

Successful treatment of abdominal aorto-right atrial fistula by vascular plug: A previously unreported cardiac malformation

Fatoş Alkan, Yüksel Pabuşcu*, Mecnun Çetin¹, Şenol Coşkun
Division of Pediatrics Cardiology, Departments of Pediatrics and
*Radiology, Faculty of Medicine, Celal Bayar University,
Manisa-Turkey

¹Departments of Pediatrics, Division of Pediatric Cardiology, Faculty of Medicine, Yüzüncü Yıl University, Van-Turkey

Introduction

Aorto-cardiac fistulas are defined as a sizable communication between the aorta and cardiac spaces, including the right atrium (RA), right ventricle, left atrium, or left ventricle. Aorto-atrial fistula is a rare condition mostly observed as an acquired or extremely rare congenital condition. Causes of the acquired condition are mainly complications of previous cardiac surgery, aortic root abscess due to bacterial endocarditis, paravalvular abscess, ruptured sinus of Valsalva aneurysm, or aortic dissection (1). Congenital cases have poorly defined characteristics with uncertain pathogenesis. Classic signs (continuous murmur) are mostly absent in congenital cases (2).

Here we report a case of congenital aorto-right atrial fistula (ARF) with a longer fistula tract. Surgical management is the mostly commonly used treatment option in these cases. In two previously reported cases of ARF originating from the ascending aorta, Amplatzer duct occluder (ADO) I and ADO II were used



Figure 1. Transthoracic echocardiogram, subcostal inferior vena cava view showing the aorto-right atrial fistula. ARF-aorto-right atrial fistula, IVC-inferior vena cava, RA-right atrium

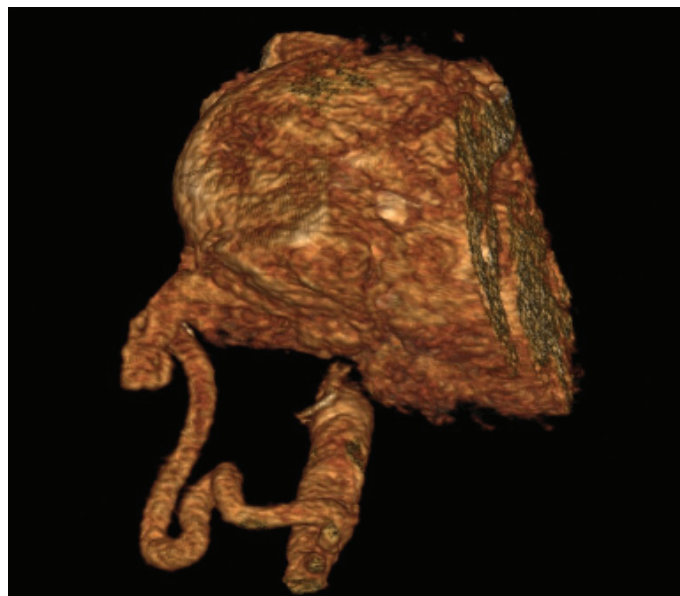


Figure 2. Computed tomography angiography showing a tortuous, large, and very long fistula between the abdominal aorta and right atrium

(3, 4). Our literature review suggested that only two reports on cases of a fistula to the RA originating from the descending thoracic aorta have been published. Surgical and Amplatzer vascular plug II were applied in these cases (5, 6).

Here we describe, for the first time, a descending abdominal aorta to the right atrial fistula, which was successfully closed by trans-catheter embolization using an Amplatzer vascular plug IV.

