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Severe Aortic Root Dilatation with Moderate-to-Severe Aortic Regurgitation in the Setting of Severe Pectus Excavatum: A Case Report

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Abstract

Severe dilatation of the aortic root is a potentially life-threatening condition that may arise in the context of heritable connective tissue disorders. We present the case of a 33-year-old male found incidentally to have a significant cardiac murmur during a pre-employment health check, who was subsequently diagnosed with severe aortic root dilatation measuring 60–67 mm at the sinuses of Valsalva, associated with moderate-to-severe aortic regurgitation. Thoracic imaging demonstrated severe pectus excavatum with a markedly elevated Haller index of 11.5, with resultant displacement of cardiac structures and a severe restrictive ventilatory defect on pulmonary function testing. Clinical examination revealed additional dysmorphic features including a high-arched palate. A family history of sudden death in a first-degree relative further raised the index of suspicion for an underlying heritable connective tissue disorder, most likely Marfan syndrome. This case highlights the importance of prompt recognition of syndromic aortopathy in young patients, the multidisciplinary complexity of management, and the urgent need for cardiothoracic surgical intervention in the context of markedly enlarged aortic root dimensions with associated valvular pathology.

Keywords: Aortic root dilatation; Aortic regurgitation; Pectus excavatum; Marfan syndrome; Heritable aortopathy; Connective tissue disorder; Cardiothoracic surgery

Introduction

Severe aortic root dilatation in young individuals is a critical clinical finding that warrants prompt evaluation for underlying heritable connective tissue disorders, particularly Marfan syndrome. Progressive enlargement of the aortic root predisposes to aortic regurgitation, dissection, and rupture, which remain the leading causes of morbidity and mortality in affected individuals [1-3]. Pectus excavatum is a common musculoskeletal manifestation associated with syndromic aortopathies and may contribute significantly to cardiorespiratory compromise. The coexistence of severe thoracic deformity and aortic pathology presents unique diagnostic and management challenges [4]. We present a case of severe aortic root dilatation with associated aortic regurgitation and marked pectus excavatum, highlighting the importance of early recognition, multidisciplinary management, and timely surgical intervention.

Case Presentation

A 33-year-old male was incidentally found to have a cardiac murmur during a routine pre-employment health examination. He was subsequently referred for echocardiographic assessment, which revealed significant dilatation of the aortic root measuring 67 mm at the sinuses of Valsalva. Left ventricular systolic function was preserved with an ejection fraction of 64%, and moderate-to-severe aortic regurgitation (AR) was demonstrated. There was no prior cardiac history, and the patient had been largely asymptomatic. The patient was referred to a secondary care centre for further evaluation. CT aortogram and CT coronary angiography were performed, confirming severe aneurysmal dilatation of the aortic root measuring up to 62 mm, with the ascending aorta remaining within normal limits at approximately 35 mm. No features of acute coronary syndrome were identified. Thoracic imaging demonstrated severe pectus excavatum with a Haller index of 11.5 — considerably above the threshold of 3.25

used to define clinically significant deformity [5] — with marked reduction of the anteroposterior thoracic dimension and displacement of cardiac structures into the left hemithorax. On clinical examination, the patient exhibited two notable dysmorphic features: a high-arched palate and longstanding severe pectus excavatum. These findings, in conjunction with the cardiovascular imaging, prompted further investigation for an underlying connective tissue disorder.

Pulmonary function testing was performed to characterise the respiratory impact of the thoracic deformity. Results demonstrated a severely reduced forced expiratory volume in one second (FEV_1) at 43% of predicted, a forced vital capacity (FVC) of 40% of predicted, a preserved FEV_1/FVC ratio of 88%, and a markedly reduced total lung capacity (TLC) of 56% of predicted. This pattern is consistent with a severe restrictive ventilatory defect, attributable to the marked reduction in thoracic volume imposed by the chest wall deformity. The patient was subsequently referred for high-resolution computed tomography (HRCT) of the chest for further characterisation. Of notable significance, the patient's father died suddenly at the age of 66 years. The precise cause of death was not formally documented; however, in the clinical context of this presentation, a heritable aortopathy or connective tissue disorder cannot be excluded and warrants formal family screening and genetic counselling. Discharge advice included restriction from moderate-to-severe exercise, weight lifting, running, and any activity involving significant straining or Valsalva manoeuvres, given the risk of acute aortic events. The patient was also advised to abstain from driving pending appropriate medical clearance. Outpatient review was arranged with cardiothoracic surgery for consideration of a Bentall procedure.

