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 FOR MARFAN SYNDROME (*FBN1*)

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male
Ordering Provider: _____ **Provider's Phone:** _____
Practice Specialty: _____ **Provider's Fax:** _____
Genetic Counselor: _____ **Counselor Phone:** _____

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

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

List country of origin (if known): _____

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 when Marfan syndrome is strongly suspected by consensus criteria.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a *FBN1* sequence change identified in a family member; copy of relative's lab result is REQUIRED.

☐ **3003144 Deletion/Duplication Analysis by MLPA:** Tests for large deletion/duplication previously identified in a family member; a copy of a relative's lab report is REQUIRED.

Master Label

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