

## Marfan Syndrome Diagnostic Criteria Checklist

De Paepe A, Deveroux RB, Dietz H, Hannekam RCM, and Pyeritz R. Revised Diagnostic Criteria for the Marfan Syndrome. American Journal of Medical Genetics 62: 417-426, 1996.

MIM 154700

NAME \_\_\_\_\_

DATE \_\_\_\_\_

EXAMINER \_\_\_\_\_

### Index case:

Major criteria in 2 different organ systems  
AND involvement of a third organ system.

### Relative of index case:

1 major criterion in family history  
AND 1 major criterion in an organ system  
AND involvement in second organ system.

## SKELETAL

**Major** (Presence of at least 4 of the following manifestations)

- pectus carinatum
- pectus excavatum requiring surgery
- reduced upper to lower segment ratio (Note 1)  
OR arm span to height ratio  $>1.05$   
Height \_\_\_\_ Arm span \_\_\_\_ Upper segment \_\_\_\_ Lower segment \_\_\_\_
- wrist (Note 2) and thumb (Note 3) signs
- scoliosis of  $>20^\circ$  or spondylolisthesis
- reduced extension at the elbows ( $<170^\circ$ )
- medial displacement of the medial malleolus causing pes planus
- protrusio acetabulae of any degree (ascertained on radiographs)

### Minor

- pectus excavatum of moderate severity
  - joint hypermobility
  - high arched palate with crowding of teeth
  - facial appearance
  - dolichocephaly,
  - malar hypoplasia,
  - enophthalmos,
  - retrognathia,
  - down-slanting palpebral fissures
- INVOLVEMENT: 2 major criteria or 1 major and 2 minor

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### **OCULAR**

#### **Major**

ectopia lentis

#### **Minor**

flat cornea

increased axial length of the globe

hypoplastic iris OR hypoplastic ciliary muscle causing decreased miosis

INVOLVEMENT: 2 minor criteria

### **CARDIOVASCULAR**

#### **Major**

dilatation 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

dilatation of the main pulmonary artery, in the absence of valvular or  
peripheral pulmonic stenosis below the age of 40 years

calcification of the mitral annulus below the age of 40 years

dilatation or dissection of the descending thoracic or abdominal aorta  
below age of 50 years

INVOLVEMENT: 1 minor criterion

### **PULMONARY**

#### **Minor (only)**

spontaneous pneumothorax

apical blebs

INVOLVEMENT: 1 minor criterion

### **SKIN AND INTEGUMENT**

#### **Minor (only)**

striae atrophicae

recurrent or incisional hernia

INVOLVEMENT: 1 minor criterion

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### DURA

#### Major

lumbosacral dural ectasia by CT or MRI

### FAMILY/GENETIC HISTORY

#### Major

first degree relative who independently meets the diagnostic criterion.

NAME \_\_\_\_\_

presence of mutation in FBN1 known to cause Marfan syndrome

presence of haplotype around FBN1 inherited by descent and unequivocally associated with diagnosed Marfan syndrome in the family

### For Differential Diagnosis:

- Homocystinuria - obtain plasma amino acids
- Congenital Contractural Arachnodactyly (MIM 121050)
- Familial Thoracic Aortic Aneurysm (MIM 132900)
- Familial Aortic Dissection (MIM 132900)
- Familial Ectopia Lentis (MIM 129600)
- Familial Marfan-Like Habitus (MIM 154705)
- MASS - myopia, mitral valve prolapse, mild aortic dilatation, skin and skeletal (at least 2, preferable 3 criteria)
- Familial mitral valve prolapse syndrome
- Stickler syndrome (MIM 108300) - myopia, vitreoretinal degeneration, retinal detachment, deafness arthropathy, spondyloepiphyseal dysplasia, joint hypermobility, midface hypoplasia, micrognathia, U-shaped cleft palate, mitral valve prolapse
- Shprintzen-Goldberg syndrome (MIM 182212) - Marfan type skeletal involvement AND craniosynostosis AND development delay

#### Notes

1. Hall JG, Froster-Iskenius UG, Allanson JE. Handbook of Normal Physical Measurements, Oxford: Oxford Univ. Press, pp 270-275.
2. wrist sign - thumb overlaps the distal phalanx of the fifth digit when grasping the contralateral wrist.
3. thumb sign - entire nail of the thumb projects beyond the ulnar border of the hand when the hand is clenched without assistance.