ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

PATIENT HISTORY FORM FOR AORTOPATHY, LOEYS-DIETZ, OR MARFAN SYNDROME TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor's Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

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)
☐ 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: _____
☐ Musculoskeletal: _____
☐ Cutaneous: _____
☐ Other: _____
☐ Ocular: _____
☐ Craniofacial: _____
☐ Gastrointestinal: _____

Has the patient undergone previous DNA testing for an aortopathy? ..... ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

_____

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.** Confirm diagnosis of an aortopathy in individuals with aortic/vascular aneurysm, dissection, or rupture.

☐ **3003947 Loeys-Dietz Syndrome Core Panel, Sequencing.** Confirm diagnosis of Loeys-Dietz syndrome in individuals meeting clinical criteria. Sequences *TGFBR1* and *TGFBR2* genes only.

☐ **3004102 Marfan syndrome (FBN1) Sequencing and Deletion/Duplication.** Confirm diagnosis of Marfan syndrome in individuals who meet clinical criteria for MFS.

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

☐ **3003144 Deletion/Duplication Analysis by MLPA.** Tests for a large deletion/duplication 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.