Radiology Case Reports 13 (2018) 295–298

Available online at www.sciencedirect.com

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Pediatric

Arterial tortuosity syndrome: An extremely rare disease presenting as a mimic of pulmonary sling

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ARTICLE INFO

Article history:
Received 29 August 2017
Received in revised form 19 October 2017
Accepted 30 October 2017
Available online 13 December 2017

Keywords:
Arterial tortuosity syndrome
Pulmonary sling

ABSTRACT

Pulmonary sling is the anatomic variant defined by the aberrant origin of the left pulmonary artery from the right pulmonary artery. This patient presented with a mimic of pulmonary sling as a result of an extremely rare condition, arterial tortuosity syndrome (ATS). The patient was first diagnosed with pulmonary sling on prenatal echocardiogram performed by cardiology. Computed tomography angiography of the chest obtained at birth to evaluate respiratory depression demonstrated ATS. The early detection of ATS has been demonstrated to improve patient outcome. This case provides an overview of the typical imaging features of ATS to aid radiologists in making this uncommon diagnosis.

Introduction

Pulmonary sling is the anatomic variant defined by the aberrant origin of the left pulmonary artery from the right pulmonary artery, which courses between the trachea and the esophagus. This anatomy often results in stridor and respiratory distress because of posterior compression of the trachea. Pulmonary sling additionally results in anterior impression of the esophagus. These features differentiate the pulmonary sling from the pulmonary rings, which result in anterior compression of the trachea and posterior impression of the esophagus.

A failure of formation of the sixth aortic arch is postulated to be the cause of pulmonary sling. The course of the pulmonary artery adjacent to the right mainstem bronchus can result in compression and air trapping in the right lung. An aberrant left pulmonary artery is associated with complete tracheal rings, which may result in a long-segment tracheal stenosis, leaving patients at higher risk of respiratory compromise [1].

Alternative causes of pulmonary sling symptomatology and pathophysiology are rarely reported in the literature. One cause identified includes duplication of the left pulmonary artery, another rare vascular anomaly [2]. Symptomatic patients with pulmonary sling are surgically managed shortly after presentation because of the high mortality associated with this condition [3].

Case report

A 27-year-old G1P001 woman with a history of chronic hypertension underwent fetal echocardiogram with pediatric

Competing Interests: The authors have declared that no competing interests exist.

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https://doi.org/10.1016/j.radcr.2017.10.023

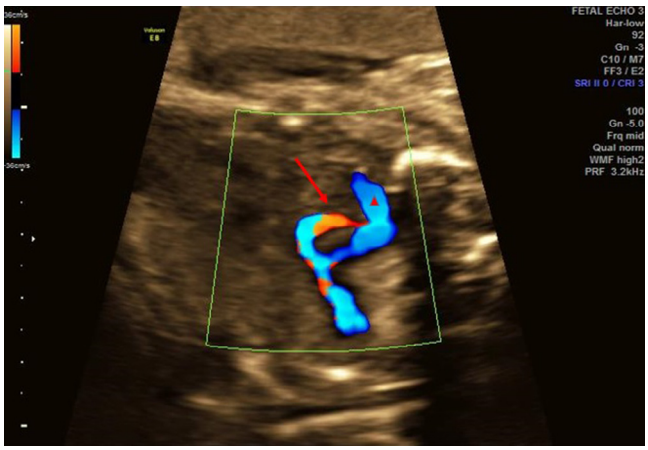

Fig. 1 – Fetal echocardiogram at 28 weeks and 4 days' gestation demonstrates an aberrant vessel with the appearance of a pulmonary sling. The arrowhead marks the main pulmonary artery, whereas the arrow marks the aberrant vessel, presumed to be the left pulmonary artery originating from the right pulmonary artery.

cardiology at 28 weeks and 4 days' gestation that demonstrated findings of a pulmonary artery sling (Fig. 1). Additional prenatal evaluation included ultrasound at 19 and 28 weeks performed by maternal fetal medicine that demonstrated a small appearance of the fetal stomach and no other evidence of fetal structural malformation.

