity ~75%.
- 0051348 Hereditary Hemorrhagic Telangiectasia (ACVRL1 and ENG) Deletion/Duplication**
 Tests for large *ACVRL1* or *ENG* gene deletions or duplications. Clinical sensitivity ~10%.
- 2001971 Juvenile Polyposis (SMAD4) Sequencing and Deletion/Duplication** - For individuals with HHT but no identifiable *ACVRL1* or *ENG* mutation. Clinical sensitivity <5%.

For questions, contact an ARUP genetic counselor at (800) 242-2787 ext. 2141

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