

**THIS IS NOT A TEST REQUEST FORM.**  
**The information below is required to perform aortopathy testing.**  
**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** \_\_\_\_/\_\_\_\_/\_\_\_\_ **Gender**  F  M

**Physician** \_\_\_\_\_ **Physician Phone** (\_\_\_\_) \_\_\_\_\_ **Practice Specialty** \_\_\_\_\_

**Genetic Counselor** \_\_\_\_\_ **Counselor Phone** (\_\_\_\_) \_\_\_\_\_

**Patient's Ethnicity** (check all that apply)

- |   |   |  |                                      |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian           | <input type="checkbox"/> Caucasian   |
| <input type="checkbox"/> Hispanic         | <input type="checkbox"/> Middle Eastern   | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

**Patient's Clinical Diagnosis**  Confirmed  Suspected  Unknown

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

**Patient's Symptoms**  No  Yes, check all that apply and describe

- Vascular/Cardiac/Thoracic: \_\_\_\_\_
- Craniofacial: \_\_\_\_\_
- Cutaneous: \_\_\_\_\_
- Ocular: \_\_\_\_\_
- Gastrointestinal: \_\_\_\_\_
- Musculoskeletal/Neurological: \_\_\_\_\_
- Other: \_\_\_\_\_

**Is there any relevant FAMILY HISTORY?**  No  Yes  Unknown

**If yes, attach a PEDIGREE** or specify the relatives 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).**

**Has the patient undergone previous DNA testing for an Aortopathy?**  No  Yes  Unknown

If yes, please describe the genes, disorder, methodology, and results \_\_\_\_\_

**Circle the test you intend to order.**

**2006540 Aortopathy Panel, Sequencing and Deletion/Duplication, 21 Genes.** Analysis of *ACTA2, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, MYH11, MYLK, PLOD1, PLOD3, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFBR1, TGFBR2* genes.

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

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

Master Label