ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform Marfan (*FBN1*) gene testing.
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_____ /_____/_____ Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)

[ ] African-American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] 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 _____

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)

Has the patient undergone previous DNA testing? [ ] No [ ] Yes [ ] Unknown
If yes, please describe the gene/disorder, methodology, and results _____

Circle the test you intend to order.

2006540 Aortopathy Panel Sequencing and Deletion/Duplication, 17 Gene: *ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, MYH11, MYLK, PLOD1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFBR1,TGFBR2.*

2005584 Marfan Syndrome (*FBN1*), Sequencing and Deletion/Duplication; Sequence analysis and MLPA of *FBN1*coding regions. Clinical sensitivity > 93% for sequencing, and is unknown for MLPA.

2005589 Marfan Syndrome (*FBN1*), Sequencing; Sequence analysis of *FBN1* coding regions. Clinical sensitivity ~93%.

2005580 Marfan Syndrome (*FBN1*), Deletion/Duplication; For patients with negative *FBN1*sequencing result. Clinical sensitivity is unknown. Also order for familial *FBN1* large deletion or duplication testing.

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

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

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