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MINI-FOCUS ISSUE: CONGENITAL HEART DISEASE

INTERMEDIATE

CASE REPORT: CLINICAL CASE

Aortoventricular Tunnel With Severely Dilated Ascending Aorta and Bicuspid Aortic Valve in a Newborn

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ABSTRACT

Aortoventricular tunnel is a rare congenital cardiovascular malformation whereby there is a paravalvular communication between the aorta and a ventricle. This unique case describes a newborn with an aortoventricular tunnel, a severely dilated ascending aorta, and a bicuspid aortic valve, which was suspected prenatally and surgically managed postnatally. (**Level of Difficulty: Intermediate.**) (J Am Coll Cardiol Case Rep 2020;2:734–9) © 2020 The Authors. Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

HISTORY OF PRESENTATION

A male infant, born at 38 weeks to a gravida 2 para 1 to 2 mother via induced vaginal delivery because of maternal hypertension, with APGAR scores of 9 and 10 at 1 and 5 min, respectively, a birth weight of 4.1 kg, and length of 21 inches, was prenatally diagnosed by fetal echocardiogram with a dysplastic bicuspid aortic valve, a severely dilated ascending aorta, and a mildly dilated and hypertrophied left ventricle (LV) with normal systolic function.

LEARNING OBJECTIVES

- Although rare, AVT is a serious cause of abnormal blood flow from the aorta to the ventricle in infancy.
- Echocardiography is the diagnostic investigation of choice and must be used to distinguish AVT from other lesions that cause rapid runoff of blood from the aorta.
- Optimal management of patients with AVT includes prompt surgical repair to prevent progression into heart failure.

DIFFERENTIAL DIAGNOSIS

Based on this prenatal echocardiogram (**Figure 1**), there was suspicion for a possible right sinus of Valsalva aneurysm into the LV versus an aortoventricular tunnel (AVT). Other less likely considerations in the differential diagnosis included coronary arteriovenous fistula, congenital isolated aortic incompetence, and aortic incompetence with ventricular septal defect.

Prenatal counseling allowed the parents to be informed about the need for delivery in a tertiary center with availability of the neonatal intensive care unit and pediatric cardiology service. Following birth, the infant did well clinically and maintained adequate oxygen saturation, blood pressure, and peripheral perfusion. On physical examination, he had a grade II/VI systolic ejection murmur over all of the precordium with no diastolic murmurs. The infant

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The authors attest they are in compliance with human studies committees and animal welfare regulations of the authors' institutions and Food and Drug Administration guidelines, including patient consent where appropriate. For more information, visit the *JACC: Case Reports* author instructions page.

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remained in the nursery and did not require transfer to the neonatal intensive care unit.

INVESTIGATIONS

A postnatal echocardiogram was performed a few hours after delivery and demonstrated a patent foramen ovale, bicuspid aortic valve with mild flow acceleration, mild aortic regurgitation, dilated aortic root and ascending aorta, and continued evidence of flow across the suspected ruptured sinus of Valsalva or AVT (**Figures 2 to 4**, Videos 1, 2, 3, 4, and 5). Mild left ventricular hypertrophy and moderate to severe dilation of the LV was also noted to be compressing the right ventricle. Another limited echocardiogram was performed 24 h later with similar findings. He was discharged from the nursery at 48 h after birth. His next follow-up at 2 weeks of age was in the outpatient setting, and he had remained asymptomatic and had regained birth weight.

He underwent a cardiac computed tomography angiogram, which reported a structure arising from the right coronary cusp just above the origin of the right coronary artery with a contained perforation into the interventricular septum. This communicated with the left ventricular outflow tract below the level of the aortic valve. A dilated ascending aorta was seen as well (**Figure 5**). He also underwent genetic evaluation and was found to have a latent transforming growth factor-beta binding protein (LTBP4) heterozygous mutation, a finding not sufficient to establish a genetic diagnosis.

MANAGEMENT

At 7 weeks of life, elective surgical repair was undergone. The delineated AVT was repaired with a sandwich technique using autologous pericardial patches on both the aortic entry side and LV exit site. The patent foramen ovale was also closed.

