7q11.23 duplication encompassing the ELN gene: a rare cause of thoracic aortic disease

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Introduction: Disease of the aortic root and ascending aorta is commonly linked to hereditary or congenital factors. Genetic disorders affecting the thoracic aorta are known as heritable thoracic aortic disease, which can present as syndromic and non-syndromic, with underlying gene defects encoding three major groups: the extracellular matrix, TGF-β signaling pathway, and the smooth muscle cell contractile apparatus.[1]

Case report: A 34-year-old female was referred for cardiologic evaluation due to elevated blood pressure and periodic chest pain. Echocardiography revealed mild to moderate aortic regurgitation with combined root and ascending aorta dilation (**Figure 1**). MR and CT aortography confirmed dilation of the ascending aorta up to 50 mm, with sinuses of Valsalva and sinotubular junction measuring up to 49 mm (**Figure 2**). Due to extra-aortic features (short stature, wide and short neck, short fourth and fifth metacarpal bones, scoliosis, small breasts), mosaic Turner syndrome was initially suspected but her karyotype analysis was normal. Further genetic testing using the Aorta Panel identified a heterozygous duplication of 7q11.23 encompassing the ELN gene, which encodes the elastin protein. 7q11.23 duplication has been associated with thoracic aneurysms, presumably due to increased ELN expression and elastin excess[2]. Interpreting the clinical relevance of 7q11.23 duplications is challenging, as phenotypic variability is wide. There are no specific prediction models that can estimate the risk of rupture or dissection in these patients. Nevertheless, considering her short stature (her height was 154 cm), we calculated the aortic size index: 30 mm/m^2, aortic height index: 32 mm/m, and the z-score: 7 - all markedly elevated. She underwent a successful valve-sparing "Florida sleeve" procedure (**Figure 3**), with uneventful postoperative course. Subsequently, her parents were tested using a

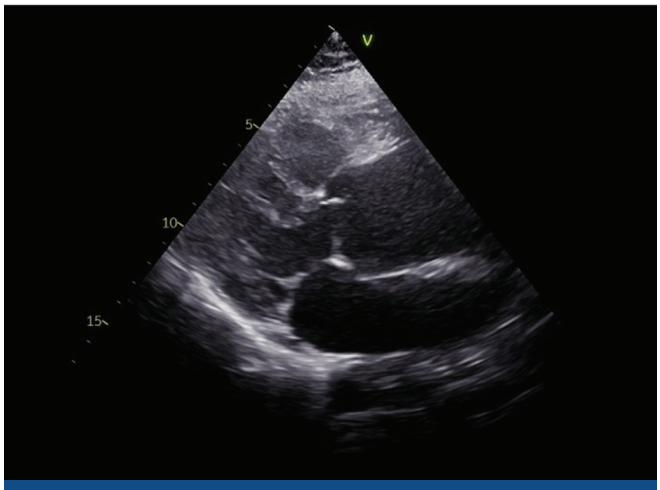

FIGURE 1. Echocardiographic image of the dilated aortic root and ascending aorta (yellow arrow).

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molecular karyotype test (array-based comparative genomic hybridization) and the results were normal, indicating that the mutation in our patient occurred de novo. Nevertheless, we performed an echocardiographic screening in both of her siblings, which confirmed normal aortic dimensions.

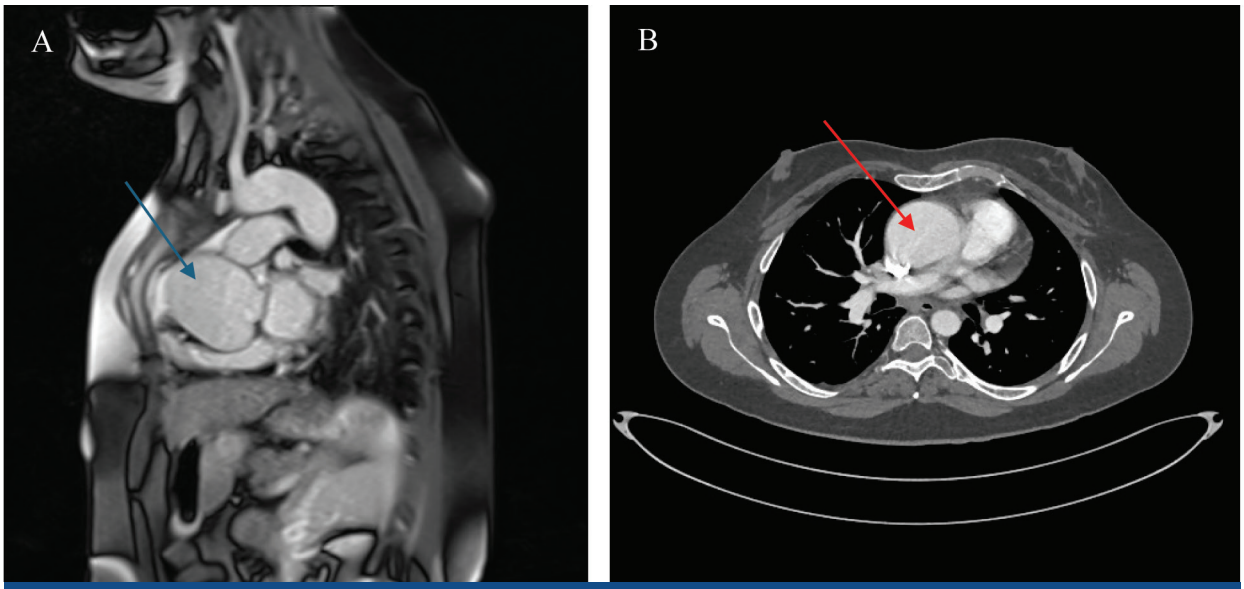

FIGURE 2. A) MR image of the dilated aortic root (blue arrow); B) dilated ascending aorta on CT scan (red arrow).

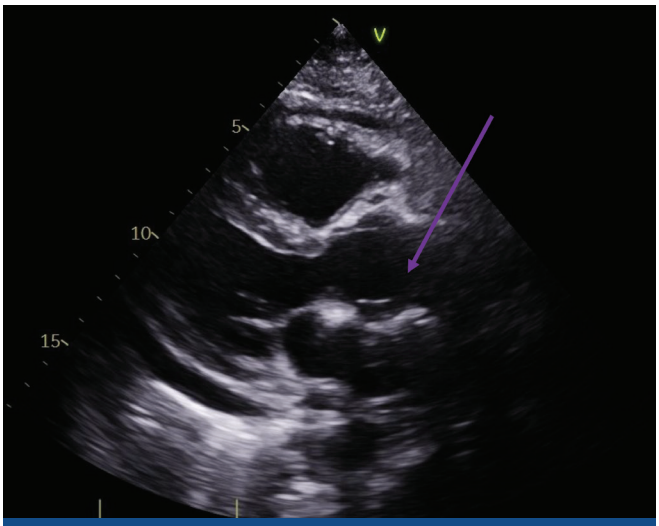

FIGURE 3. Echocardiographic image of the ascending aorta (purple arrow) after the „Florida sleeve" procedure.

Conclusion: There is a need for better characterization and risk stratification models in rare genetic aortopathies.

LITERATURE

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