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Case Report

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Spontaneous rupture of a left subclavian artery pseudoaneurysm in a child

Abstract

A 14-year-old boy with history of generalized joint hyper mobility and ongoing genetics work-up for connective tissue disorder presented to our hospital for one day of acute left-sided chest pain and shortness of breath after feeling a "pop" in his chest while trying to put on a jacket. CT chest and subsequent angiography demonstrated a large left hemothorax with a psuedoaneurysm of the left subclavian artery at the base of the vertebral artery, which was repaired with urgent successful placement.

Keywords: pseudoaneurysm, ehlers-danlos syndrome, pediatric interventional cardiology

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Case presentation

A 14-year-old boy with history of generalized joint hypermobility and ongoing genetics work-up for connective tissue disorder presented to our hospital for one day of acute left-sided chest pain and shortness of breath after feeling a "pop" in his chest while trying to put on a jacket. He was tachycardic and had absent breath sounds in the left lobe. A chest X-ray showed a large left pleural effusion and a subsequent CT chest demonstrated a large left hemothorax with a psuedoaneurysm of the left subclavian artery (LSCA) at the base of the vertebral artery. Angiography confirmed the presence of a LSCA pseudoaneurysm that measured 15 by 8mm (Figure 1A video/Panel A). A 5mm by 3 cm Cordis OptaPro balloon was then placed in the LSCA just distal to the vertebral artery and proximal angiogram demonstrating that the bleed could be occluded without obstructing flow to the vertebral artery. Subsequently, the patient underwent successful placement of a 6 mm Gore VBX stent (Figure 1B video/Panel B) via positioning with an Amplatz Extra Stiff wire in the distal LSCA without obstruction to flow to the vertebral artery. At follow-up, the patient is doing well and was recently confirmed to have *PLOD1*-related kyphoscoliotic Ehlers-Danlos syndrome (1902+1G>T variant). Causes of subclavian artery pseudoaneurysms include connective-tissue disorders, congenital defects and infections.

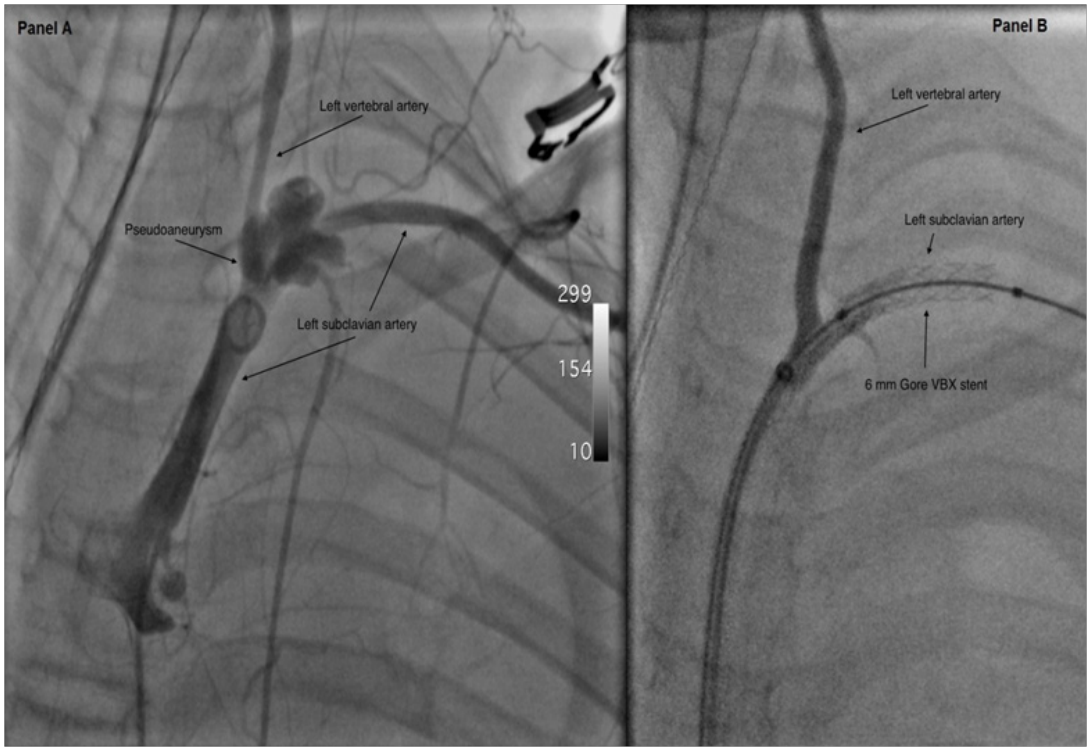

Figure 1A & 1B A chest X-ray showed a large left pleural effusion and a subsequent CT chest demonstrated a large left hemothorax with a psuedoaneurysm of the left subclavian artery (LSCA) at the base of the vertebral artery.

Discussion

Ehlers-Danlos Syndrome (EDS) is a heterogeneous group of disorders with overlapping clinical characteristics including skin hyper extensibility, joint hypermobility and vascular fragility. While the original classification of EDS comprised of six main EDS subtypes, after the introduction of next-generation sequencing techniques, the list has since expanded to include non-collagenous extracellular matrix (ECM) types and now comprises 13 distinct clinical subtypes in 19 different genes.[1] Depending on the EDS subtype and the underlying genetic defect, their clinical consequences, including vascular complications can range from mild to severely debilitating and even life-threatening. *PLOD1*-related kyphoscoliotic Ehlers-Danlos syndrome (kEDS) is an autosomal recessive subtype caused by a defect in the protein, lysylhydroxylase 1 (LH1), encoded by the *PLOD1* gene, which plays an essential role in the formation of intra- and intermolecular collagen crosslink. To date, 83 patients from 73 families with confirmed kEDS_*PLOD1*have been reported.[1]In addition to the hallmark features of EDS, patients with this subtype have an increased risk for rupture of medium-sized arteries (up to 20% in a recent systematic review), the majority of which have been severe and potentially life-threatening.[2] To date, these include six cases of antenatal or neonatal intracerebral hemorrhage and a series of case reports involving arterial dissections or aneurysms, especially in teenagers and young adults.[3–8] Possible association between LH1 deficiency and increased vascular fragility has been studied in a *Plod1*$^{-/-}$ mouse model mice where about 15% of the knockout mice died because of aortic rupture thought to be related to degenerated aortic smooth muscles and abnormal collagen fibrils in the aortic wall.[9] Our case is the first to describe a subclavian pseudoaneurysm in the kEDS subtype and highlights both the dramatic presentation of EDS as a massive hemothorax from a ruptured LSCA aneurysm and subsequent importance of regular cardiovascular assessment and imaging in patients with both vascular and non-vascular forms of EDS. Future research focusing on guidelines for specific long-term management of vascular complications in EDS remains ongoing.

Acknowledgments

None.

Conflicts of interest

The authors do not have any conflicts of interest.

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