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Case Report

Arterial tortuosity syndrome presented as nonvisualization of thoracic aorta in preoperative transthoracic echocardiogram: A case report ☆,☆☆

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ABSTRACT

Arterial tortuosity syndrome is a rare genetic disorder characterized by dilation, elongation, and significant tortuosity of major arteries. Approximately 100 cases of this disorder have been reported worldwide, including 3 reports in Iran. We describe a case of arterial tortuosity syndrome suspected during the preoperative evaluation for hypertrophic pyloric stenosis, where the thoracic aorta was not visualized appropriately in transthoracic echocardiography. Our report focuses on identifying the disease through diagnostic imaging.

Introduction

Arterial tortuosity syndrome is a rare genetic disorder characterized by significant tortuosity and dilation of major arteries, including the aortic arch and its branches- along with several other connective tissue abnormalities throughout the body [1]. A literature review reveals approximately 100 cases of this disorder, with very few reports of concomitant hypertrophic pyloric stenosis [2,3]. In this report, we present a case of arterial tortuosity syndrome diagnosed with preoperative CT angiography.

Case report

A 4-week-old male infant was presented to our center by his parents with the complaint of projectile vomiting. He was the third child of consanguineous parents with 2 previous healthy

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children. He was born full term via an uncomplicated Cesarean section.

The patient was symptom-free until 8 days before admission when he started to have projectile non-bilious vomiting episodes after feeding.

On physical examination, he was noticed to have dysmorphic facial features, such a high palate, micrognathia, large ears, and an elongated face. Heart sounds were regular without any specific murmurs. There was a significant blood pressure discrepancy of 90-95 mmHg systolic in the upper extremities and 60 mmHg systolic in the lower extremities.

An abdominal ultrasound exam revealed elongation of the pyloric canal (18 mm), associated with increased muscle thickness (3.8 mm), suggestive of hypertrophic pyloric stenosis (HPS); and a pyloromyotomy surgery was planned.

Due to the blood pressure discrepancy, a preoperative chest x-ray and echocardiogram were done. The chest X-ray did not reveal significant abnormality and was interpreted as a normal neonatal chest (Fig. 1). Transthoracic echocardiography showed normal heart structure and function, although the aortic arch and descending aorta were not visualized properly.

Computed tomography angiography (CTA) was performed for further evaluation of possible congenital vascular anomalies of the aortic arch and thoracic aorta. Imaging findings showed dilation, elongation, and significant tortuosity of the aortic arch and its branches, including bilateral common carotid, subclavian, intercostal, and internal mammary arteries, associated with the tortuous course of the right pulmonary artery. The tortuosity and elongation of the aortic arch had caused multiple kinking in its course (Fig. 2). Similar changes were also visible in the abdominal aorta and all its branches (Fig. 3). The hypertrophied pyloric canal muscle was also visualized in sections of the abdomen (Fig. 3).

Arterial tortuosity syndrome was suggested based on characteristic CTA findings. A retrograde detailed history revealed that the patient's brother was previously evaluated for vascular problems after the detection of a heart murmur during his routine examinations, in which he was found to have significant tortuosity of the aortic arch and thoracic aorta (medical images are not available).

A pyloromyotomy surgery was performed, and the patient recovered uneventfully.

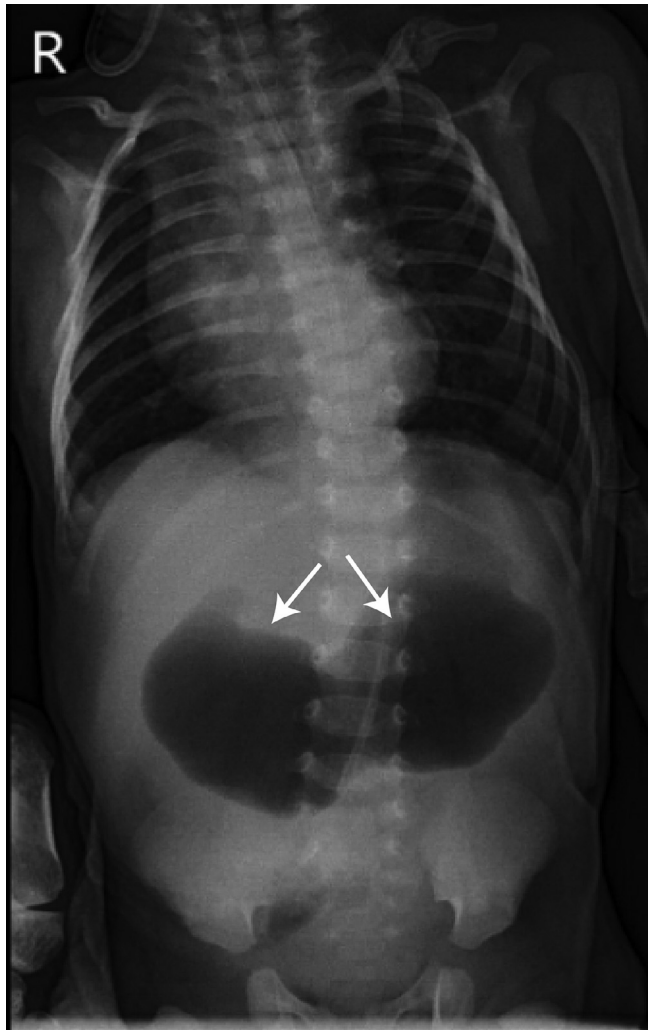

Fig. 1 – Chest X-ray shows the normal radiographic appearance of the heart and pulmonary vascularity. A distended stomach (white arrows) is apparent in the abdomen, consistent with the patient's hypertrophic pyloric stenosis.

