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Case Report

A rare case of arterial tortuosity syndrome in an adult ☆

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ABSTRACT

Arterial tortuosity syndrome (ATS) is rare autosomal recessive connective tissue disorder. It affects large and medium-sized arteries inducing tortuosity and elongation. Typical skeletal manifestations are dysmorphic features, hyperextensible skin, hypermobile joints, and congenital contractures.

We present a case of a 33-year-old female, with history of multiple abdominal wall hernias, who was diagnosed with ATS by preoperative investigations based on typical vascular manifestations. We will present the radiological findings of this rare condition.

Introduction

Arterial tortuosity syndrome (ATS) is rare autosomal recessive connective tissue disorder. ATS was first described by Ertugrul in 1967 [1].

It affects large and medium-sized arteries inducing tortuosity and elongation. Patients can be asymptomatic or present with wide range of manifestations. It is associated with a higher incidence of vascular complications such as aneurysms formation, vascular dissection, and pulmonary arteries stenosis. Typical skeletal manifestations are dysmorphic features, hyperextensible skin, hypermobile joints, and congenital contractures.

It affects both sexes equally and usually presents early depending on the severity of the participation of multiple systems and the existence of complications in childhood [1].

While most cases are diagnosed in infancy, this is a case of a 33-year-old female with manifestations of ATS who was not previously investigated.

Case presentation

We present a 33-year-old female, who presented with a complaint of abdominal pain and swelling and prolonged constipation. She had a past surgical history, abdominal wall hernia repair twice at different sites, umbilical and right inguinal. Examination showed 2 large nonreducible abdominal wall hernias with 2 surgical scars. The patient also had lax hyperextensible skin.

☆ Competing Interests: All authors have no interests to declare.

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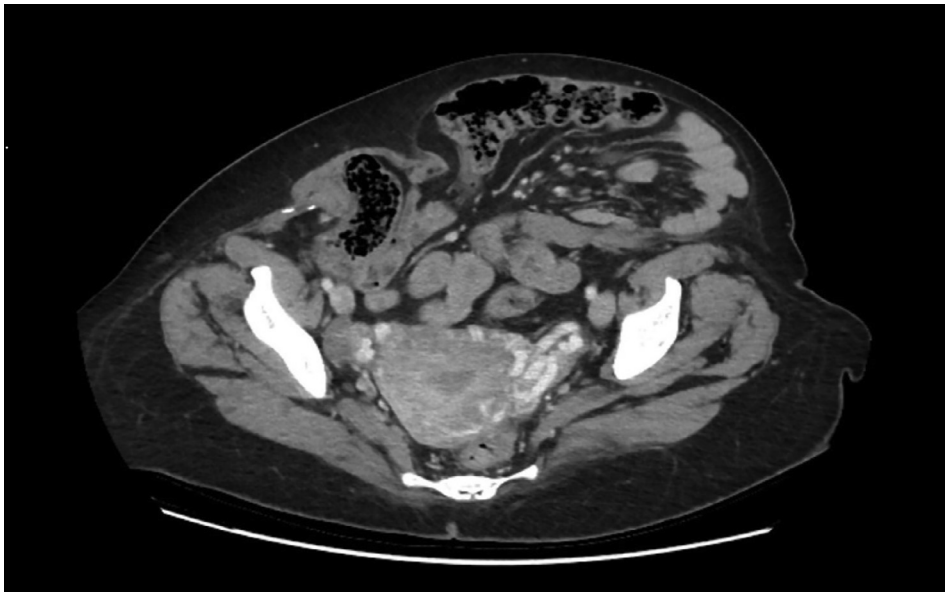

Fig. 1 – Contrast-enhanced CT scan of the abdomen and pelvis showed sizable anterior abdominal wall hernia containing small bowel loops and mesenteric fat. Prominent para uterine veins denoting pelvic venous congestion.

