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 AORTOPATHY 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 _____

Clinical Diagnosis / Reason for Referral: ☐ Confirmed ☐ Suspected ☐ Unknown

☐ Arterial tortuosity syndrome (ATS)
☐ Congenital contractural arachnodactyly (CCA)
☐ Ehlers-Danlos syndrome Type I/II, classic (EDS I/II)
☐ Ehlers-Danlos syndrome Type IV, vascular (EDS IV)
☐ Ehlers-Danlos syndrome Type VI, kyphoscoliotic (EDS VI)
☐ Familial ectopia lentis
☐ Familial thoracic aortic aneurysm (TAAD)
☐ Homocystinuria due to cystathionine beta-synthase deficiency (HCY)
☐ Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (JPHT)
☐ Loeys-Dietz syndrome (LDS)
☐ Shprintzen-Goldberg syndrome
☐ Marfan syndrome (MFS)
☐ Other _____

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

☐ Vascular/Cardiac/Thoracic: _____
☐ Craniofacial: _____
☐ Cutaneous: _____
☐ Ocular: _____
☐ Gastrointestinal: _____
☐ Musculoskeletal/Neurological: _____
☐ Other: _____

Has the patient undergone previous DNA testing for an Aortopathy? ☐ No ☐ Yes ☐ Unknown
If yes, describe the genes, disorder, methodology, and results: _____

_____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown
If yes, 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 these family member(s)? ☐ No ☐ Yes ☐ Unknown
Please attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing.)

Check the test you intend to order.

_____ **2006540 Aortopathy Panel, Sequencing and Deletion/Duplication.** Analysis of *ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, MYH11, MYLK, PLOD1, PLOD3, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFBR1, TGFBR2* genes.

_____ **2001691 Familial Mutation, Targeted Sequencing.** Tests for a mutation previously identified in a family member; copy of relative's lab result is REQUIRED.

Master Label

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