An unusual and rare form of truncus arteriosus in an asymptomatic woman

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Abstract

Persistent truncus arteriosus is a rare congenital cardiac malformation. It is characterized by a single great artery arising from the heart which supplies the aorta, the origin of coronary arteries and pulmonary arteries. Without surgery, prognosis is poor and 90% of these patients die before one year of age. We report a rare case of an asymptomatic 35-year-old woman with uncorrected persistent truncus arteriosus and hypoplastic right and left pulmonary arteries. Hypoplastic branch pulmonary arteries prevented the development of severe pulmonary arterial hypertension.

Keywords

Congenital heart defect, pulmonary artery, truncus arteriosus, truncus

Date received: 28 October 2016; accepted: 21 April 2017

Introduction

Persistent truncus arteriosus (PTA) is a rare congenital cardiac malformation, comprising 2–4% of all congenital heart defects.¹ It is characterized by a single great artery arising from the base of the heart which supplies the aorta, the origin of coronary arteries and pulmonary arteries.² This anomaly was first described by Maurice Lev and Otto Saphir in 1942. Since then, different classifications have been defined by other researchers.³ Without surgery, prognosis is poor and 90% of these patients die before one year of age. Survival after the first decade is even rarer.⁴ Patients who have not undergone cardiac surgery are prone to over circulation-induced pulmonary arterial hypertension, severe heart failure and early death.⁵ We report a rare case of an asymptomatic 35-year-old woman with uncorrected PTA who was referred by her primary care physician.

Case report

A 35-year-old woman came to our cardiology clinic as recommended by her primary care physician. She did not have any symptom related to the heart other than dyspnea on strenuous exercise. She had one prior uncomplicated vaginal delivery 12 years ago that a girl was born with cleft lip and palate without any cardiac anomaly. On physical exam, there was central

cyanosis and clubbed fingers. Furthermore, a continuous murmur was heard in the sternal borders. Her blood pressure was 100/60 mmHg and heart rate was 72 beats /minute. An electrocardiogram demonstrated sinus rhythm with a right bundle branch block. Trans thoracic echocardiography followed by trans esophageal echocardiography showed situs solitus, inferior vena cava continuity, D-loop, atrio ventricular concordance, significant right ventricular hypertrophy (the thickness of the right ventricular wall was 10 mm), flattening of the inter ventricular septum (D-shaped left ventricle) throughout the cardiac cycle and no significant left ventricular systolic impairment. Also, a non-restrictive sub arterial ventricular septal defect (VSD) and a single arterial trunk arising from the base of both ventricles with moderate to severe regurgitation of the tricuspid truncal valve were seen. (Figure 1). Truncal valve did not appear to be significantly dysplastic

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Figure 1. Transthoracic two-dimensional echocardiography and color Doppler flow velocity mapping in a parasternal long-axis view revealed a non-restrictive subarterial VSD (ventricular septal defect) and a single arterial trunk with moderate to severe regurgitation of truncal valve.

RVOT: Right ventricular outflow tract; LV: Left ventricle.

Figure 3. Transthoracic two-dimensional echocardiography in a five-chamber view demonstrated branch pulmonary arteries arising from postero-lateral wall of the truncus.

RV: Right ventricle; LA: Left atrium; RVOT: Right ventricular outflow tract; LV: Left ventricle, PA: Pulmonary artery.

Figure 2. Transthoracic two-dimensional echocardiography in an apical long-axis view revealed a non-restrictive subarterial VSD (ventricular septal defect) and a single arterial trunk that overrides both ventricles.

LV: Left ventricle; LA: Left atrium; RVOT: Right ventricular outflow tract.

Figure 4. Transthoracic two-dimensional echocardiography in a supra-sternal view showed a hypoplastic pulmonary arterial branch arising from arterial trunk.

PA: Pulmonary artery.

was 55 mmHg (Figure 8). The patient did not give consent for further workup or receive therapy.

Discussion

and it was only mildly thickened. Furthermore, the main pulmonary artery was absent and hypoplastic right and left pulmonary arteries had arisen from the posterolateral aspect of the truncus, with diameters of approximately 1 cm (Figures 2 to 6). Color-Doppler echocardiography showed continuous left-to-right shunt from flow through the truncus into pulmonary arterial branches (Figure 7). The peak velocity of the shunt flow was 3.72 m/s and the estimated pressure gradient

PTA is rare congenital heart anomaly. Anatomically, a single arterial trunk arises from the heart, supplies the coronary arteries, the pulmonary arteries, and the systemic arterial circulation. In the majority of instances, there is a subarterial VSD and a single semilunar valve. The semilunar valve usually overrides the true inter-ventricular septum, and thus both ventricles.⁶ The two main classification of PTA has been introduced: In the one that has been used by Collett-Edwards,

Figure 5. Trans esophageal echocardiography in mid-esophageal ascending aortic short-axis views (a, b) [0 degree] demonstrated pulmonary arterial branches arising from postero lateral wall of the truncus. PA: Pulmonary artery.

