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Short Report

Spontaneous Brachial Pseudo-aneurysm in a 12-year-old with Kyphoscoliosis-type Ehlers–Danlos Syndrome

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ABSTRACT

The Ehlers–Danlos Syndrome (EDS) is a rare connective tissue disorder characterised by fragility of the soft connective tissues and widespread manifestations in skin, ligaments, joints, blood vessels and internal organs. We report a case of a 12-year-old boy, previously diagnosed with kyphoscoliosis-type EDS (type VI), presenting with a left brachial artery pseudo-aneursym with history of multiple spontaneous and post-traumatic arterial ruptures. Surgical management of this patient was performed successfully by primary repair of brachial artery lesion.

The kyphoscoliotic type of Ehlers–Danlos syndrome (EDS), type VI (MIM 225400), is a rare autosomal recessive connective tissue disorder characterised at birth by severe muscular hypotonia often requiring invasive neuromuscular work-up, kyphoscoliosis which is progressive and severe, marked joint hypermobility and luxations and severe skin hyperelasticity.[1,2] Vascular rupture is a major life-threatening complication in this disorder, but is less frequent than in vascular-type EDS (type IV).

Case Report

A 12-year-old Caucasian male was referred to our outpatient clinic from another cardiovascular surgery centre due to a failed repair for a left brachial artery pseudo-aneurysm with typical clinical presentation and molecular diagnosis of kyphoscoliosis-type EDS (type VI) verified in another university hospital. He is the third child of healthy Turkish parents with no known consanguinity. He was diagnosed at an early age with DNA analysis and urinary pyridinoline investigation at another university hospital. Diagnosis was based on clinical findings observed by a geneticist and confirmed by showing an increased ratio of lysylpyridinoline (LP) to hydroxylysylpyridinoline (HP) cross-links in the urine. Sequencing of *PLOD1* gene revealed a homozygous deletion in exon 13 (c.1362delC), leading to decreased activity of lysyl hydroxylase, an enzyme necessary for collagen biosynthesis.

The patient had a 5 × 5 cm pulsatile mass on the distal left arm in addition to characteristics of the syndrome such as marked joint hypermobility, skin fragility with abnormal scarring and bluish cornea with normal intellect. This patient and four of his siblings showed progressive kyphoscoliosis, joint contractures and hypermobility, and skin laxity (Table 1). He had a history of consecutive surgical repairs for right iliac artery rupture and a left deep femoral artery rupture at the age of 9 and 11, respectively. He had severe kyphoscoliosis and pectus excavatum deformity hindering a straight supine or erect posture (Fig. 1A, B). He also had a severe flexion contracture on the elbow joint of the affected arm.

Computerised arterial tomography (CAT) revealed a 5 × 3 cm pseudo-aneurysm of the left brachial artery (Fig. 1A, C). It was repaired with 6.0 polypropylene sutures under general anaesthesia due to postural deformities. He was discharged from the hospital on day 5. Follow-up CAT scans at 6 months demonstrated a normal arterial tree with no signs of pseudo-aneurysm (Fig. 2).

Discussion

The kyphoscoliotic-type of Ehlers–Danlos syndrome (EDS), type VI (MIM 225400), is a rare autosomal recessive connective tissue disorder, which in the first years of life affects the musculoskeletal

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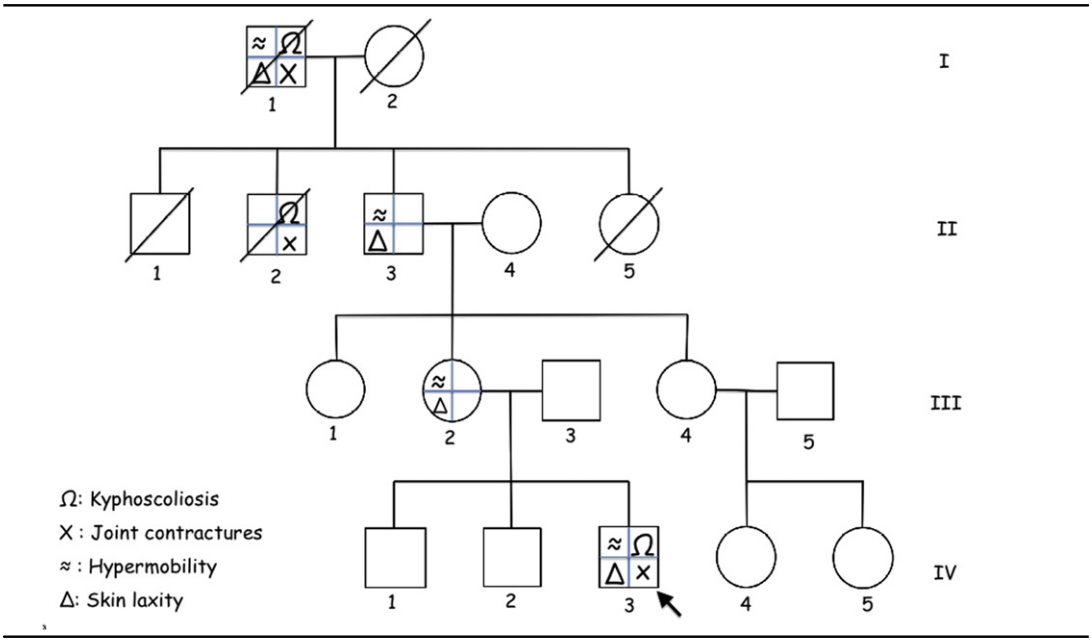
Table 1
Family pedigree of EDS in four generations.



system leading to hypotonia, kyphoscoliosis and joint hyperextensibility.[1,2] Our patient was able to walk with assistance. He had multiple spontaneous or post-traumatic arterial ruptures leading to consecutive operations and no past history of gastrointestinal events or pneumothorax which is among the diagnostic criteria of the autosomal dominant vascular type. His other two brothers did not show any of the phenotypical features of the syndrome and were negative for the mutation.

