Marfan Syndrome

Diagnostic Criteria

Ghent Nosology revised 2010

No Family History

1. Aortic Root Dilatation Z score ≥ 2 AND Ectopia Lentis = Marfan syndrome
    1. The presence of aortic root dilatation (Z-score ≥ 2 when standardized to age and body size) or dissection and ectopia lentis allows the unequivocal diagnosis of Marfan syndrome, regardless of the presence or absence of systemic features
    2. Except where these are indicative of Shprintzen Goldberg syndrome, Loeys-Dietz syndrome, or vascular Ehlers Danlos syndrome.
2. Aortic Root Dilatation Z score ≥ 2 AND FBN1 = Marfan syndrome
    1. The presence of aortic root dilatation (Z ≥ 2) or dissection and the identification of a bona fide FBN1 mutation are sufficient to establish the diagnosis, even when ectopia lentis is absent.
3. Aortic Root Dilatation Z score ≥ 2 AND Systemic Score ≥ 7pts = Marfan syndrome
    1. Where aortic root dilatation (Z ≥ 2) or dissection is present, but ectopia lentis is absent and the FBN1 status is either unknown or negative, a Marfan syndrome diagnosis is confirmed by the presence of sufficient systemic findings (≥ 7 points, according to a scoring system) confirms the diagnosis.
    2. However, features suggestive of Shprintzen Goldberg syndrome, Loeys-Dietz syndrome, or vascular Ehlers Danlos syndrome must be excluded and appropriate alternative genetic testing (TGFBR1/2, SMAD3, TGFB2, TGFB3, collagen biochemistry, COL3A1, and other relevant genetic testing when indicated and available upon the discovery of other genes) should be performed.

Family History Present

1. Ectopia lentis AND Family History of Marfan syndrome (as defined above) = Marfan syndrome
    1. The presence of ectopia lentis and a family history of Marfan syndrome (as defined in 1-4 above) is sufficient for a diagnosis of Marfan syndrome.
2. A systemic score ≥ 7 points AND Family History of Marfan syndrome (as defined above) = Marfan syndrome
    1. A systemic score of greater than or equal to 7 points and a family history of Marfan syndrome (as defined in 1-4 above) is sufficient for a diagnosis of Marfan syndrome.
    2. However, features suggestive of Shprintzen Goldberg syndrome, Loeys-Dietz syndrome, or vascular Ehlers Danlos syndrome must be excluded and appropriate

alternative genetic testing (TGFBR1/2, SMAD3, TGFB2, TGFB3 collagen biochemistry, COL3A1, and other relevant genetic testing when indicated and available upon the discovery of other genes) should be performed.

3. Aortic Root Dilatation Z score ≥ 2 above 20 yrs. old, ≥ 3 below 20 yrs. old + Family History of Marfan syndrome (as defined above) = Marfan syndrome
    1. The presence of aortic root dilatation (Z ≥ 2 above 20 yrs. old, ≥ 3 below 20 yrs. old) and a family history of Marfan syndrome (as defined in 1-4 above) is sufficient for a diagnosis of Marfan syndrome.
    2. However, features suggestive of Shprintzen Goldberg syndrome, Loeys-Dietz syndrome, or vascular Ehlers Danlos syndrome must be excluded and appropriate alternative genetic testing (TGFBR1/2, SMAD3, TGFB2, TGFB3, collagen biochemistry, COL3A1, and other relevant genetic testing when indicated and available upon the discovery of other genes) should be performed

Aortic Root Z-Scores

- Calculator for Children
- Calculator for Adults

References

- Loeys et al. Journal Medical Genetics 2010
- The Marfans Foundation

syndrome