ARUP LABORATORIES

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 a PEDIGREE or specify the relatives RELATIONSHIP to the patient. List their symptoms & age of onset:

____

Has DNA testing been performed for these family member(s)? [ ] No [ ] Yes [ ] Unknown

Please attach a copy of the relative's DNA laboratory result. <u>(REQUIRED for familial mutation testing).</u>

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

<u>Circle the test you intend to order.</u>

0051382 Hereditary Hemorrhagic Telangiectasia (*ACVRL1* and *ENG*) Sequencing and Deletion/Duplication
Sequencing of ***ACVRL1*** and ***ENG*** and deletion/duplication analysis. Clinical sensitivity ~85%.

0051381 Hereditary Hemorrhagic Telangiectasia (*ACVRL1* and *ENG*) Sequencing - Clinical sensitivity ~75%.

0051348 Hereditary Hemorrhagic Telangiectasia (*ACVRL1* and *ENG*) Deletion/Duplication
For patients with negative *ACVRL1* and *ENG* sequencing result. Clinical sensitivity ~10%. **Also order for familial *ACVRL1* or *ENG* large deletion or duplication testing.**

2001961 Familial Mutation, Targeted Sequencing- For familial *ACVRL1* or *ENG* small sequence change. A copy of a relative's DNA laboratory result is **REQUIRED**.

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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