Case Report

A 2-year-old asymptomatic boy was referred to our clinic because of a continuous murmur that was more evident at the left upper sternal border. No other pathological finding was detected in his physical and chest X-ray examinations. During echocardiographic examination, a small shunt from the proximal descending aorta to the main pulmonary artery was observed and diagnosed as patent ductus arteriosus (PDA). We noted an abnormal, continuous, high-velocity flow into the RA, which was dilated, with a gradient of 75 mm Hg. The left and right ventricles were slightly dilated without any functional loss. An extra-cardiac structure was found to be open to the RA on the posterior lateral side of the inferior vena cava (Fig. 1). Computed tomography (CT) angiography confirmed a tortuous, large, and very long fistula (Fig. 2). No connection was observed between this fistula and the liver or the portal system on both CT and abdominal ultrasonography. This ruled out the possibility of hepatic arteriovenous fistula and hepatoportal fistula. Right and left heart catheterization was performed, which revealed a Qp/Qs ratio of 1.8: 1. Descending aorta angiography confirmed the diagnosis of a very small PDA and unusual tortuous fistula between the descending abdominal aorta and the RA (Fig. 3). A catheter was



Figure 3. Anterior–posterior angiograms of the abdominal aorta showing a tortuous fistulous tract between the abdominal aorta and right atrium (arrow)

advanced to the fistula orifice directly from the abdominal aorta. The fistula was selectively engaged using a 5-Fr right Judkins catheter. A hydrophilic guidewire was then passed through the proximal fistula into the narrowing proximal portion of the fistula. This area measured approximately 5 mm. A 7-mm Amplatzer vascular plug IV device was loaded into the catheter, and the distal skirt of the device was placed on the narrow and curved part, 5-6 mm away from the proximal fistula. After device replacement, significant residual flow was observed, and thereafter, a second 8-mm Amplatzer vascular plug IV device was advanced close the residual leak (Fig. 4a). A non-selective contrast agent was injected into the abdominal aorta, which revealed occlusion of

the fistulous tract with the Amplatzer vascular plug IV device in stable position at the proximal mouth of the fistula (Fig. 4b). PDA could not be closed as it could not pass through the catheter. No procedure-related complication occurred. The patient was discharged from the hospital on the second day with acetylsalicylic acid (5 mg/kg/day), and he remained well during the 6 months of follow-up.

Discussion

ARF is an uncommon condition with unclear pathogenesis. Possible embryological abnormalities in the sixth aortic arch (arterial component) or sinus venous or cardinal veins (venous component) were accused for the fistula formation (6). Mostly, it was thought that congenital deficiency of the elastic lamina in the aortic wall (cystic medial necrosis) leads to this type of fistula (7). Most ARFs occur due to complications of surgical procedures, bacterial endocarditis, paravalvular abscess, ruptured sinus of Valsalva, and aortic dissection, and few of them are congenital (8, 9).

Our patient had no history of any surgical procedure to suggest an acquired origin. Presentation may be asymptomatic but once detected, closure is recommended to prevent potential complications. If complications, such as congestive cardiac failure, bacterial endocarditis, aneurysm formation, or spontaneous rupture, occur, they can be fatal.

Conclusion

Various occlusion devices and techniques are available for treating ARF. The percutaneous closure of ARF with an Amplatzer vascular plug type device IV can be considered as a therapeutic option in cases with favorable anatomy.