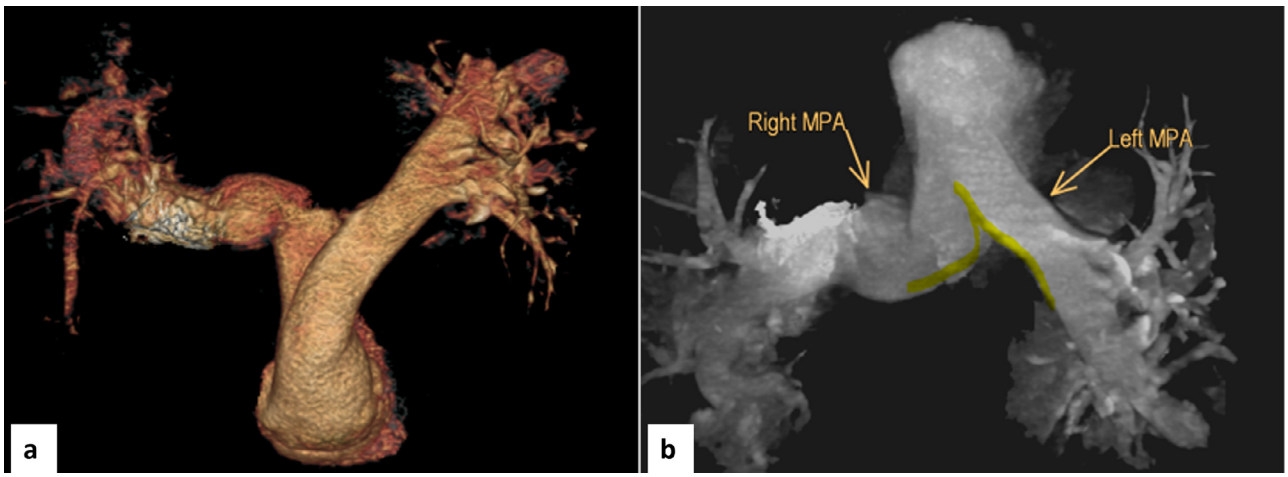

Fig. 2 – Volume-rendered CT pulmonary angiogram (a) and MIP (b) images showing elongation and early branching of pulmonary artery with V-shaped configuration.

The patient underwent CT scan of the abdomen and pelvis with intravenous contrast which showed 2 lower anterior abdominal wall paraumbilical hernias. The larger one is an inferior para umbilical location measuring 17.8 × 17.6 × 8.0 cm. It contains omentum, bowel loops, and mesentery with no signs of strangulation. The other smaller hernia is a supra umbilical fat containing hernia measuring 9.5 × 8.3 × 5.4 cm (Fig. 1). Another recurrent hernia at a previous surgical scar was noted as well at the same level of the large hernia. Abdominal muscle thinning is noted.

Preoperative chest radiography showed widened mediastinum. Evaluation by CT pulmonary angiogram showed pulmonary artery elongation with narrowed bifurcation angle giving inverted V-shaped configuration however with no evidence of stenosis or aneurysmal dilatation (Fig. 2).

CT aortogram showed an elongated, tortuous descending thoracic aorta as it courses anteriorly and to the left to form the aortic arch. The aortic arch is attaining high location seen at the level of the thoracic inlet with abnormal configuration and orientation of its branches originating from its posterior aspect as following:

The left subclavian artery (SCA) arises as to the first branch from the left lateral aspect of the arch and courses superiorly, forming a loop at the root of the neck.

- The left common carotid artery (CCA) arises as to the second branch from the posterior aspect of the aortic arch; it is also elongated and tortuous, forming 2 loops; the first one is directed left postero-laterally then forms the 2nd loop

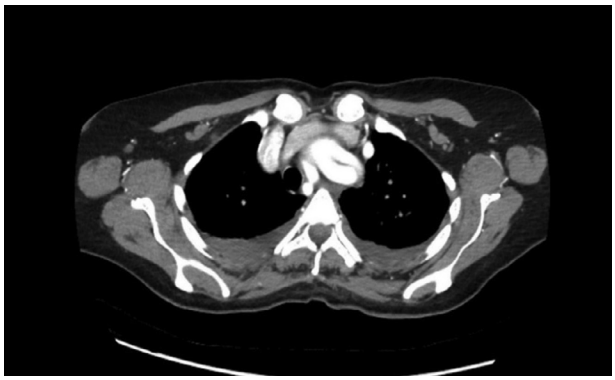

Fig. 3 – Axial CT aortogram showing elongated tortuous left common carotid artery.

has right anterior-medial direction at the root of the neck (Fig. 3).