FOLLOW-UP

On his 2-week post-operative follow-up, he was overall thriving without any cardiac symptoms and had a reassuring physical examination. His echocardiogram revealed a residual shunt across his left ventricular patch end, a small atrial shunt, a severely dilated ascending aorta, a dysplastic aortic valve with mild aortic valve regurgitation, and a hypertrophied LV with borderline normal systolic function (**Figures 6** and **7**). He was followed serially, and at 3 months of age he had normalization of his left ventricular size and systolic function. At 14 months of age, he continued to be doing well clinically with improvement of dilation in ascending aorta (decreasing *z*-score toward normal range). There was no evidence of aortic stenosis nor worsening of mild aortic regurgitation.

DISCUSSION

AVT is a paravalvular connection between the aorta above the sinotubular junction and the ventricle. It was first reported in 1963 by Levy et al. (1) and continues to be a rare defect that only accounts for 0.1% to 0.5% of all congenital cardiac defects. Although the etiology remains uncertain, it has been suggested to result from a combination of maldevelopment of the endocardial cushions, which give rise to the pulmonary and aortic roots, and abnormal separation of these structures (2). Many times, AVTs are associated with other structural abnormalities. Associated lesions of the aortic valve occur in about 20% of patients, ranging from 2-leaflet valves without obstruction to severe dysplasia or atresia.

Hovaguimian et al. (3) proposed a classification of 4 anatomic types of AVTs and suggested that repair should be individualized according to the anatomic type. They described a slit-like opening at the aortic end with no valve distortion in 24% of the cases (type 1), a large extracardiac aneurysm in 44% (type 2), an intracardiac aneurysm of the septal portion of the tunnel with or without right ventricular outflow tract obstruction in 24% (type 3), and a combination of

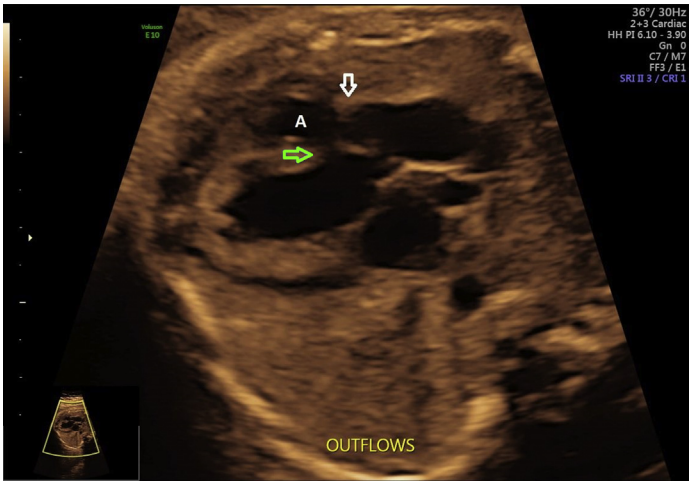
ABBREVIATIONS AND ACRONYMS

AVT = aortoventricular tunnel

LTBP4 = latent transforming growth factor-beta binding protein

LV = left ventricle

FIGURE 1 Prenatal Fetal Echocardiogram



White arrow points toward right aortic cusp. **Green arrow** points toward opening of the tunnel into the left ventricle. Structure marked **A** depicts the aortoventricular tunnel.

