

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform JPS/HHT testing.
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** ____/____/____ **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 Unknown

If yes, check all that apply:

- Juvenile polyposis (number of polyps _____, location(s) _____, age of onset _____)
 GI carcinoma (location _____, age of diagnosis _____)
 Spontaneous, recurrent nosebleeds (frequency: _____)
 Telangiectasia (location(s): _____)
 Pulmonary arteriovenous malformation (AVM)
 Cerebral AVM
 Spinal AVM
 Hepatic AVM
 Other _____

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

Is there a FAMILY HISTORY of: JPS HHT JP/HHT Neither Unknown

If yes, please attach PEDIGREE or specify the relationship(s) of affected family member(s) to the patient, the symptoms and age at diagnosis in each affected relative: _____

 Please attach a copy of the relative's laboratory result (REQUIRED for familial mutation testing)

Please circle the JPS/ HHT test you intend to order.

2004992 Juvenile Polyposis (BMPRIA) Sequencing and Del/Dup Sequencing of *BMPRIA* 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 (BMPRIA) Sequencing Sequencing of *BMPRIA* 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 A copy of the affected relative's DNA lab result is REQUIRED.

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

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