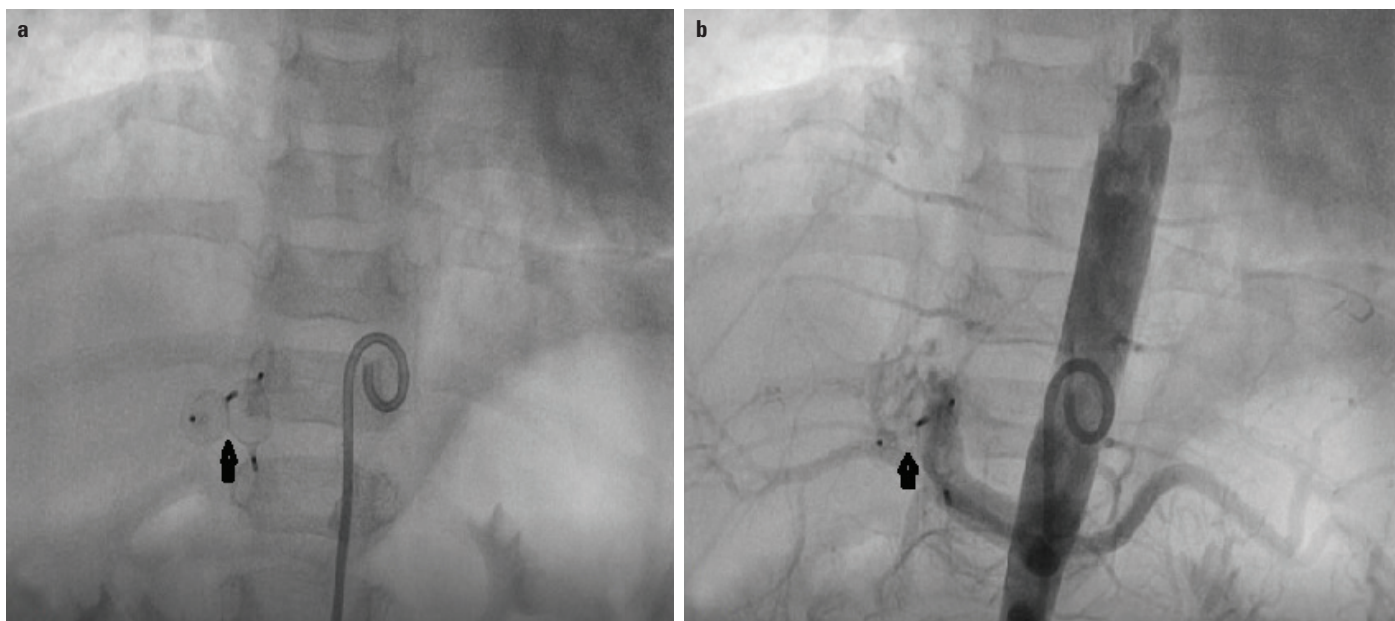


Figure 4. (a) Devices were deployed to the narrow and curved part at a distance of approximately 5-6 mm from the proximal fistula; (b) no residual shunt was observed after detachment. Arrow indicates the position of the Amplatzer vascular plug IV

References

1. Darwazah A, Kiswani M, Ismail H, Hawari M, Awad S. Aorto-right atrial fistula: a complication of prosthetic aortic valve endocarditis. A case report. *J Heart Valve Dis* 2006; 15: 142-5.
2. Shah BN, Livesey SA, Rakhit DJ. Aorto-atrial fistula in the absence of infective endocarditis diagnosis by 2- and 3-Dimensional transesophageal echocardiography. *Tex Heart Inst J* 2012; 39: 146-7.
3. Chandra S, Vijay S, Kaur D, Dwivedi S. Congenital aorta right atrial fistula: successful transcatheter closure with the Amplatzer occluder. *Pediatr Cardiol* 2011; 32: 1057-9. [CrossRef]
4. Ece I, Üner A, Cüce F, Şahin M. Transcatheter closure of tortuous aorto-right atrial fistula. *Cardiovasc Interv Ther* 2015;30:151-4.
5. Elwatidy AF, Galal AN, Rhydderch D, Ashmeg AK. Aorto-right atrial fistula. *Ann Thorac Surg* 2003; 76: 929-31. [CrossRef]
6. Ho AB, Magee AG, Hayes N. Descending thoracic aorta to right atrial fistula: Presenting with neonatal collapse. *Catheter Cardiovasc Interv* 2017 May 30. Epub ahead of print. [CrossRef]
7. Gajjar T, Voleti C, Matta R, Iyer R, Dash PK, Desai N. Aorta-right atrial tunnel: clinical presentation, diagnostic criteria, and surgical options. *J Thorac Cardiovasc Surg* 2005; 130: 1287-92. [CrossRef]
8. Arnett EN, Roberts WC. Valve ring abscess in active infective endocarditis; frequency, location and clues to clinical diagnosis from study of 95 necropsy patients. *Circulation* 1976; 54: 140-5. [CrossRef]
9. Sakakibara S, Konno S. Congenital aneurysms of sinus of Valsalva. A clinical study. *Am Heart J* 1962; 63: 708-19. [CrossRef]