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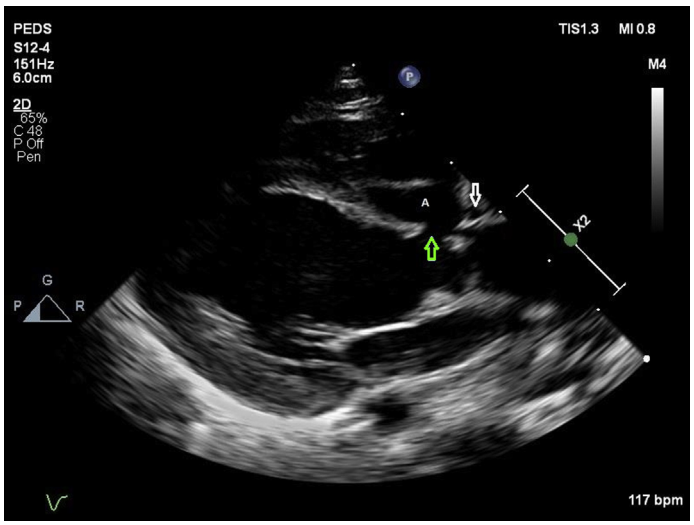
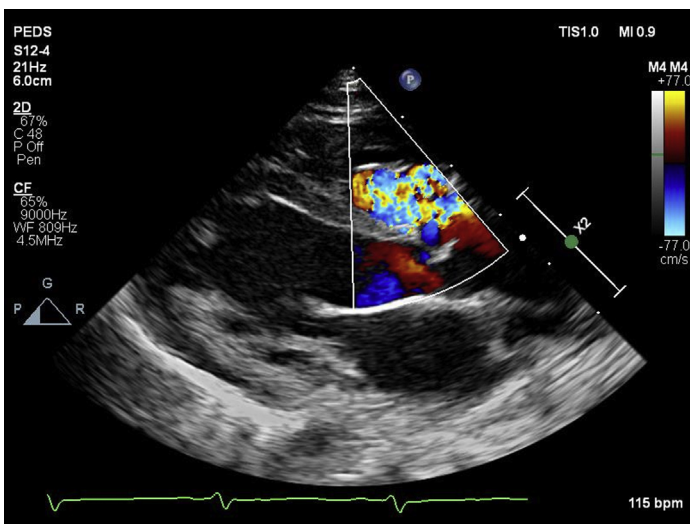
FIGURE 2 Postnatal Echocardiogram: Parasternal Long View



White arrow points toward right aortic cusp. **Green arrow** points toward opening of the tunnel into the left ventricle. Structure marked **A** depicts the aortoventricular tunnel.

types 2 and 3 in 8% (type 4). The differences between the 4 types most likely represent different degrees and stages of the same disease process and have a strong bearing on the mortality, morbidity, repair techniques, and outcome of the surgical treatment

FIGURE 3 Postnatal Echocardiogram: Parasternal Long View With Color Doppler



Doppler portraying shunting across opening of the tunnel into the left ventricle and color aliasing in the aortoventricular tunnel.

(3). In this case, the patient's AVT was more consistent with the more advanced, type 3 to 4 variant.

Echocardiography is the diagnostic investigation of choice. Transthoracic cross-sectional imaging in a parasternal long-axis view allows for viewing of the tunnel itself, and its aortic origin and left ventricular opening. On color Doppler studies, diastolic flow can be seen passing from the aorta to the LV and systolic flow from the ventricle to the aorta. In utero diagnosis of AVT is made possible by the finding of fetal left ventricular enlargement and hypertrophy, aortic root enlargement, and aortic regurgitation on routine obstetric ultrasound screening as early as the 18th to 20th gestational week (4).

AVTs must be distinguished from other lesions that cause rapid runoff of blood from the aorta and produce cardiac failure. An AVT differs from a ruptured sinus of Valsalva aneurysm in that an AVT orifice is located in the tubular ascending aorta rather than in a sinus of the aortic valve. In addition, an AVT passes outside of the heart into the tissue plane between the muscular subpulmonary infundibulum and the aortic valvular sinuses. Echocardiography may be misleading and distinguishing between the two is difficult when there is an aneurysm at the level of the right sinus of Valsalva with aortic incompetence. This is because of prolapse of the right aortic cusp occluding the ventricular orifice of the tunnel during diastole. It is important to note that the ostium of a coronary artery may lie within an AVT and absence of the origin of the left or right have both been observed with this anomaly. It is, therefore, of utmost importance to visualize the ostium of the coronary arteries before any intervention.

Although this patient was found to have a mutation in LTBP4 (expressed in cardiac, skeletal muscle, and lung tissue), a single variant in the LTBP4 gene is not sufficient to establish a genetic diagnosis. There are currently no known molecular markers for AVT, and it is not associated with any recognized genetic syndrome. Recent findings of cystic medial necrosis within the wall of an ascending aortic aneurysm resected 15 years after repair of an AVT raises the possibility that markers of an associated or underlying connective tissue disorder may emerge in the future.

