A Rare Case of a Giant Aortic Arch Saccular Aneurysm in a 16-year-old Filipino with Marfan Syndrome.

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Abstract

Background:

Marfan syndrome (MFS) is an autosomal dominant connective tissue disorder affecting the fibrillin 1 gene. It mainly affects the skeletal, ocular, and cardiovascular and system, with cardiovascular complications being the most devastating. The most notable of the cardiovascular complications are aortic root dilatation, aortic aneurysm, and dissection. Giant saccular aneurysms of the aortic arch are extremely rare in Marfan patients particularly in adolescents, mainly because it usually affects the aortic root. Only a few of such case have been reported in the literature.

Case Presentation:

We report the case of a 16-year-old Filipino male with phenotypic features fulfilling the revised Ghent criteria for MFS. The patient presented with a two-month history of nonproductive cough and was found incidentally to have a widened mediastinum on chest radiograph. CT aortogram imaging revealed a giant wide-necked saccular aneurysm of the aortic arch measuring 12 × 10 cm, causing compression of the trachea, left mainstem bronchus, and esophagus. Echocardiography also showed a dilated aortic root (z-score +3.24) with trivial mitral and aortic regurgitation.

Management and Outcome:

The patient underwent a successful hemiarch replacement with reimplantation of supra-aortic vessels using a 2 vessel island flap under hypothermic circulatory arrest. Histopathology of the

resected specimen demonstrated medial degeneration with elastic fiber fragmentation and smooth muscle cell loss, consistent with Marfan aortopathy. The postoperative course was uneventful, and follow-up imaging showed an intact aortic graft without evidence of leak or dissection.

Conclusion:

This case highlights the rare occurrence of a giant saccular aneurysm of the aortic arch in an adolescent with Marfan syndrome. While aortic root dilatation is the most common vascular manifestation, atypical aneurysm morphology and location can occur and may remain asymptomatic until incidental discovery or life-threatening complications arise. Prompt recognition, surgical intervention, and long-term surveillance are essential to improve outcomes in this high-risk population.