

FBN1/TGFBR2/TGFBR1 Gene Testing Patient Information Sheet



Instructions: The accurate interpretation and reporting of the genetic results is contingent upon the reason for referral, clinical information, ethnic background, and family history. To help provide the best possible service, please supply the information requested below and **send paperwork with the specimen.**

Patient Information

Patient Name <i>(Last, First, Middle)</i>	Birth Date <i>(Month DD, YYYY)</i>	Sex <input type="checkbox"/> Male <input type="checkbox"/> Female
Referring Physician Name	Phone	Fax
Other Contact	Phone	Fax

Clinical History

Patient's Diagnosis/Suspected Diagnosis: <input type="checkbox"/> Marfan syndrome <input type="checkbox"/> Marfan-related syndrome <input type="checkbox"/> Loews-Dietz syndrome <input type="checkbox"/> Familial thoracic aortic aneurysm and dissection <input type="checkbox"/> Other <i>(fill in)</i> _____																																																								
Indicate whether the following are present <i>(Adapted from Loeys, et al. (2010) J Med Genet 47: 476-485 and Loeys, et al. (2005), Nat Genet; 37: 275-281)</i>																																																								
<input type="checkbox"/> Aortic diameter at sinuses of valsalva Z-score ≥ 2 <input type="checkbox"/> Aortic dissection <input type="checkbox"/> Ectopia lentis <input type="checkbox"/> Systemic score ≥ 7 points (see table to the right for calculation) <input type="checkbox"/> Aortic dilatation/aneurysm (Z-score < 2) <input type="checkbox"/> Family history of independently diagnosed Marfan syndrome using the revised Ghent criteria	Systemic Score Calculation <table border="1" style="width: 100%; border-collapse: collapse;"> <thead> <tr> <th style="width: 70%;">Feature</th> <th style="width: 10%;">Value</th> <th style="width: 20%;">Enter Value If Present</th> </tr> </thead> <tbody> <tr><td>Wrist AND thumb sign</td><td style="text-align: center;">3</td><td></td></tr> <tr><td>Wrist OR thumb sign</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Pectus carinatum</td><td style="text-align: center;">2</td><td></td></tr> <tr><td>Pectus excavatum or chest asymmetry</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Hindfoot deformity</td><td style="text-align: center;">2</td><td></td></tr> <tr><td>Plain flat foot (pes planus)</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Pneumothorax</td><td style="text-align: center;">2</td><td></td></tr> <tr><td>Dural ectasia</td><td style="text-align: center;">2</td><td></td></tr> <tr><td>Protrusio acetabulae</td><td style="text-align: center;">2</td><td></td></tr> <tr><td>Reduced upper/lower segment AND increased armspan/height</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Scoliosis or thoracolumbar kyphosis</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Reduced elbow extension</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>3 of 5 facial features: • dolichocephaly • enophthalmos • downslanting palpebral fissures • malar hypoplasia • retrognathia</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Skin striae</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Myopia >3 diopters</td><td style="text-align: center;">1</td><td></td></tr> <tr><td>Mitral valve prolapse</td><td style="text-align: center;">1</td><td></td></tr> <tr><td style="text-align: right;">Total</td><td></td><td></td></tr> </tbody> </table>		Feature	Value	Enter Value If Present	Wrist AND thumb sign	3		Wrist OR thumb sign	1		Pectus carinatum	2		Pectus excavatum or chest asymmetry	1		Hindfoot deformity	2		Plain flat foot (pes planus)	1		Pneumothorax	2		Dural ectasia	2		Protrusio acetabulae	2		Reduced upper/lower segment AND increased armspan/height	1		Scoliosis or thoracolumbar kyphosis	1		Reduced elbow extension	1		3 of 5 facial features: • dolichocephaly • enophthalmos • downslanting palpebral fissures • malar hypoplasia • retrognathia	1		Skin striae	1		Myopia >3 diopters	1		Mitral valve prolapse	1		Total		
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Additional Features <input type="checkbox"/> Talipes equinovarus <input type="checkbox"/> Hypertelorism <input type="checkbox"/> Craniosynostosis <input type="checkbox"/> Cleft palate <input type="checkbox"/> Bifid uvula <input type="checkbox"/> Blue sclerae <input type="checkbox"/> Arterial tortuosity <input type="checkbox"/> Patent ductus arteriosus <input type="checkbox"/> Velvety/ translucent skin <input type="checkbox"/> Easy bruising <input type="checkbox"/> Widened atrophic scars <input type="checkbox"/> Spontaneous organ rupture <input type="checkbox"/> Other _____																																																								
See the diagnostic criteria at the bottom of this page and the following website for specific information about using the revised Ghent criteria: http://www.marfan.org/marfan/4265/2010																																																								

Ethnic Background and Family History - Attach pedigree if available

<input type="checkbox"/> European Caucasian <input type="checkbox"/> African American <input type="checkbox"/> Hispanic <input type="checkbox"/> Asian <input type="checkbox"/> Other <i>(specify)</i> _____ Are other relatives known to be affected? <input type="checkbox"/> Yes <input type="checkbox"/> No If Yes, indicate their relationship to the patient _____ Have other relatives had molecular genetic testing for FBN1/TGFBR2/TGFBR1 mutations? <input type="checkbox"/> Yes <input type="checkbox"/> No For FBN1 Known Mutation (89311), TGFBR2 Known Mutation (89462), and TGFBR1 Known Mutation (89460) testing, a familial mutation MUST be provided Indicate: Gene _____ Exon _____ Amino Acid _____ Nucleotide _____ OR Intron _____ Nucleotide _____ If relative was tested at the Mayo Clinic, name and relationship of relative or Family Number from relative's report _____ Attach a copy of the genetic test lab report if available

For the diagnosis of Marfan syndrome (see Loeys, et al. (2010) J Med Genet 47:476-485 for details):

In the absence of family history 1. Ao ($Z \geq 2$) and EL = MFS 2. Ao ($Z \geq 2$) and FBN1 = MFS 3. Ao ($Z \geq 2$) and Syst (≥ 7 pts) = MFS 4. EL and with known Ao = MFS	In the presence of family history 5. EL and FH of MFS = MFS 6. Syst (≥ 7 pts) and FH of MFS = MFS 7. Ao ($Z \geq 2$ above 20 yrs old, ≥ 3 below 20 years old) + FH of MFS=MFS
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