Optimal management of symptomatic AVT consists of diagnosis by echocardiography complimented with cardiac catheterization, as needed to elucidate coronary arterial origins or associated defects, followed by prompt surgical repair. The goal of any treatment modality is to obliterate the tunnel. The techniques that have been described in previous reports are summarized as follows: 1) closure of the aortic orifice of the tunnel with or without a patch (Dacron,

pericardium Teflon); 2) closure of the ventricular end of the tunnel; 3) obliteration of the tunnel (i.e., ligation of the tunnel, or partial resection of the tunnel, or filling of the tunnel with gel-foam); or 4) closure of both orifices (aortic and ventricular [i.e., sandwich technique]) (1,3,5). Ideally, both ends of the tunnel should be closed, providing support to the right aortic leaflet. This is critical to the preservation of the aortic valve competency and avoidance of subpulmonary obstruction (4,6).

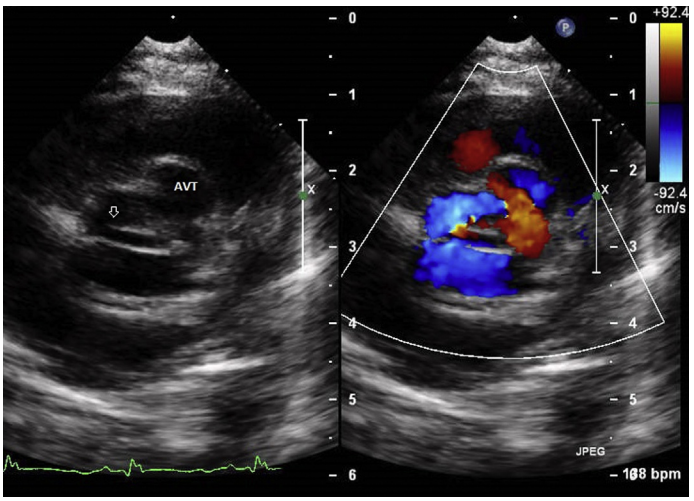
The closure of the aortic orifice requires surgical opening of the ascending aorta. The same approach can be used for closure of the LV outlet through the aortic valve. Risks of this approach include potential aortic valve injury. Serino et al. (7) support closing the aortic defect by direct suture. However, this technique could distort the cusps by pulling them toward the weak aortic wall, which remains unsupported within the dilated aortic sinus. Consequently, the aortic regurgitation may persist and progress even if repaired in infancy. The surgical patch technique is believed to reduce this risk. Using a direct approach, advancing through the tunnel and closing both ostia with pericardial patches avoids distortion of the aortic valve while supporting the aortic wall.

Only patients having undergone repair in the first 6 months of life have been shown to have subsequent normalization of left ventricular size and function. Observation of the exceedingly rare, asymptomatic patient with a small tunnel may be justified by occasional spontaneous closure seen in a single documented case (7). However, most patients develop symptoms of heart failure during the first year of life. The onset, severity, and progression of heart failure is, however, variable, and ranges from many years of asymptomatic compensation to rapid decompensation, sudden death, or death in utero. Therefore, prompt surgical repair is advised at time of diagnosis. Martins et al. (5) illustrated that most patients were asymptomatic at post-surgical follow-up (median, 5 years; 1 month to 35 years), and a minority had residual AVT with, at most, mild aortic regurgitation. All patients require lifelong follow-up for recurrence of the tunnel, aortic valve incompetence, left ventricular function, and aneurysmal enlargement of the ascending aorta as well as risk of endocarditis (8).

