

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	Female
Name of Patient		Ethnic origin: Asian Black or African Caucasian Hispanic Other	

(1) **Patient's Diagnosis:** _____

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

Full Sequencing: *FBN1* *TGFBR2* *TGFBR1*

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

(4) Please Attach Pedigree