Diagn Interv Radiol 2010; 16:132–133

CASE REPORT

Cervical aortic arch in a patient with Turner syndrome

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ABSTRACT
We report a case of a young girl with Turner syndrome presenting with a pulsatile left-sided supraclavicular swelling since birth, which proved to be the rare anomaly of a cervical aortic arch. Though elongation of the transverse aortic arch is well known in Turner syndrome, to the best of our knowledge, a cervical aortic arch has not been described in the literature.

Key words: • *aortic arch syndromes* • *magnetic resonance angiography* • *Turner syndrome*

The cervical aortic arch is defined as an arch that is supraclavicular (1). Though often isolated, it may be associated with certain congenital aortic or cardiac anomalies, including ventricular septal defect, double outlet right ventricle, tricuspid atresia, and atrial septal defect (2). Herein, we report a case of a young girl with Turner's syndrome presenting with a pulsatile left-sided supraclavicular swelling since birth, which proved to be the rare anomaly of a cervical aortic arch. Though elongation of the transverse aortic arch is well known in Turner's syndrome, to the best of our knowledge, a cervical aortic arch has not been described in the literature.

Case report

An 18-year-old girl presented with a pulsatile swelling present since birth in the left side of her neck, which had gradually increased in size. There were no specific symptoms related to cardiac disease. She was also under evaluation for delayed mental and physical milestones and absence of secondary sexual characteristics.

On clinical examination, she had a short stature with webbing of the neck. A pulsatile swelling was seen in the left side of the neck. Normal blood pressure was noted with pulses equal in both upper and lower limbs. Cardiovascular examination was normal.

On echocardiography, the left carotid artery was thought to be aneurysmal. On catheter angiography, the catheter could not be advanced beyond the point of a looped segment of the aortic arch.

A contrast-enhanced 3D magnetic resonance angiogram (MRA) was then performed on a 1.5 T magnetic resonance imaging (MRI) scanner (Siemens Avanto, Erlangen, Germany). This revealed a left-sided aortic arch with the right common carotid artery arising from the proximal arch. The arch subsequently passed into the neck up to the C3-C4 level where the left internal and external carotid arteries arose directly from the arch (Fig. 1). The arch then bent acutely inferiorly passing into the upper thorax where it formed a loop from which the left subclavian artery arose. The right subclavian artery was aberrant in origin arising from the descending thoracic aorta. This artery showed a long proximal segment of diffuse stenosis (Fig. 2). The remainder of the descending thoracic and abdominal aorta and its other branches were normal in caliber (Fig. 3).

In view of the fact that her symptoms were minor, the patient was managed conservatively.

Discussion

During embryonic development, there are five or six pairs of aortic arches that connect the truncus arteriosus with the paired dorsal aortae. The normal arch is formed through progressive involution of seg-

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Received 28 April 2008; revision requested 5 June 2008; revision received 5 June 2008; accepted 15 July 2008.

Published online 19 October 2009
DOI 10.4261/1305-3825.DIR.1860-08.1

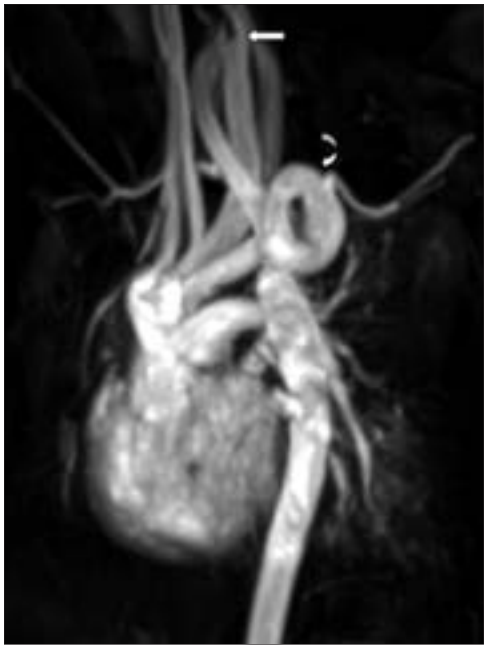

Figure 1. Contrast-enhanced 3D MRA showing the left-sided arch passing into the neck after giving off the right common carotid artery. Direct origin of the internal and external carotid arteries from the superior-most part of the arch is noted *(arrow)* with an acute bend distally, and a loop in the chest *(curved arrow)*.