- The brachiocephalic trunk is arising from the posterior aspect of the aortic arch; shortly, it gives off the right CCA and right SCA; both are elongated, tortuous, and looping at the thoracic inlet.

No appreciable aneurysms, significant stenosis, or cardiomegaly were noted. The trachea was shifted to the right side. The esophagus was encased by the vascular loops. Echocardiography showed left ventricular hyperdynamic wall motion and an overall systolic function of 70%.

The diagnosis of ATS was made due to typical radiological findings. Genetic or molecular testing was not done for the patient.

Discussion

ATS is rare autosomal recessive disorder. The genetic defect in ATS is caused by loss-of-function mutations in the SLC2A10 gene encoding the facilitative glucose transporter GLUT 10 [2].

The central hypothesis is that SLC2A10 expresses a dehydroxyascorbic acid transporter that is present on both mitochondrial and endoplasmic reticulum membranes. Ascorbic acid is required for the maturation of elastin and collagen by acting as a hydroxylation cofactor for prolyl and lysyl residues [3]. Reduced hydroxylation of these residues appears to impair vessel walls due to a disruption in the transforming growth factor-beta pathway caused by GLUT10 deficiency [4,5].

This disorder affects multiple systems, notably the cardiovascular, musculoskeletal, and gastrointestinal systems. Skeletal manifestations include facial dysmorphic features, skin and joint hyperlaxity, chest wall deformity as pectus excavatum and kyphoscoliosis. Gastrointestinal manifestations include esophageal dilatation, elongations, and recurrent ab-