Discussion

Marfan Syndrome and Heritable Connective Tissue Disorders

Marfan syndrome is an autosomal dominant disorder of connective tissue caused by pathogenic variants in the FBN1 gene, encoding fibrillin-1, a critical structural glycoprotein of the extracellular matrix [1]. It has an estimated prevalence of approximately 1 in 5,000 to 1 in 10,000 individuals, with around 25% of cases representing de novo mutations in the absence of a positive family history [3]. The condition exhibits considerable phenotypic variability and affects multiple organ systems, including the cardiovascular, musculoskeletal, ocular, and pulmonary systems. The cardinal cardiovascular manifestation of Marfan syndrome is progressive dilatation of the aortic root at the level of the sinuses of Valsalva, which predisposes to aortic regurgitation, aortic dissection, and rupture — the leading causes of premature death in untreated individuals [2]. Prophylactic aortic root replacement is generally recommended when the aortic root diameter reaches 45–50 mm in adults with Marfan syndrome, and at lower thresholds in the presence of rapid expansion, significant aortic regurgitation, family history of dissection at smaller dimensions, or in women planning pregnancy [2,6]. An aortic root diameter of 60–67 mm, as documented in this case, represents a significantly enlarged measurement well above accepted intervention thresholds and confers a markedly elevated risk of dissection or rupture. Skeletal manifestations of Marfan syndrome include tall stature, arachnodactyly, pectus excavatum or pectus carinatum, scoliosis, pes planus, and joint hypermobility. Pectus excavatum is present in up to 70% of individuals with Marfan syndrome and can contribute meaningfully to cardiorespiratory compromise, as demonstrated by the severe restrictive ventilatory defect observed in this case [3]. Ocular manifestations — most notably ectopia lentis (superior lens dislocation) — are a hallmark feature of the condition and are incorporated into the revised Ghent nosology, the internationally accepted diagnostic framework for Marfan syndrome [1] (Figures 1,2).

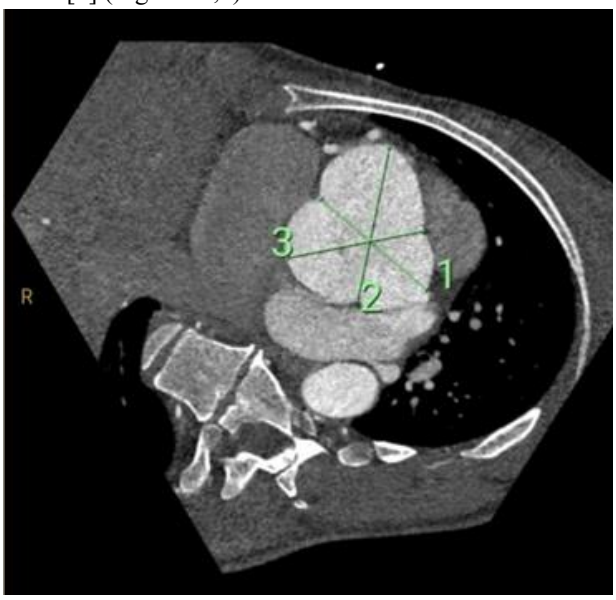

Figure 1: *CT aortogram demonstrating severe aneurysmal dilatation of the aortic root at the level of the sinuses of Valsalva.*

The differential diagnosis of heritable thoracic aortopathy is broad. Loeys-Dietz syndrome, caused by mutations in TGFBR1, TGFBR2, SMAD3, or TGFB2, shares several features with Marfan syndrome but is associated with more aggressive aortic disease at smaller diameters, hypertelorism, bifid uvula, and arterial tortuosity [7]. Ehlers-Danlos syndrome — particularly the vascular subtype (vEDS), caused by COL3A1 mutations — is characterised by fragility of medium and large arteries, spontaneous arterial rupture, and a notably high mortality risk; aortic root dilatation is less prominent but vascular events may

occur at any site. Bicuspid aortic valve-associated aortopathy represents a distinct and common cause of ascending aortic dilatation and should be excluded by echocardiography [8]. A comprehensive genetic panel for heritable thoracic aortopathies is warranted in cases such as this, particularly where a family history of premature aortic or cardiovascular events is present. The co-occurrence of aortic root dilatation, aortic regurgitation, pectus excavatum with a severely elevated Haller index, high-arched palate, and a first-degree family history of sudden death in this patient represents a compelling clinical picture for Marfan syndrome. Formal evaluation using the revised Ghent criteria — including ophthalmological assessment for ectopia lentis and systemic scoring — alongside molecular genetic testing is essential to confirm the diagnosis and guide long-term surveillance and management [1].

