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Case Report

Right-sided cervical aortic arch in Loeys–Dietz syndrome

Behnam Shakerian (MD)[a], Mohammad Hossein Mandegar (MD)[a], Bahieh Moradi (MD)[b,*], Farideh Roshanali (MD, FASE)[b]

[a] Department of Cardiothoracic Surgery, Shariati General Hospital, Tehran University of Medical Science, Tehran, Iran
[b] Department of Echocardiography, Day General Hospital, Tehran, Iran

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ABSTRACT

Loeys–Dietz syndrome is an autosomal dominant connective tissue disorder that is characterized by skeletal abnormalities, craniofacial malformations, and predisposition for aortic aneurysm with tortuosity. We report a case of a right-sided cervical aortic arch associated with the Loeys–Dietz syndrome. To the best of our knowledge, this combination has not been described in the literature.
<**Learning objective:** The Loeys–Dietz syndrome is characterized by vascular findings such as arterial aneurysm or tortuosity with skeletal and craniofacial malformation. We report for the first time a patient with right-sided cervical aortic arch associated with this syndrome. The patient underwent surgical reconstruction.>

Introduction

The Loeys–Dietz syndrome (LDS) is a connective tissue disorder. It is an autosomal dominant disorder caused by heterozygous mutation in the gene that encodes transforming growth factor B (TGFB) receptor type 1 or 2 as well as SMAD3. The LDS is characterized by vascular findings such as arterial aneurysm or tortuosity and skeletal and craniofacial manifestation [1].

Natural history is significant for aortic dissection at smaller aortic diameter and arterial aneurysms throughout the arterial tree [2].

Case report

A 13-year-old girl presented complaining of a pulsatile mass on the right side of her neck since birth, which had gradually increased in size. Her medical history was otherwise unremarkable. On physical examination, she had a short stature, there was a pulsatile swelling on the right side of the neck (Fig. 1), and the blood pressure was 110/70 mmHg with pulses equal in both upper and lower limbs. Cardiac examination was normal. Examination of her head and face revealed an abnormal cranial structure consistent with hypertelorism, retrognathia, and low-set ears, and inspection of her oral cavity showed a bifid uvula and a high arched palate. Skeleton findings included scoliosis and joint laxity. Chest X-ray showed the absence of a normal aortic knob. The echocardiographic study revealed normal cardiac structure. Aortic valve was normal and tricuspid. Computed tomography angiography revealed a right-sided cervical aortic arch (Fig. 2), reaching the upper margin of the C-7 vertebral body and descending to the left of the mid line. The first branch of the ascending aorta was the left common carotid artery; the second branch was the right common carotid artery, arising at the C-5 vertebral level; the third branch was the right subclavian artery; the next branch was the right vertebral artery at the C-7 level; and the last branch was the left subclavian artery. Genetic study with DNA sequencing was performed and revealed a c.1069 G>A/p.G357R missense mutation in TGFBR2 gene.

Cardiac surgery with median sternotomy and extension to the right side of the neck over sternocleidomastoid muscle was performed. The anatomy was confirmed visually (Fig. 3). The ascending aorta and aortic arch in neck was released and aortic arch was dissected. The excess part of cervical aortic arch over right common carotid artery up right subclavian artery was resected. Aortic stump was repaired after the right carotid take-off, and then re-anastomosis of the beginning of descending aorta to the right side of ascending aorta was performed. In this way, the right subclavian artery was saved. Pathological findings were consistent with loss of elastin content and myxoid degeneration of media with subadventitial hemorrhage.

The patient remains in good general condition two years after the operation.

* Corresponding author at: Day General Hospital, Vali-Asr Ave., Abbaspour St., Tehran 1434873111, Iran. Tel.: +98 21 84942875; fax: +98 21 66005214.
E-mail address: faraviolet@yahoo.com (B. Moradi).

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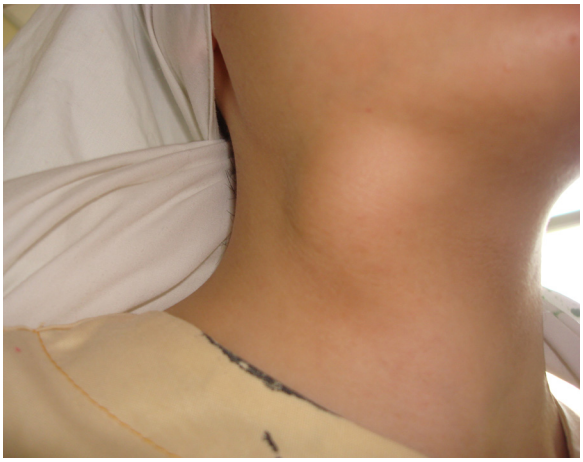

Fig. 1. A pulsatile swelling on the right side of the patient's neck.

