ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR JUVENILE POLYPOSIS SYNDROME (JPS) / HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT) TESTING

Patient Name _____ Date of Birth _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply and describe)

☐ Cerebral arteriovenous malformation (AVM)

☐ GI carcinoma (location: _____; age of diagnosis: _____)

☐ Hepatic AVM

☐ Juvenile polyposis (number of polyps: _____; location(s): _____; age of onset: _____)

☐ Pulmonary AVM

☐ Spinal AVM

☐ Spontaneous, recurrent nosebleeds (frequency: _____)

☐ Telangiectasia (location(s): _____)

☐ Other symptom(s): _____

Has the patient had an allogeneic bone marrow or umbilical cord blood transplant? ☐ No ☐ Yes ☐ Unknown

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, specify: ☐ JPS ☐ HHT ☐ JP/HHT ☐ Neither ☐ Unknown

Attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset: _____

_____

_____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Check the test you intend to order.

☐ **2004992 Juvenile Polyposis (*BMPR1A*) Sequencing and Del/Dup:** Sequencing of *BMPR1A* coding regions and duplication/deletion analysis for large gene rearrangements. Clinical sensitivity 20–25% for JPS.

☐ **2001971 Juvenile Polyposis (*SMAD4*) Sequencing and Del/Dup:** Sequencing of *SMAD4* coding regions and duplication/deletion analysis for large gene rearrangements. Clinical sensitivity ~ 28% for JPS, unknown for JP/HHT. Order for HHT patients negative for *ENG* and *ACVRL1* mutations.

☐ **2004988 Juvenile Polyposis (*BMPR1A*) Sequencing:** Sequencing of *BMPR1A* coding regions and intron/exon boundaries. Clinical sensitivity is 18% for JPS.

☐ **0051510 Juvenile Polyposis (*SMAD4*) Sequencing:** Sequencing of the *SMAD4* coding regions and intron/exon boundaries. Clinical sensitivity of 21% for JPS, unknown for JP/HHT.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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