Musculoskeletal Problems

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Clinical picture

Marfan syndrome and the thumb sign

A 74-year-old woman presented with congestive heart failure exacerbation. She was known to have moderate-to-severe aortic regurgitation and echocardiogram exam demonstrated ascending aortic aneurysm of 4.6 cm diameter on her recent hospitalization, 1 month ago. The patient’s appearance was suggestive for Marfan syndrome, and complete physical examination, in addition to the known aortic root disease, further strengthened the suspicion by demonstrating arachnodactyly (including thumb and wrist signs) and several other suggestive features (pectus excavatum, pes planus, thoracolumbar kyphosis and suggestive facial features). A positive thumb sign indicates that the distal phalanx of the adducted thumb extends beyond the ulnar border of the palm (Figure 1a, b).

Though exhibiting quite impressive morphological features, our patient was never officially diagnosed before and was never evaluated systematically for disease manifestations. The diagnosis of Marfan syndrome relies on a set of defined clinical criteria (the Ghent nosology) developed to facilitate accurate recognition of the syndrome and improve patient management and counseling. According to the 2010 revised Ghent nosology,\(^1\) due to lack of known family history and lack of current ocular findings, her diagnosis could have been established during hospitalization by means of combination of ascending aortic aneurysm and systemic features score of seven points. She was referred immediately to her chest surgeon and to genetic testing (as it helps to validate the clinical diagnosis, exclude alternative diagnoses and facilitates the diagnosis in the patients’ relatives).\(^2\) Her first degree relatives were recommended to undergo aortic imaging, as aortic dissection is by far the commonest cause of death in the natural history of Marfan syndrome.\(^3\)

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References


Figure 1. Positive thumb sign: the distal phalanx of the adducted thumb extends beyond the ulnar border of the palm.