CASE REPORT

Interrupted aortic arch with isolated persistent left superior vena cava in patient with Turners syndrome

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ABSTRACT

We present a case of 13-year-old female with Turner syndrome (TS), who presented with unexplained lower limbs swelling and ejection systolic murmur at the left second intercostal space. Suspicion of mild aortic coarctation was made by echocardiography. Computed tomography angiography (CTA) showed a complete interruption of the aortic arch (IAA) below the left subclavian artery with persistent left superior vena cava (PLSVC) and absent right SVC, defined as an isolated PLSVC. The patient underwent successful surgical correction after unsuccessful trial of transcatheter stent placement. We present this case of asymptomatic IAA to draw attention to the importance of CTA in diagnosing such rare anomalies and ruling out asymptomatic major cardiovascular abnormalities in patient with TS.

Key words: Coarctation of aorta, interrupted aortic arch, left-sided superior vena cava, Turner syndrome

INTRODUCTION

The prevalence of Turner syndrome (TS) is approximately 50/100,000 female. It is characterized by the absence of an X chromosome that results in features like, but not limited to, short stature, webbed neck, primary amenorrhea, and cardiovascular anomaly.[1] The prevalence of cardiovascular malformations ranges from 17% to 45% of TS patients, with the aortic coarctation and bicuspid aortic valve being the most common. Complete interruption of aortic arch (IAA) is a quite rare congenital anomaly, seen in 3 per million live births, and characterized by luminal discontinuity between ascending and descending aorta.[2] It can be isolated or associated with chromosomal abnormalities such as DiGeorge syndrome and rarely with TS.[3,4]

CASE REPORT

A 13-year-old female patient who is known to have TS was referred to the outpatient clinic with a complaint of both lower limbs swelling for the past 8 weeks. The patient was otherwise asymptomatic with no history of dyspnea or lower limb claudication. Physical examination showed a short stature, webbed neck, ejection systolic murmur 2/6 at the left second intercostal space, and nonpitting edema of both lower limbs. A difference of 20 mmHg in systolic blood pressure between upper and lower limbs and diminished pulses in the lower extremities were found.

Laboratory test results were normal, and electrocardiogram showed left ventricular hypertrophy (LVH). Transthoracic echocardiography showed LVH with suspicion of aortic coarctation and detectable pressure gradient at the descending aorta of 20 mmHg. Computed tomography angiography (CTA) of the aorta showed complete IAA with collaterals involve the vertebrobasilar system, and persistent left-sided superior vena cava (PLSVC) [Figure 1a-c].

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Left heart catheterization showed the same finding [Figure 2a-d]. The patient was referred for a trial of dilation and stent placement in the descending aorta at the site of interruption but, unfortunately, it was unsuccessful and complicated by dissection in one of the three large collaterals. Three months later, the patient was referred for surgery and underwent successful surgical repair where all collaterals were divided with resection, and end-to-end anastomosis of juxtaductal interruption was carried out via left thoracotomy. The surgical procedure was uneventful, and the patient was discharged with no perioperative complications.

DISCUSSION

TS is a common sex chromosomal abnormality that occurs in approximately 50/100,000 female and can be associated with multiple cardiovascular abnormalities, commonly bicuspid aortic valve, and coarctation of the aorta.[1] Extremely uncommon, TS can be associated with complete IAA. The incidence of complete IAA, which is a rare congenital malformation characterized by luminal discontinuity of ascending and descending aorta, is about 3 per million live births.[2] It is considered incompatible with life if the ductus arteriosus closes and no correction by surgical intervention made. There are three types of IAA according to the site of interruption. Type A (43%) interruption occurs distal to the origin of the left subclavian artery, Type B interruption, the most common type (53%), occurs between the left carotid and left subclavian artery, and Type C where the interruption occurs between the innominate and left carotid artery (4%).[5] It can be isolated or associated with chromosomal abnormalities, commonly DiGeorge syndrome. Rarely, IAA can be associated with TS, more frequently the 45, X karyotype. The form of IAA associated with other cardiovascular malformations, such as ventricular septal defect, transposition of great arteries, aortic stenosis, bicuspid aortic valve, truncus arteriosus is more common than the isolated form of complete IAA.[2,6]

In the neonatal or early childhood period, patient presents with symptoms of congestive heart failure which carries poor prognosis with approximately 90% mortality at a median age of 4–10 days without treatment, usually after the physiologic closure of patent ductus arteriosus.[7] In adult patients, the presence of collateral circulation helps to maintain blood flow to the organs and enable survival. However, these collateral vessels are vulnerable to pathological changes, such as atherosclerosis and atrophy, which results in the appearance of symptoms. Although patient with IAA can be asymptomatic, symptoms are usually due to refractory hypertension, congestive cardiac failure, claudication, and aortic insufficiency. Other reported presentations include coronary artery disease, intracranial hemorrhage, and biventricular heart failure.[8] A prenatal diagnosis of IAA can be suspected on echocardiographic examination. In addition, diagnosis of thymic hypoplasia or aplasia during fetal echocardiography can help identifying a high-risk group for 22q11 deletion and possible associated IAA and then postnatal intervention.

