

TABLE E-1 Ghent Diagnostic Criteria for Marfan Syndrome<sup>4\*</sup>

Skeletal system
Major (presence of at least four of the following)
• Pectus carinatum
• Pectus excavatum requiring surgery
• Reduced upper segment-to-lower segment ratio or arm span-to-height ratio >1.05
• Wrist and thumb signs
• Reduced extension at the elbows (<170°)
• Scoliosis >20°
• Pes planus
• Protrusio acetabuli of any degree (ascertained on radiographs)
Minor
• Pectus excavatum
• Joint hypermobility
• High arched palate
• Facial
Dolichocephaly
Malar hypoplasia
Enophthalmos
Retrognathia
Down-slanting palpebral fissures
Ocular system
Major
• Ectopia lentus
Minor
• Flat cornea
• Increased axial length of the globe (>23.5 mm)
• Hypoplastic iris or hypoplastic ciliary muscle causing decreased miosis
Cardiovascular system
Major
• Dilation of the ascending aorta with or without aortic regurgitation and involving at least the sinuses of Valsalva
• Dissection of the ascending aorta
Minor
• Mitral valve prolapse with or without mitral valve regurgitation
• Dilation of the main pulmonary artery, in the absence of valvular or peripheral pulmonic stenosis before the age of 40 years
• Calcification of the mitral annulus before the age of 40 years
• Dilation or dissection of the descending thoracic or abdominal aorta before the age of 50 years
Pulmonary system
Minor (only)
• Spontaneous pneumothorax
• Apical blebs
Skin and integumentary system
Minor (only)
• Striae atrophicae
• Recurrent or incisional hernia
Neurologic system
Major
• Lumbar dural ectasia by computed tomography or magnetic resonance imaging
Family/genetic history
Major
• First-degree relative who independently meets the diagnostic criteria
• Presence of mutation in FBN1 gene
• Presence of haplotype around FBN1 inherited by descent and unequivocally associated with diagnosed Marfan syndrome in the family

\*An index case demonstrates major criteria in two different organ systems and involvement of a third organ system. An index case may be identified by confirming the gene mutation to be present and one major criterion and involvement of a second organ system. The relative of an index case requires one major criterion in family history, one major criterion in an organ system, and involvement of a second organ system.