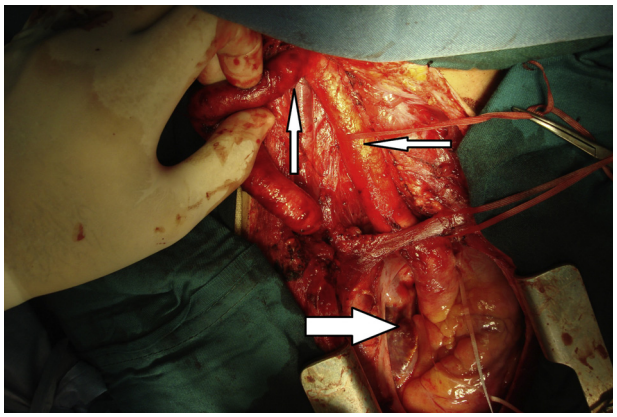

Fig. 3. Surgical view of the heart (thick arrow) and extended median sternotomy to the right side of the neck; the anatomy of cervical aortic arch confirmed visually (narrow arrows).

Discussion

In 2005, Loeys et al. characterized a newly identified genetic syndrome by a triad of hypertelorism, cleft palate or bifid uvula, and arterial tortuosity or aneurysm. Other manifestations include blue sclera, scoliosis, retrognathia, craniosynostosis, pectus deformity, joint laxity, velvety skin, congenital heart defect, and mild developmental delay [1,3]. About 75% of the patients belong to type 1, which presents with craniofacial and vascular disorders. The remaining patients belong to type 2, in which craniofacial abnormalities, with the exception of an isolated bifid uvula, are typically absent. Although the LDS initially was considered as a subgroup of the Marfan syndrome, additional features (e.g. hypertelorism, cleft palate, and uvular abnormalities) along with cardiovascular abnormalities such as tortuosity and widespread aneurysm distinguish it from the Marfan syndrome. Aneurysms are prone to rupture at a smaller size; consequently, the aggressive treatment of aneurysms should be considered [4]. Approximately, half of the individuals with LDS have an aneurysm distant from the aortic root. The diagnosis is suspected by clinical characteristics but confirmed by molecular genetic testing of TGFB receptors [5]. A cervical aortic arch is a rare vascular anomaly. The prevalence of a cervical aortic arch is less than 1 in 10,000 live births [6]. Theories regarding the etiology of the cervical aortic arch include: the persistence of the second or third branchial arch and the resorption of the fourth arch, which normally forms the aortic arch; the failure of the normal fourth arch to descend into the neck; and the fusion of the third and fourth arches and failure to descend into the neck. The cervical aortic arch is often asymptomatic or presents as a pulsatile neck mass, dysphagia, dyspnea, discrepancy of arterial blood pressure between the upper and lower extremities, cough, hoarseness, and mild stroke. The recognized cardiac anomalies associated with the cervical aortic arch are ventricular septal defect, tetralogy of Fallot, pulmonary atresia with ventricular septal defect, and double-outlet right ventricle without pulmonary stenosis [7]. The interesting feature in our case is the occurrence of the cervical aortic arch in the LDS. In this case because of growing neck mass and high risk of dissection of aorta, as well as the annoying appearance and risk of cervical aortic rupture during physical activity, exercise, and probable trauma, surgical reconstruction was undertaken.

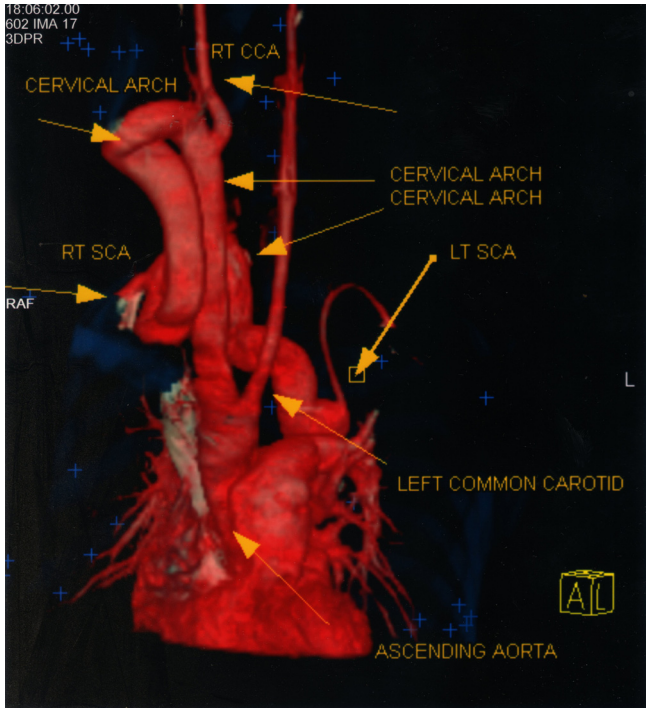

Fig. 2. A three-dimensional image of computed tomography angiography: ascending aorta, large cervical aortic arch, and branches (left and right common carotid arteries, left and right subclavian arteries) are shown. LT SCA, left subclavian artery; RT SCA, right subclavian artery; RT CCA, right common carotid artery.

Conflict of interest

Authors declare no conflict of interest.

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