Discussion

Arterial tortuosity syndrome is a rare congenital disorder characterized by significant elongation and tortuosity of large arteries, mostly the aorta and pulmonary arteries. Approximately 100 cases of this syndrome are reported in the literature [1], including only 3 cases in Iranian patients [4–6]. Our case is the second reported pediatric patient with arterial tortuosity syndrome in the Iranian population.

Clinical presentations of this syndrome include characteristic craniofacial features (including a long face and hypertelorism) and variable connective tissue presentations; such as skin hyperelasticity or inguinal and diaphragmatic hernias. The association of arterial tortuosity syndrome with HPS has been reported in a few cases [7]. This association can be justified as part of the connective tissue disorders of this syndrome [2,3]. Also, the concomitant occurrence of HPS with various congenital heart defects has been rationalized in several previous studies based on common genetic pathways in the development of the pyloric muscle and the heart and vascular structures [8,9]. However, due to the rarity of this syndrome, and even rarer association with HPS, it is not possible to confidently judge whether or not this association is incidental, and further studies are required on this matter.

Genetically, arterial tortuosity syndrome is caused by mutations in the *SLC2A10* gene, which has a role in encoding the glucose transporter GLUT10. The exact pathogenic mechanism of these mutations remains unclear; however, it is hypothesized that GLUT10 has a role in elastin and collagen maturation through its activities in the endoplasmic reticulum of the cells. Hence, its mutations can lead to the fragmentation of the elastic fibers in the medial layer of the arterial wall, a

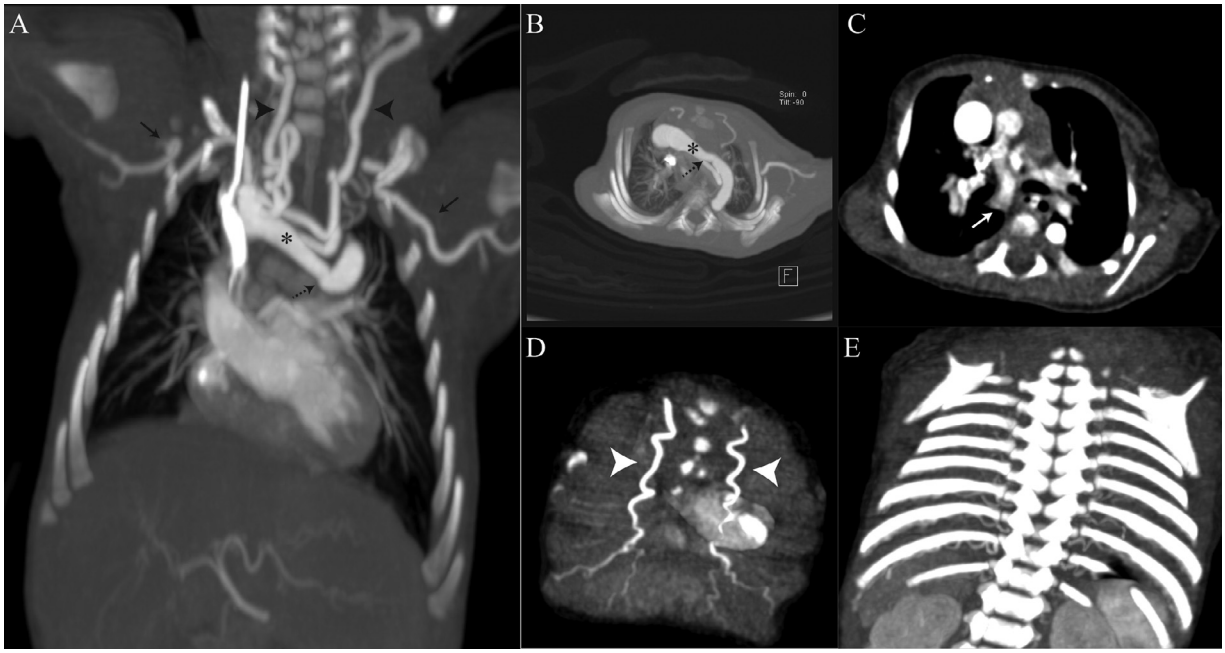

Fig. 2 – Images from CTA of the thoracic aorta and pulmonary arteries show dilation, elongation, and significant tortuosity of the aortic arch (asterisk, A and B) and its branches, including bilateral common carotid arteries (black arrowheads, A) and subclavian arteries (black arrows, A), internal mammary arteries (white arrowheads, D), and intercostal arteries (E), associated with the tortuous course of the right pulmonary artery (white arrow, C). The tortuosity and elongation of the aortic arch cause multiple kinking in its course (dashed arrows, A and B).

