

**DIAGNOSTIC CRITERIA FOR MARFAN SYNDROME
 (GHENT NOSOLOGY)**

Organ System	Major Criteria	Minor Criteria	Needed for Involvement
Cardiovascular	One of the following: Dilation of ascending aorta and sinuses of Valsalva Aortic dissection	Mitral valve prolapse Annulus mitralis calcification in patients < 40 yr Pulmonary artery dilation Descending or abdominal aorta dilation or dissection in patients < 50 yr	One major or 1 minor criterion
CNS	Lumbosacral dural ectasia (detected by MRI or CT)	N/A	One major criterion
Musculoskeletal	Four of the following: Pectus carinatum Pectus excavatum requiring surgery Arm span/height ratio > 1.05 or upper segment/lower segment ratio < 0.86 (in adults) Wrist and thumb signs Scoliosis > 20° or spondylolisthesis Elbow extension < 170° Pes planus (flat feet) Protrusio acetabuli	Typical facies (eg, dolichocephaly, malar hypoplasia, enophthalmos, retrognathia, down-slanting palpebral fissures) Joint hypermobility Pectus excavatum not requiring surgery High-arched palate with crowded teeth	Two of the 8 components comprising the major criteria or 1 of these plus 2 minor criteria
Ocular	Ectopia lentis (lens dislocation)	Myopia Flat cornea Iris or ciliary muscle hypoplasia	Two minor criteria
Pulmonary	N/A	Pneumothorax Apical blebs (seen on chest x-ray study)	One minor criterion
Skin	N/A	Atrophic striae not secondary to pregnancy or weight change Recurrent or incisional hernia	One minor criterion

N/A = not applicable.

If family/genetic history is not contributory, index case diagnosis requires major criteria in 2 organ systems and involvement in a 3rd. Alternatively, if a mutation known to cause Marfan syndrome is present, index case diagnosis requires 1 major criterion in 1 organ system and involvement in a 2nd. Diagnosis of an index case's relative requires 1 major criterion in family history, 1 major criterion in 1 organ system, and involvement in a 2nd. Minor criteria are used only to score organ system involvement and in themselves do not count toward the diagnosis.

Adapted from De Paepe A, Devereux RB, Dietz HC, et al: Revised criteria for Marfan syndrome. *American Journal of Medical Genetics* 62:417–426, 1996; Loeys B, Nuytinck L, Delvaux I, et al: Genotype and phenotype analysis of 171 patients referred for molecular study of the fibrillin-1 gene *FBNI* because of suspected Marfan syndrome. *Archives of Internal Medicine* 161:2447–2454, 2001.