The mother was Group B Streptococcus positive and her pregnancy was further complicated by chorioamnionitis. The mother ultimately underwent emergent cesarean section at 37 weeks and 2 days for a prolapsed cord. The neonate was a 3.21-kg male with Apgars of 5 and 7 at 1 and 5 minutes (decreased tone, little spontaneous activity, and poor respiratory effort). The patient required positive-pressure ventilation and was admitted to the neonatal intensive care unit (NICU) for continuous positive airway pressure.

Given the prenatal findings of pulmonary artery sling and respiratory depression at birth, the patient underwent a computed tomography angiography (CTA) using our congenital heart protocol on the first day of life (Figs. 2 and 3). Significant vascular findings included a markedly tortuous aorta, a dilated main pulmonary artery, and patent ductus arteriosus.

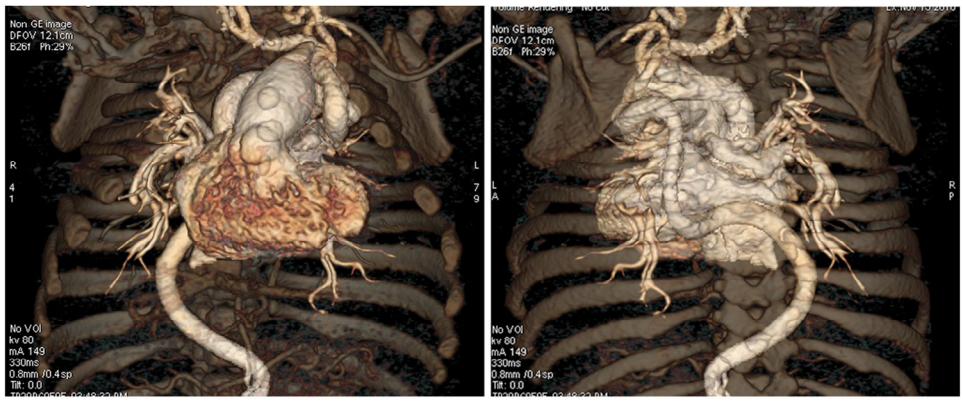

Fig. 2 – Anterior and posterior projections of the 3-dimensional reconstructions of the heart and vessels from computed tomography angiography congenital heart protocol obtained on day 1 of life demonstrate a marked tortuosity of the aorta, patent ductus arteriosus, and a dilation of the main pulmonary artery. DFOV, display field of view; GE, General Electric.

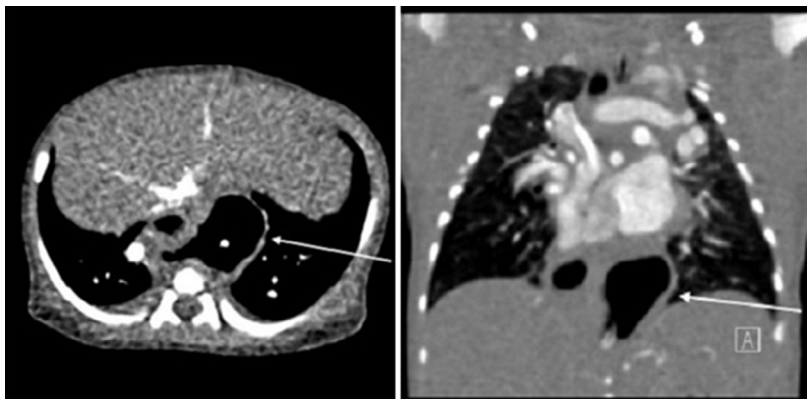

Fig. 3 – Axial and coronal images from a computed tomography angiography congenital heart protocol obtained on day 1 of life also demonstrate a large hiatal hernia with nasogastric tube in place. The hiatal hernia is marked with arrows.