In adult, Imaging modalities such as echocardiography can be used as well, but gadolinium-enhanced three-dimensional magnetic resonance angiography (MRA), multislice CTA,

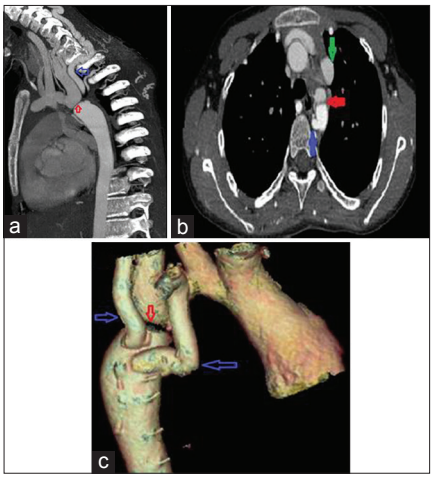

Figure 1: Computed tomography angiography of the aorta images (a-c) showing interrupted aortic arch Type A (red arrow), with complete discontinuity of the aortic lumen distal to the origin of the left subclavian artery, two large collateral vessels involve the vertebrobasilar circulation bilaterally connected to the distal aorta (blue arrow) and isolated persistent left superior vena cava (green arrow)

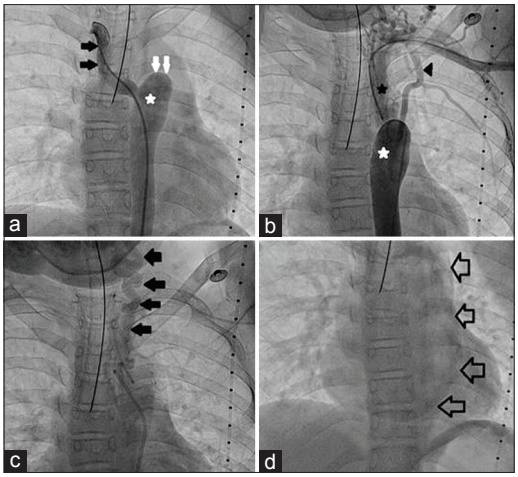

Figure 2: (a) Percutaneous angiography of the distal part of the aorta (white asterisk), with the contrast, injected through femoral access, showing large collateral form vertebrabasilar circulation (closed black arrow) and the site of interruption (white arrow). (b) Early and (c) late images of simultaneous contrast injection to proximal (black asterisk) and the distal part of the aorta through radial and femoral access, respectively; collateral through left internal mammary artery (black arrowhead). (d) Venogram with contrast through the right cubital vein showing the left superior vena cava (open black arrows)

and percutaneous angiography are more reliable for diagnosis of IAA.[8,9]

Treatment in infants and adults appears to be the same. Surgical correction with sternotomy or lateral thoracotomy, either single or two stages approach, can be performed. Percutaneous wire perforation, with subsequent dilation, and graft stent placement within the aorta have been reported as well.[7]

PLSVC with the absence of the right SVC is defined as isolated PLSVC. It is a rare congenital anomalous of the venous drainage that usually occurs in 0.09–0.13% of patients. Echocardiography may show a dilated coronary sinus that can be confirmed by performing a bilateral "bubble study" with injection of agitated saline from both peripheral arm veins. Usually, variation in venous drainage is physiologically not important, but should be recognized before some intervention, such as radiofrequency ablation, permanent pacemaker implantation or cardiopulmonary bypass surgery, to avoid procedure-related complications.[10]

CONCLUSION

Although extremely rare, IAA should be considered in the differential diagnosis along with aortic coarctation in adult patient. It is one of the rare cardiovascular malformations seen in TS. Surgical intervention is necessary to prevent fatal complication or death and improve patient's symptoms and quality of life. It is based on the associated conditions and anatomy. Percutaneous stent placement appears to be an effective mean of repairing IAA in the adult patient as well. The role of multislice CTA is important in evaluating such rare anomalies, especially when a clear diagnosis cannot be made by other tests, for example, echocardiography, or when other modalities, like MRA cannot be used.

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Conflicts of interest

There are no conflicts of interest.

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