

**THIS IS NOT A TEST REQUEST FORM.**  
**The information below is required to perform Marfan (FBNI) gene testing.**  
**Please fill out this form and submit it with the test request form or electronic packing list.**

**PATIENT HISTORY FOR MARFAN SYNDROME (FBNI) GENE TESTING**

Patient's 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 _____ |

Does the patient have a diagnosis of Marfan syndrome (MFS)?  Confirmed  Suspected  Unknown

Does the patient have SYMPTOMS?  No  Yes, check all that apply

- |   |   |  |
|---|---|--|
| <input type="checkbox"/> <b>Vascular/Cardiac/Thoracic</b>   | <input type="checkbox"/> <b>Ocular</b>            | <input type="checkbox"/> <b>Musculoskeletal/Neurological</b> |
| <input type="checkbox"/> Aortic dilatation/dissection       | <input type="checkbox"/> Ectopia lentis           | <input type="checkbox"/> Wrist and/or thumb sign             |
| <input type="checkbox"/> Mitral valve prolapse              | <input type="checkbox"/> Myopia                   | <input type="checkbox"/> Pectus carinatum/excavatum          |
| <input type="checkbox"/> Tricuspid valve prolapse           |   | <input type="checkbox"/> Hindfoot deformity                  |
| <input type="checkbox"/> Enlarged proximal pulmonary artery |   | <input type="checkbox"/> Acetabular protrusion               |
| <input type="checkbox"/> Pneumothorax                       |   | <input type="checkbox"/> Scoliosis or thoracolumbar kyphosis |
|   | <input type="checkbox"/> <b>Cutaneous</b>         | <input type="checkbox"/> Reduced upper/lower segment ratio   |
| <input type="checkbox"/> <b>Craniofacial</b>                | <input type="checkbox"/> Skin striae              | <input type="checkbox"/> Increased arm/height ratio          |
| <input type="checkbox"/> Facial features of MFS             | <input type="checkbox"/> Cutis laxa               | <input type="checkbox"/> Reduced elbow extension             |
| <input type="checkbox"/> Progeroid-like appearance          | <input type="checkbox"/> Congenital lipodystrophy | <input type="checkbox"/> Dural ectasia                       |
| <input type="checkbox"/> 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.**

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

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

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

**2001961 Familial Mutation, Targeted Sequencing;** Tests for a *FBNI* 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**

Master Label