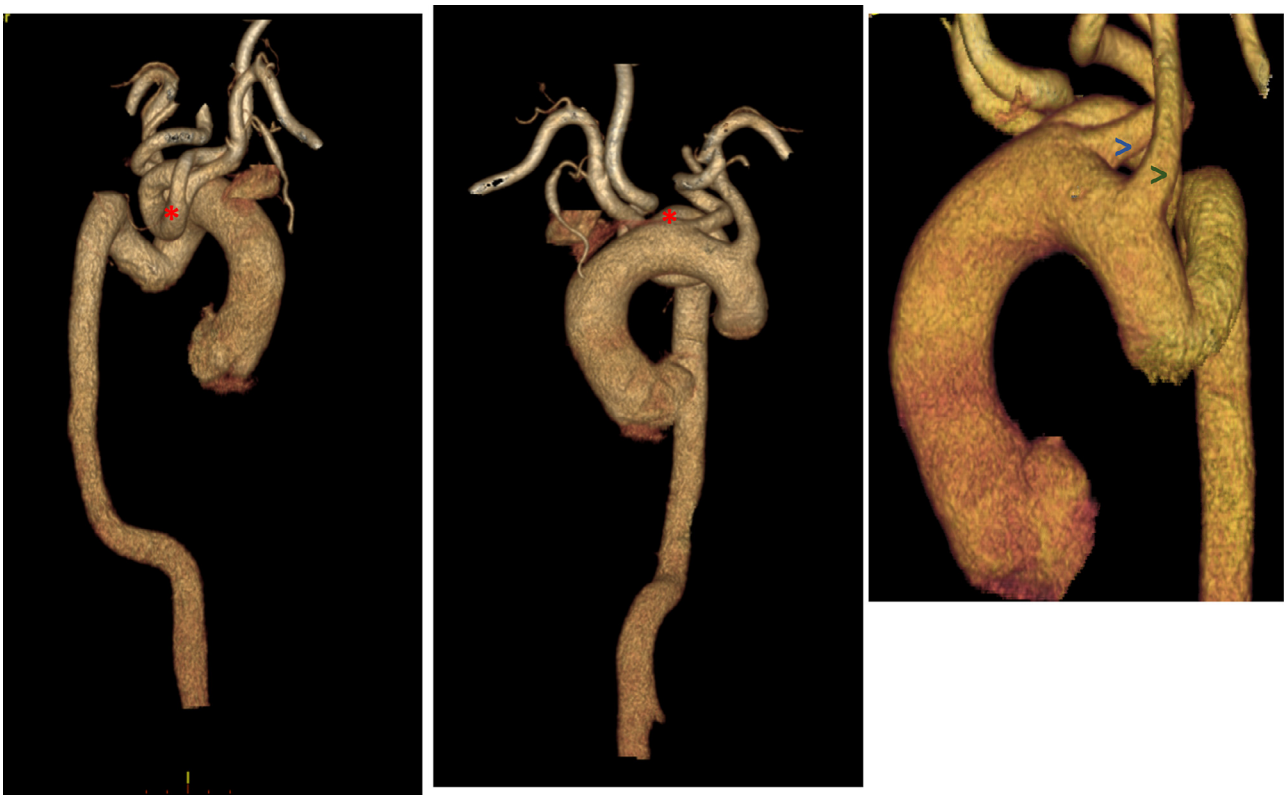

Fig. 4 – CT aortogram volume-rendered scan showing tortuous and mandering descending aorta with looping of its branches. *Brachiocephalic artery > Left CCA > left SCA.

dominal wall hernias as in our case. Morgagni hernia, and less frequently Bochdalek's hernia [2].

Vascular manifestations like tortuosity and elongation are the most common cardiovascular imaging finding, which may be localized or generalized. Usually, it affects large vessels as the aortic arch and its branches, middle-sized vessels as coronary, cerebral, and iliac arteries, and is less likely to affect small-sized arteries such as subungual capillaries and retinal arteries. Intracranial vessels are more susceptible to multiple aneurysms [6].

Arterial tortuosity syndrome has multiple radiological signs. Aortic elongation sign describes prolonged aortic knuckle. A tortuous vessel that extends more than the anticipated arterial course it is described as the mandering vessel [7]. Clustering of vessels is a sign that describes excessive arterial tortuosity on axial contrast-enhanced CT (CECT) images (Fig. 4). V-shaped sign (Fig. 2) describes early pulmonary artery bifurcation with small caliber of main branches appearing on coronal CECT, while inverted V shape sign describes this on axial CECT images.

Tortuosity may attain S- or C-shaped configuration. It might have acute angulation, which is called kinking. Kinking is graded as mild (angle $\geq$60°) to moderate (angle between 30° and 60°) and severe (angle less than 30) [8,9]. Kinking can be seen in the visceral arteries (the celiac and superior mesentery arteries). Excessive tortuosity of the femoral artery can make femoral access difficult. Similarly, there may be problems with venous access [9].

Many syndromes have clinical similarities to ATS and should be differentiated as Marfan syndrome, which has typical marfanoid features, and vascular changes often affect the aortic root in the form of dilatation or aneurysm formation. Other differentials include Loeys-Dietz syndrome and Menkes disease. Pulmonary arteries narrowing is specific to ATS, as well as, diffuse involvement of the aorta, and arch branches [10].

Conclusion

Tortuosity, dilation, aneurysm formation, and stenosis of big and medium-sized arteries are all symptoms of ATS, a rare connective tissue illness. Imaging plays a role in diagnosis and detection of complications if present. Radiologists should be aware of the imaging features of ATS as for timely diagnosis.

Patient consent

A written informed consent was obtained from the patient, consented for publishing patient case details, imaging data, without personal images. The article was approved for publishing by the research and ethics committee of King Hamad University Hospital on January 18, 2022.

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