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 MARFAN SYNDROME (*FBN1*) GENE 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 a diagnosis of Marfan Syndrome (MFS)? ☐ Confirmed ☐ Suspected ☐ Unknown

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

☐ **Vascular/Cardiac/Thoracic**
- ☐ Aortic dilatation/dissection
- ☐ Mitral valve prolapse
- ☐ Tricuspid valve prolapse
- ☐ Enlarged proximal pulmonary artery
- ☐ Pneumothorax

☐ **Craniofacial**
- ☐ Facial features of MFS
- ☐ Progeroid-like appearance

☐ **Ocular**
- ☐ Ectopia lentis
- ☐ Myopia

☐ **Cutaneous**
- ☐ Skin striae
- ☐ Cutis laxa
- ☐ Congenital lipodystrophy

☐ **Musculoskeletal/Neurological**
- ☐ Wrist and/or thumb sign
- ☐ Pectus carinatum/excavatum
- ☐ Hindfoot deformity
- ☐ Acetabular protrusion
- ☐ Scoliosis or thoracolumbar kyphosis
- ☐ Reduced upper/lower segment ratio
- ☐ Increased arm/height ratio
- ☐ Reduced elbow extension
- ☐ Dural ectasia

☐ **Other symptom(s):** _____

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown

If yes, describe the gene/disorder, methodology, and results: _____

_____

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

If yes, attach a pedigree or specify the affected 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

☐ **2006540 Aortopathy Panel, Sequencing and Deletion/Duplication:** Confirm diagnosis of an aortopathy in individuals with aortic/vascular aneurysm, dissection, or rupture.

☐ **2005584 Marfan Syndrome (*FBN1*), Sequencing and Deletion/Duplication:** Sequence analysis and MLPA of *FBN1* coding regions. Preferred test to confirm diagnosis when Marfan syndrome is strongly suspected by consensus criteria.

☐ **2005589 Marfan Syndrome (*FBN1*), Sequencing:** Sequence analysis of *FBN1* coding regions. Acceptable test to confirm diagnosis for individuals with clinical phenotype of Marfan syndrome.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a *FBN1* sequence change 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