Vascular events, as commonly seen in the vascular type, may be more frequent than expected in this type of disease.[3] In the vascular type, the generalised vascular fragility largely dominates the clinical picture. Apart from excessive bruising and bleeding, it may lead to arterial rupture, potentially resulting in sudden death, usually in the third or the fourth decade of life. Unlike other EDS types, the skin is not hyperextensible, but rather thin and translucent, showing a visible venous pattern over the chest, abdomen and extremities in the vascular type which is different from that seen in our patient.[3]

In conclusion, this case report emphasised the need to use zonoperative therapy when possible for EDS VI patients with vascular complications. When operative intervention is required, simple vessel repair is more convenient than reconstruction.

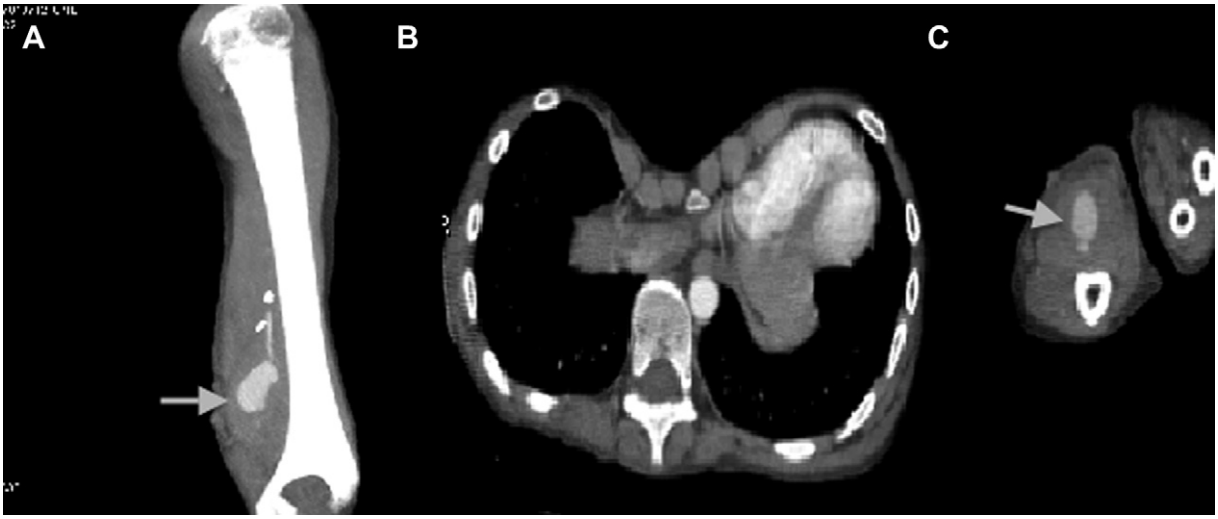


Figure 1. Preoperative CT angiography of the patient. Note the pseudoaneurysm in proximity of the distal brachial artery (A and C). Please, note the asymmetry of the bilateral thoracic cavities reflecting the posture deformity (B).

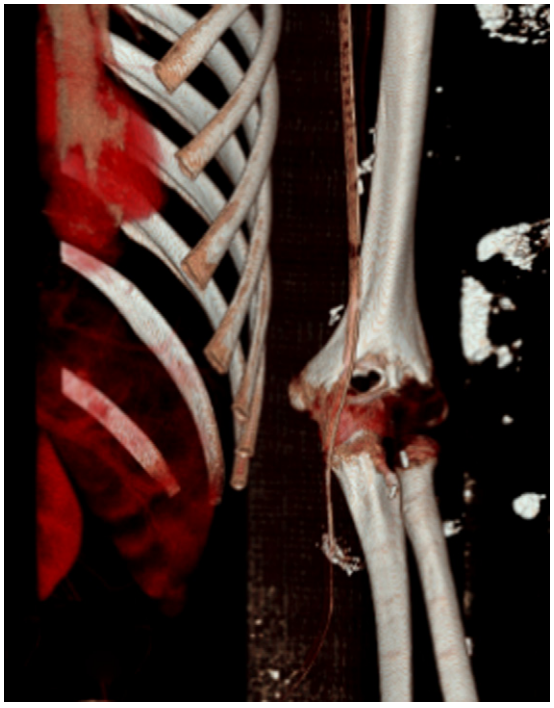


Figure 2. Postoperative CT angiography of the patient 6 months following successful surgical repair of the initial arterial defect.

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