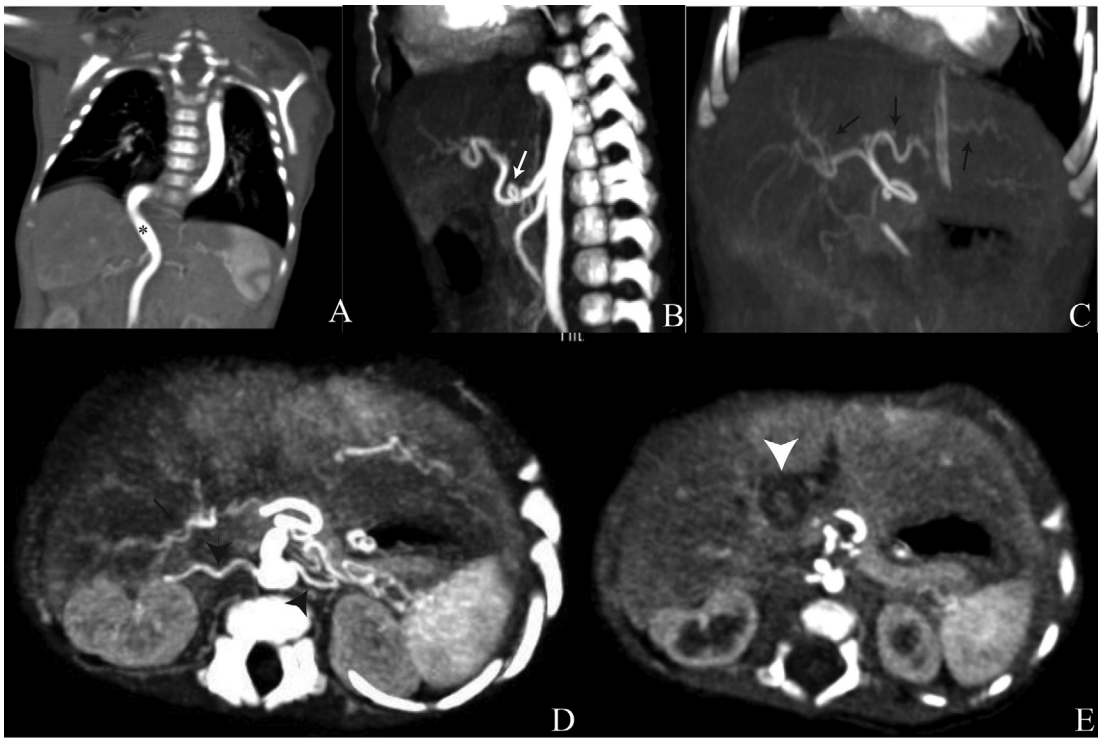

Fig. 3 – Images from CTA of the thoracic and abdominal aorta show significant tortuosity in the abdominal aorta (asterisk, A) and its branches, including the celiac artery (white arrow, B) intrahepatic branches of the proper hepatic artery (black arrows, C and D) and renal arteries (black arrowheads, D). The hypertrophied pyloric canal muscle is also visualized in the sections of the abdomen (white arrowhead, E).

finding observed in the histopathologic evaluation of arterial walls in patients with ATS [1].

Cross-sectional Imaging findings of this syndrome have been described in very few reports, with most reports concentrating on genetic and clinical findings. Conventional and CT angiographic findings of this syndrome include elongation and tortuosity of major arteries, mostly the aortic arch and its branches, and the pulmonary arteries, which can lead to multiple arterial stenoses due to kinking of vessels [10]. Our case excellently demonstrates the CT angiographic findings of arterial tortuosity syndrome, significant dilation and tortuosity of the aortic arch and all of its branches, including intercostal and internal mammary arteries, associated with a tortuous course of the right pulmonary artery. Multiple kinking in the aortic arch was also present, which could potentially be the cause of the blood pressure discrepancy between the upper and lower limbs.

An interesting imaging point in our case is the demonstration of concomitant significant tortuosity in the abdominal aorta and its branches, including intrahepatic branches of the proper hepatic artery, and renal arteries. Our case also highlights the unusual clinical presentations and the diagnostic challenges that can be encountered in this syndrome due to the abnormal course of the tortuous thoracic aorta, such as blood pressure discrepancy and nonvisualization of the thoracic aorta in transthoracic echocardiography. This unique presentation has not been described in previous cases.

Conclusion

Arterial tortuosity syndrome can cause diagnostic challenges related to an abnormal course of the aortic arch and other major arteries. Familiarity with this entity could help physicians manage further diagnostic workups. CT angiography can accurately demonstrate the position and anatomy of the aortic arch and thoracic aorta and can be problem-solving in challenging cases of a non-diagnostic transthoracic echocardiogram.

Patient consent

Written informed consent was obtained from the patient's guardian for publication of clinical data and diagnostic images.

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