Figure 6. Trans esophageal echocardiography in mid-esophageal short-axis view [45 degree] demonstrated tricuspid truncal valve. Furthermore, a small and blind ending RVOT was seen anterior to truncal valve. LA: Left atrium, RA: Right atrium, TV: Tricuspid valve.

Figure 8. Trans esophageal echocardiography and Doppler flow velocity mapping in the mid-esophageal view demonstrated continuous turbulent flow from the arterial trunk toward pulmonary arterial branch.

Figure 7. Trans esophageal echocardiography and color Doppler flow velocity mapping in mid-esophageal long-axis view [130 degree] demonstrated continuous turbulent flow from the arterial trunk toward pulmonary arterial branch. PA: Pulmonary artery, LV: Left ventricle, RVOT: Right ventricular outflow tract.

there are four subtypes. In type I, a main pulmonary artery segment is present that is originated from the proximal left lateral wall of the main truncus. The branch pulmonary arteries are arising from main pulmonary artery segment. This subtype represents the most common form (48–68%). Type II is described by absence of the aortico pulmonary septum and the separate origin of the branch pulmonary arteries from the posterior aspect of the common trunk (29–48%). Type III is characterized with absence of the main pulmonary artery and the separate origin of branch pulmonary arteries from the lateral aspect of the common trunk (6–10%). Type IV is described with the origin of branch pulmonary arteries from descending aorta that recently considered as a variant of Tetralogy of Fallot with pulmonary atresia and a sub-aortic VSD (pseudo-truncus).⁷ Another classification has been described by Van Praagh and Van Praagh⁸: type 1 is similar to type I of Collett–Edwards. Type 2 is identical to Collett–Edwards types II and III. Type 3 includes cases with the origin of one branch pulmonary artery (usually the right) from the common trunk, with pulmonary blood

supply to the other lung provided by a pulmonary artery arising from the ductal or aortic arch origin. Type 4 is defined by the coexistence of an interrupted aortic arch in addition to types 1 and 2. The type B interruption, that occurs between the left common carotid and left subclavian arteries, is the most common form.⁸ This classification also identifies the presence of a VSD (type A), or the absence of a VSD (type B).⁶ Our patient belongs to Collett–Edwards type II and Van Praagh type 2A. This form is probably the most common form in unoperated patients who survive into adulthood because of high risk of developing Eisenmenger syndrome and death in Collett–Edwards type I (Van Praagh type 1). Echocardiogram is the best imaging modality for the diagnosis and surgical planning. The parasternal short axis view should be used for the evaluation of truncal valve morphology, while the apical and subcostal views can appropriately determine the truncal valve stenosis or regurgitation. Furthermore, the suprasternal view should be used for the diagnosis of an interrupted aortic arch.⁹ Survival into adulthood is rare without surgical intervention and nearly all of the survivors have severe vascular obstructive disease or Eisenmenger syndrome that is defined as pulmonary hypertension due to a high pulmonary vascular resistance ($>10 \text{ WU} \times \text{m}^2$) with right to left or bidirectional shunt at arterial, ventricular, or atrial level.⁹ The pulmonary arterial hypoplasia in these patients may induce protection of the pulmonary circulation and thus may be beneficial as seen in our patient.¹ Our patient had hypoplasia of both branch pulmonary arteries. Protection of her pulmonary circulation from developing Eisenmenger syndrome might be induced by pulmonary arterial hypoplasia. In our patient, the peak velocity of the shunt flow through the truncus into pulmonary arterial branches was 3.72 m/s and the estimated pressure gradient was 55 mmHg. However, a limitation of our study is that hypoplastic pulmonary arteries with long tubular stenosis can be associated with significant underestimation of pressure gradient due to viscous losses.¹⁰

Chromosome 22q11 deletion is present in 30% of cases of truncus arteriosus. On the other hand, DiGeorge syndrome and conotruncal anomaly face syndrome are associated with this chromosomal anomaly.¹¹ Tbx1 is a T-box transcription factor on chromosome 22q11 that seems to be associated with DiGeorge syndrome and conotruncal anomaly face syndrome. Furthermore, it is a principal factor in the development of heart tube, including the outflow tract and the pulmonary infundibulum.⁹ Our patient did not have any craniofacial anomaly. However, her daughter was born with cleft lip and palate. This chromosomal association is an explanation for occurrence of craniofacial anomaly in her daughter. Recent advances in pediatric

cardiac surgery have led to early complete repair in these patients. In older unoperated children, surgery may be complicated or contraindicated by the presence of severe pulmonary hypertension.¹

Declaration of Conflicting Interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

Ethical approval

Written consent was received from this patient to publish the study and images.

Guarantor

This study belongs to Cardiovascular Research Center of Mazandaran University of Medical Sciences.

Contributors

This study was prepared and written by corresponding author.

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