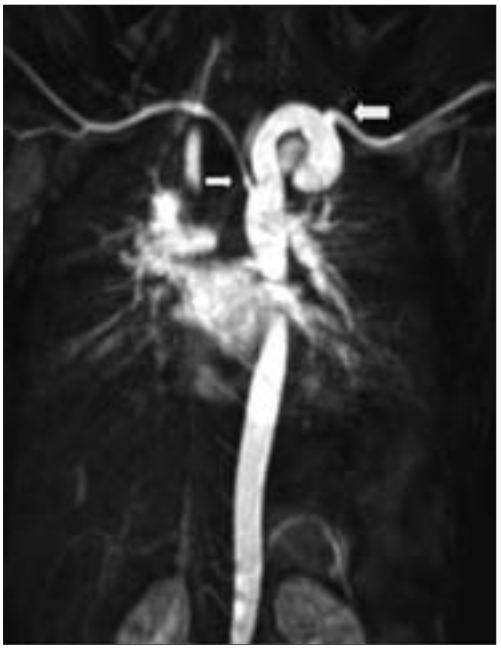

Figure 2. Contrast-enhanced 3DMRA demonstrating the left subclavian artery *(thick arrow)* and the aberrant right subclavian artery *(thin arrow)*, the latter showing a long-segment stenosis proximally.

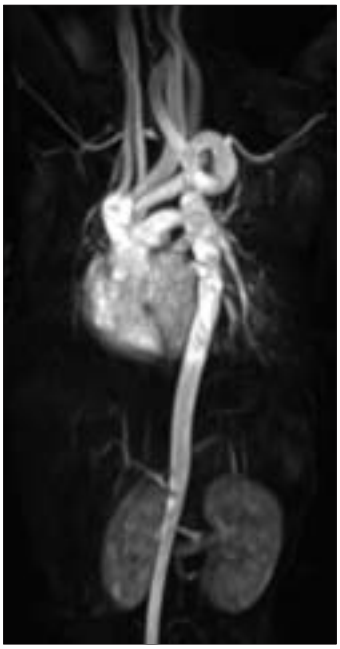

Figure 3. Contrast-enhanced 3D MRA demonstrating the remainder of the descending thoracic and abdominal aorta showing normal caliber and course.

ments of the third and fourth arches and the dorsal aorta, with migration of the arch from the cervical to the thoracic region (3). Theories regarding the etiology of the cervical aortic arch include persistence of the second or third branchial arch and resorption of the fourth arch, which normally forms the aortic arch; failure of the normal fourth arch to descend into the neck; and fusion of the third and fourth arches and failure to descend into the neck (1).

Cervical aortic arch is often asymptomatic, or presents as a pulsatile swelling in the neck. It may also be associated with symptoms related to compression of the trachea or esophagus, such as dyspnea or dysphagia, or recurrent respiratory infections.

Different morphological types of cervical aortic arches have been described with variations in branching. Redundancy of the arch with areas of acute angulation as seen in our case is a known feature with similar findings of acute angulation and bends described by Morris and Ruttley (4). Other associated anomalies of the cervical aortic arch and its branches include aneurysms, coarctation/pseudocoarctation, and stenosis of the arch or its branches, as was seen in our case.

There have been several reports of association of anomalies of the cervical aortic arch with deletions of chromosome 22q11, suggesting that the cervical aortic arch may be part of the DiGeorge spectrum of defects (5). Turner syndrome is well known to be associated with arch anomalies, including elongation of the transverse aortic arch, coarctation of the aorta, and aberrant right subclavian artery (6). Association of Turner syndrome with a cervical aortic arch as seen in our case, however, has not been described in the literature, to the best of our knowledge.

A leading hypothesis considered in the origin of cardiovascular anomalies in Turner syndrome is lymphatic hypoplasia/obstruction, which affects a large number of 45X fetuses (6).

Clinically, cervical arch anomalies, though rare, should be considered in the differential diagnosis of a pulsatile neck mass with the potential for being mistaken for a carotid aneurysm noted.

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