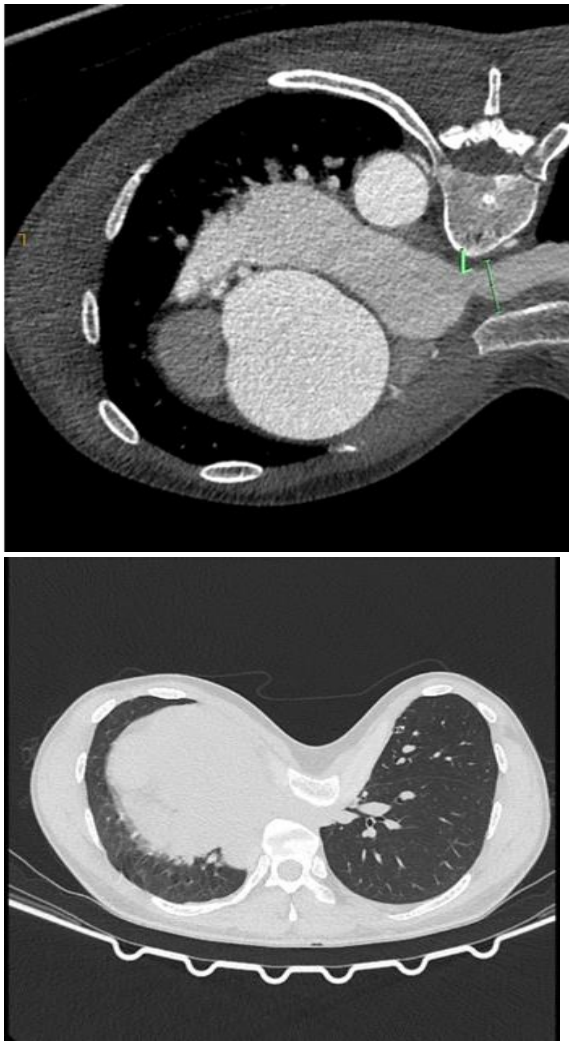

Figure 2*: Axial CT chest image demonstrating severe pectus excavatum with marked reduction in the anteroposterior thoracic diameter, associated cardiac displacement, and a Haller index of 11.5.*

Management Considerations

Valve-sparing aortic root replacement, where technically feasible and anatomically appropriate, offers the potential for definitive correction of the aneurysm while preserving native aortic valve function, which is of particular relevance in a young patient with an expected prolonged lifespan [9]. Concurrent assessment of the severe restrictive respiratory deficit and its interaction with surgical risk will require multidisciplinary input from respiratory medicine and anaesthetics.

Conclusion

This case illustrates a high-risk presentation of severe aortic root dilatation in a young male, identified incidentally during a pre-employment health screen. The combination of markedly enlarged aortic root dimensions (60–67 mm), moderate-to-severe aortic regurgitation, severe pectus excavatum with a Haller index of 11.5, high-arched palate, and a family history of sudden death in a first-degree relative is strongly suggestive of an underlying heritable connective tissue disorder, most likely Marfan syndrome. The aortic root dimensions documented in this case significantly exceed established thresholds for prophylactic surgical intervention and represent a clear and urgent indication for cardiothoracic surgical assessment. This case underscores the importance of thorough clinical evaluation of incidental murmurs in young individuals, the need for systematic assessment of dysmorphic features that may indicate an underlying syndromic aetiology, and the critical role of multidisciplinary care in the comprehensive management of heritable aortopathies. Cascade screening of first-degree relatives is strongly recommended given the potential hereditary basis of this presentation and the associated risk to family members.

Event Findings

Pre-employment health examination Incidental cardiac murmur detected. Echocardiography Aortic root 67 mm at sinuses of Valsalva, LVEF 64%, moderate-to-severe AR. CT aortogram / CT coronary angiography Aortic root 62 mm, ascending aorta 35 mm, severe pectus excavatum (Haller index 11.5). Pulmonary function testing Severe restrictive defect (FEV_1 43%, FVC 40%, TLC 56%). Discharge planning Activity restriction, driving abstinence, cardiothoracic surgery referral.

Ethics & Consent

Ethics Approval: Not required for single case reports as per institutional policy.

Consent: Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Competing Interests: The authors declare that they have no competing interests.

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Authors' Contributions: All authors contributed to the clinical care of the patient, conception of the report, and drafting of the manuscript. All authors read and approved the final manuscript.

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