CONCLUSIONS

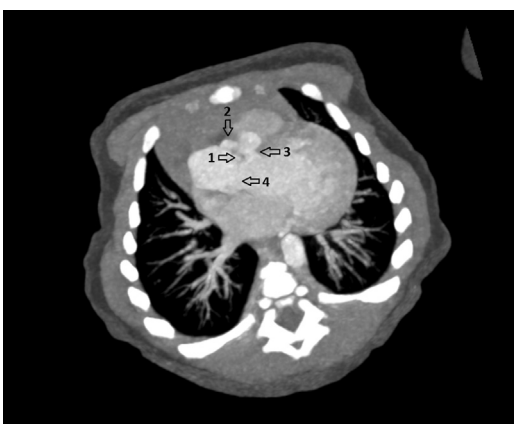
AVT is a rare cardiac malformation that may cause progressive postnatal heart failure and is often associated with other cardiac lesions. In utero diagnosis currently improves the neonatal management of AVT through assessment of its prognosis and programmed assisted delivery. High suspicion is

FIGURE 4 Postnatal Echocardiogram: Parasternal Short View With Color Compare



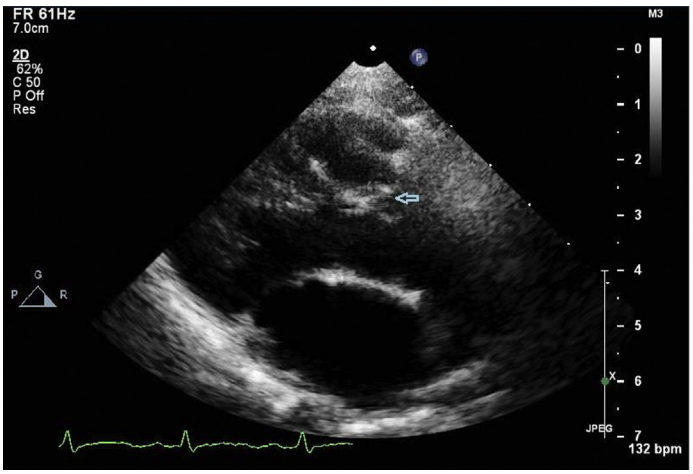
(Left) Aortoventricular tunnel. **Arrow** points toward dysplastic aortic valve. **(Right)** Color Doppler depicting the flow across the aortic end of the tunnel. AVT = aortoventricular tunnel.

FIGURE 5 Heart Computed Tomography Angiogram



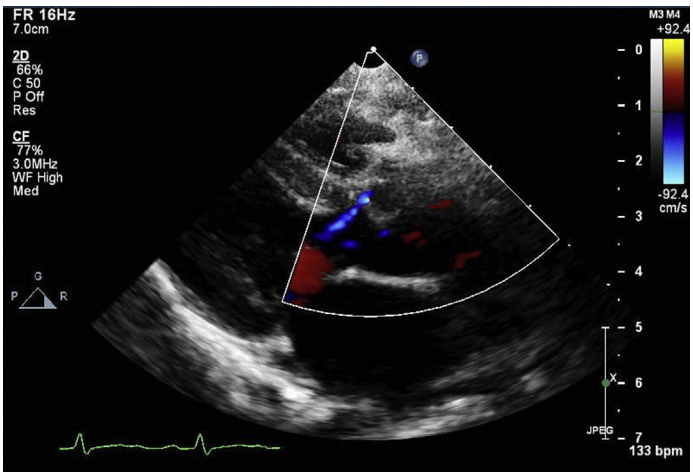
Arrows pointing toward right coronary cusp and ostium **(1)**, aortic end of the left aortoventricular tunnel **(2)**, ventricular end of the left aortoventricular tunnel **(3)**, and aortic valve **(4)**.

FIGURE 6 **2 Weeks Post-Operative Echocardiogram: Parasternal Long View**



Arrow points toward surgical patch repair of aortoventricular tunnel.

FIGURE 7 **Post-Operative Echocardiogram: Parasternal Long View With Color Doppler**



Indicates residual shunt across the left ventricular patch end **(blue jet)**, and mild aortic valve regurgitation **(red jet)**.

warranted when a prenatal fetal echocardiogram indicates aortic regurgitation and left ventricular dysfunction (9). Patch closure is the surgical procedure of choice for repair and has good long-term outcomes (10).

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KEY WORDS aortoventricular tunnel, bicuspid aortic valve, dilated ascending aorta, fetal echocardiography

APPENDIX For supplemental videos, please see the online version of this paper.