

## FBN1/TGFBR2/TGFBR1 Patient Information Form

**Providing the following clinical information is critically important in helping us to interpret genotype variations and to provide optimal genotype analysis on your patient:**

Date	Date of Birth	Gender: Male <input type="checkbox"/> Female <input type="checkbox"/>
Name of Patient		Ethnic origin: Asian <input type="checkbox"/> Black or African <input type="checkbox"/> Caucasian <input type="checkbox"/> Hispanic <input type="checkbox"/> Other <input type="checkbox"/>

**(1) Patient's Diagnosis:** \_\_\_\_\_

**(2) Test ordered and indication (check all that apply):**

- Full Sequencing:       *FBN1*       *TGFBR1*       *TGFBR2*  
 Deletion analysis:       *FBN1*       *TGFBR1*       *TGFBR2*  
 Known Mutation Detection: (indicate mutation and proband's identifier): \_\_\_\_\_

**(3) Clinical Diagnosis (please check all that apply) (adapted from De Paepe, et al. (1996), Am J Med Genet; 62: 417-426)**

<p><b><u>Skeletal System:</u></b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> pectus carinatum</li> <li><input type="checkbox"/> pectus carinatum requiring surgery</li> <li><input type="checkbox"/> reduced upper to lower segment ratio or arm span to height ratio greater than 1.05</li> <li><input type="checkbox"/> wrist and thumb signs</li> <li><input type="checkbox"/> scoliosis of &gt;20° or spondylolisthesis</li> <li><input type="checkbox"/> reduced extension at the elbows (&lt;170°)</li> <li><input type="checkbox"/> medial displacement of the medial malleolus causing pes planus</li> <li><input type="checkbox"/> protrusion acetabulae of any degree</li> <li><input type="checkbox"/> <i>pectus excavatum of moderate severity</i></li> <li><input type="checkbox"/> <i>joint hypermobility</i></li> <li><input type="checkbox"/> <i>highly arched palate with crowding of teeth</i></li> <li><input type="checkbox"/> <i>facial appearance (dolichocephaly, malar hypoplasia, enophthalmos retrognathia, down-slanting palpebral fissures)</i></li> </ul>	<p><b><u>Cardiovascular System:</u></b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> dilatation of the ascending aorta involving at least the sinuses of Valsalva</li> <li><input type="checkbox"/> dissection of the ascending aorta</li> <li><input type="checkbox"/> <i>mitral valve prolapse</i></li> <li><input type="checkbox"/> <i>dilation or dissection of the descending thoracic or abdominal aorta (&lt;50 yrs)</i></li> <li><input type="checkbox"/> Other:</li> </ul>
<p><b><u>Ocular System:</u></b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> ectopia lentis</li> <li><input type="checkbox"/> <i>abnormally flat cornea</i></li> <li><input type="checkbox"/> <i>increased axial length of globe</i></li> <li><input type="checkbox"/> <i>hyperplastic iris or hypoplastic ciliary muscle causing decreased miosis</i></li> <li><input type="checkbox"/> Other:</li> </ul>	<p><b><u>Pulmonary:</u></b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> <i>spontaneous pneumothorax</i></li> <li><input type="checkbox"/> <i>apical blebs</i></li> <li><input type="checkbox"/> Other:</li> </ul>
<p><b><u>Skin and Integument:</u></b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> <i>striae atrophicae (stretch marks) without obvious cause</i></li> <li><input type="checkbox"/> <i>recurrent or incisional herniae</i></li> <li><input type="checkbox"/> Other:</li> </ul>	<p><b><u>Dura:</u></b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> lumbosacral dural ectasia by CT or MRI</li> <li><input type="checkbox"/> Other:</li> </ul>

**(4) Please Attach Pedigree**