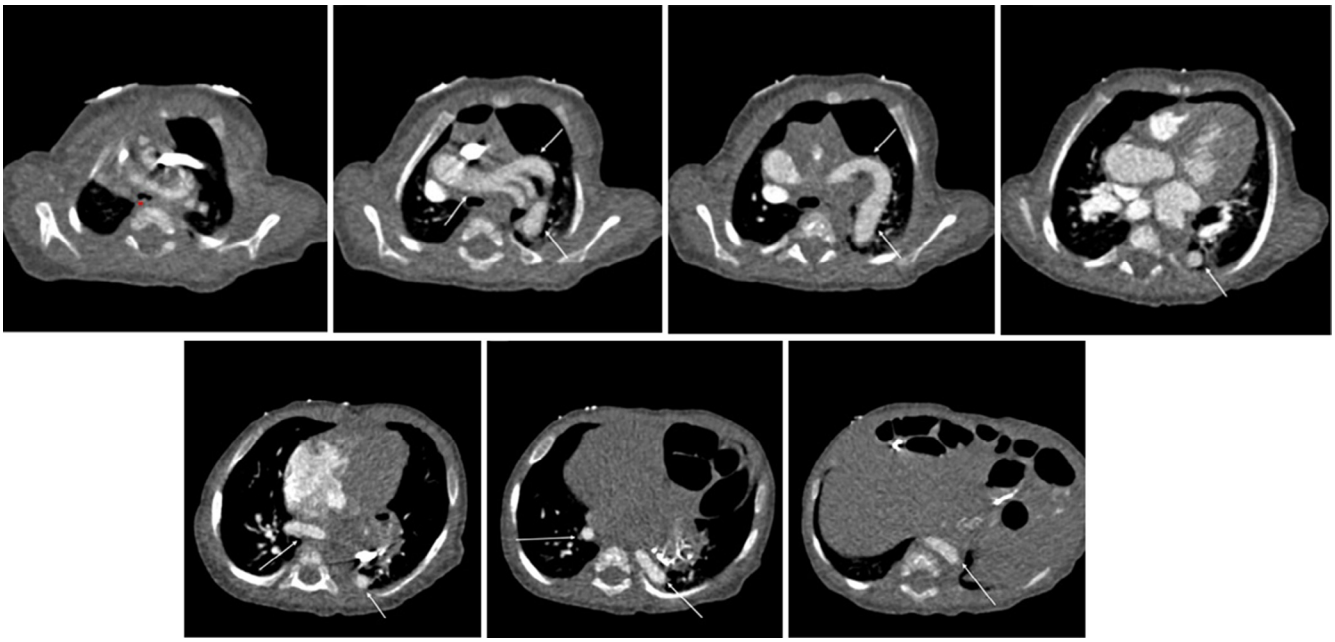

Fig. 4 – Sequential axial images from a computed tomography angiography congenital heart protocol obtained at 8 weeks old. The first image demonstrates severe tracheal stenosis, marked with an arrowhead. Subsequent images demonstrate marked tortuosity of the thoracic aorta, marked by arrows, which crosses from left to right 3 times during its course through the thorax.

CTA additionally demonstrated a large hiatal hernia not identified on prenatal ultrasound. The patient was given prostaglandins and remained in the NICU for monitoring, given concern that arch obstruction could occur as the ductus closed. Magnetic resonance angiography of the brain and renal ultrasound on day 2 of life did not demonstrate additional vascular abnormalities. The patient remained stable and was transferred to the floor. The patient had emesis with oral feeds and required supplementation with nasogastric tube feeds. He underwent Nissen fundoplication and gastrostomy tube placement on day 15 of life and was discharged from the hospital shortly thereafter. He underwent elective left inguinal and umbilical hernia repairs at 8 weeks old.

The patient represented at 9 weeks old after an episode of cyanosis and apnea. Bronchoscopy demonstrated an 80% obstruction of the trachea with secondary tracheomalacia. He underwent repeat CTA congenital heart that again demonstrated marked tortuosity of the thoracic aorta with interval closure of the ductus arteriosus, a normal caliber of the main pulmonary artery, and severe tracheal stenosis (Fig. 4). Additional radiological workup included barium esophagram, which demonstrated an anterior impression on the esophagus (Fig. 5). Given these findings, the decision was made to proceed with aortopexy. The patient had an uneventful postoperative course and was discharged.

At 4 months old, the patient developed recurrent emesis and decreased tolerance of tube feeds. He underwent a redo open Nissen fundoplication with hiatal hernia repair. His postoperative course was uneventful and he was discharged home after routine care. At his wellness visit at 6 months of age, the patient had developmental delay, but was otherwise asymptomatic. He was using his gastrostomy tube for medications only. His growth velocity was appropriate. He continues to undergo follow-up with cardiology and ophthalmology evaluation had been scheduled. Genetics is also involved in his care.