Address for Correspondence: Dr. Fatoş Alkan
Celal Bayar Üniversitesi, Tıp Fakültesi,
Pediyatrik Kardiyoloji Bölümü
45030, Manisa-Türkiye
Phone: +90 236 444 42 28 Fax: +90 236 233 80 40
E-mail: fatos.alkan@hotmail.com

©Copyright 2017 by Turkish Society of Cardiology - Available online
at www.anatoljcardiol.com
DOI:10.14744/AnatolJCardiol.2017.7973



A novel mutation in the desmoplakin gene in two female siblings with a rare form of dilated cardiomyopathy: Carvajal syndrome

Mehmet G. Ramoğlu, Tayfun Uçar, Serdar Ceylaner¹, Semra Atalay, Ercan Tutar

Department of Pediatric Cardiology, Faculty of Medicine, Ankara University, Ankara-Turkey

¹Department of Medical Genetics, Intergen Genetic Centre, Ankara-Turkey

Introduction

Carvajal syndrome is a cardiocutaneous syndrome characterized by dilated cardiomyopathy (DCM), woolly hair, and keratoderma (1). Here we present the case of two female siblings with Carvajal syndrome and a new homozygous frameshift mutation in desmoplakin (DSP).

Case Report

A 5-year-old female patient, who was the first child of second-degree consanguineous parents, with no significant medical history was admitted with complaints of malaise and abdominal pain that persisted for 3 months. She had tachypnea and tachycardia. Her vital signs were as follows: heart rate, 140 beats/min; respiratory rate, 32 breaths/min; and blood pressure, 95/64. Gallop rhythm and jugular venous distension were noted. The liver and spleen were palpable 10 and 5 cm below the costal margin, respectively, and edema was present on the legs. Laboratory findings were as follows: brain natriuretic peptide, 2667 pg/mL (normal <100); creatine kinase-MB, 8.1 ng/mL (normal <6.3); and cardiac troponin I, 0.03 ng/mL (normal <0.06); complete blood count and other biochemical laboratory findings were normal except for mildly elevated liver enzyme levels. Echocardiography revealed markedly dilated cardiac chambers, prominently the left chambers, marked left ventricular dysfunction (ejection fraction: 25.7%, fractional shortening: 13%, LVIDd: 47 mm) and moderate mitral regurgitation. After being treated for congestive heart failure (CHF) for 2 years, left ventricular assist device was implanted, and on the 501th day after implantation, the patient underwent heart transplantation. The biopsy of the heart revealed widespread multifocal myofibrillar damage and interstitial fibrosis. The patient's 18-month-old younger sibling, who was successfully treated for neuroblastoma at the age of 1 year, was treated for DCM for 1 year and was referred to our hospital at the age of 6 years. Besides signs of CHF, peculiar woolly hair (Fig. 1a), palmoplantar (1b & 1c) keratoderma, and wart-like lesions on the hand were strikingly forefront in both siblings. Both patients had normal eyelash, eyebrow, and nail and teeth development. At admission, both siblings had ventricular arrhythmias, voltage suppression, and left-sided cardiomyopathy. All screening test results for DCM (metabolic screening tests, viral serologic tests, and upper respiratory viral and bacterial panel) were normal. Genetic screening revealed a normal JUP gene and a new homozygous frameshift mutation, c.4650_4651delTG (p. V155Efs*75), in DSP in both siblings. Both parents were also heterozygous for the DSP frameshift mutation. The parents did not have any abnormal echocardiographic, electrocardiographic, or cutaneous findings. The younger sibling has been on anti-congestive medication for 4 years and the older sibling had a successful heart transplantation 23 months ago.

Discussion

DCM can be caused by a variety of factors or may be inherited as a hereditary disease. Although most commonly cytoskeletal, sarcomere, or Z-disk proteins are affected, mutations in ion channels and desmosome-encoding genes have also been reported (2).

Desmosomes are major cell adhesion junctions that provide mechanical stability, and desmoplakin is the most abundant constituent (3). Dysfunction of desmosomes leads to cell death and