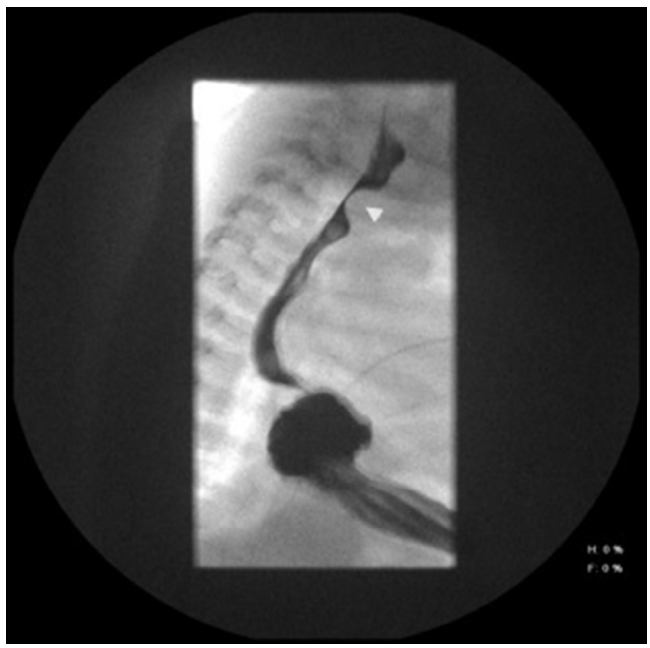

Fig. 5 – Lateral view from fluoroscopic images obtained during a barium esophagram at 9 weeks old demonstrates a fixed anterior impression on the esophagus, marked with an arrowhead.

Discussion

Arterial tortuosity syndrome (ATS) is a rare autosomal recessive connective tissue disease caused by a mutation of the SLC2A10 gene. Associated anomalies are similar to sequelae in patients with other connective tissue disorders, including joint laxity, pes excavatum, hernias, and ocular abnormalities. Patients additionally demonstrate atypical facial features, including micrognathia, large ears, and a long face [4]. A review of the English language literature did not disclose any additional cases in which this syndrome resulted in a pulmonary sling pathophysiology.

Our patient's diagnosis was confirmed with genetic testing. He demonstrated many of the typical features of ATS, including complex cardiovascular disease, inguinal and hiatal hernias, and characteristic facial features. Early intervention to prevent complications related to the abnormal vasculature improves prognosis [5]. Active surveillance of these patients includes annual echo and magnetic resonance imaging or computed tomography every 3 years as patients are at increased risk of developing aneurysms and dissections that require treatment. Routine ophthalmologic visits to evaluate for keratoconus (thinning of the cornea) and orthopedic evaluation during periods of rapid growth are additionally recommended to monitor patients for developing complications.

This is presumed to be the first case of ATS presenting as a mimic of pulmonary sling by echocardiogram. As in this case, the condition may be mistaken for more common congenital heart conditions or other connective tissue disorders. Once suspected, the diagnosis of ATS can be established with genetic testing. Although a rare condition, the proper diagnosis of ATS is essential to ensure proper monitoring of these medically complex patients.

REFERENCES

[1] Berdon WE. Rings, slings, and other things: vascular compression of the infant trachea updated from the midcentury to the millennium—the legacy of Robert E. Gross, MD, and Edward B. D. Neuhauser, MD. Radiology 2000;216(3):624–32. PubMed PMID: 10966687.
[2] Giudici V, Kanani M, Muthialu N, Carr M, Calder AD, Owens CM, et al. Duplicated left pulmonary artery: an unknown disease? Three case reports and review of the literature. Cardiol Young 2016;26(2):340–436. PubMed PMID: 25739970.
[3] Fiore AC, Brown JW, Weber TR, Turrentine MW. Surgical treatment of pulmonary artery sling and tracheal stenosis. Ann Thorac Surg 2005;79(1):38–46. PubMed PMID: 15620911.
[4] Satish G, Nampoothiri S, Kappanayil M. Arterial tortuosity syndrome: phenotypic features and cardiovascular manifestations. Circulation 2008;117(23):477–8. PubMed PMID: 18541745.
[5] Pichler K, Ralser E, Resch M, Steichen-Gersdorf E, Maurer K, Trawoger R, et al. A newborn with arterial tortuosity syndrome: the importance of timely diagnostic work-up in patients presenting with cutis laxa. J Funct Morphol Kinesiol 2016;1(2):249–53.