Unrepaired Transitional Atrioventricular Septal Defect in a 52-Year-Old Patient

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INTRODUCTION

Atrioventricular septal defects (AVSDs), also called atrioventricular (AV) canal defects, represent 4% to 5% of congenital heart diseases.[1] They encompass a spectrum of defects involving AV valves and both atrial and ventricular septa.[2] In 2000, there was an agreement on the classification and nomenclature based on the anatomy and severity, and the defects were categorized as partial, transitional, intermediate, or complete defects.[3,4] The majority of patients with AVSD are diagnosed early in life, with previously reported cases of milder defects diagnosed in the fourth and fifth decades of life.[4] A late first presentation of milder degrees of AVSD is not uncommon.[5]

While surgical repair remains the treatment option of choice with good long-term outcomes, there are no well-studied nonsurgical or transcatheter treatment options.[6,7] However, we present this case of unrepaired grown-up transitional AVSD to emphasize the pivotal role of advanced cardiac imaging in adult congenital heart disease diagnosis and management and to shed light on nonsurgical options for patients who have social and financial hardships.

CASE PRESENTATION

A 52-year-old man presented to our emergency department complaining of exertional dyspnea with minimal activity for 10 days, bilateral lower extremities edema, orthopnea, nonproductive cough, and palpitation. The patient was incidentally diagnosed with atrial septal defect (ASD) at the age of 4 but had no interventions at that time. The patient was asymptomatic until age 50 when they were admitted to an outside facility because of dyspnea and volume overload. An ASD with a left-to-right shunt and a coexisting anterior left AV valve (LAVV) leaflet cleft with a moderate regurgitation were visualized in the transesophageal echocardiogram (TEE), resulting in a diagnosis of a partial AVSD. After that first encounter, they had multiple subsequent hospitalizations due to hypoxemic respiratory failure and fluid overload. The patient was offered surgical intervention repeatedly but declined due to social and financial hardships. The other relevant cardiovascular risk factors were morbid obesity, insulin-dependent diabetes mellitus type 2, hypertension, heart failure with preserved ejection fraction, mild pulmonary hypertension, hyperlipidemia, and a history of tobacco smoking. The patient took atorvastatin, aspirin, furosemide, insulin, lisinopril, and metoprolol succinate at home.

Vital signs on presentation were blood pressure, 159/99 mm Hg; heart rate, 106 bpm; respiratory rate, 22; O_2 saturation (SPO_2) 95% on nasal cannula oxygen, 3L/min; and temperature 36.7 °C. On physical examination, the patient had 2/6 systolic murmur on the left sternal border, bibasilar crackles, and +3 bilateral lower extremities edema. Chest x-ray demonstrated cardiomegaly, bilateral pulmonary edema, and a small right pleural effusion (Figure 1). Electrocardiography showed sinus tachycardia with left-axis deviation and intraventricular conduction delay. Hs-troponin was normal, and brain natriuretic peptide was 186 pg/mL (normal, 0-100). The patient was admitted for acute exacerbation of chronic congestive heart failure and treated with intravenous furosemide 40 mg every 12 hours, lisinopril 40 mg daily, and supplemental O_2.

During hospitalization, extensive workup was undertaken that included transthoracic echocardiography, TEE, cardiac computed tomography scan, right and left heart catheterization with coronary angiography, and cardiovascular magnetic resonance imaging (CMR; Table 1). The left ventricular (LV) ejection fraction and volumes were within normal ranges, whereas the right atrium and ventricle were dilated. The Qp/Qs ratio was invasively calculated at 2.1/1 at arterial O_2 saturation of 94%. The patient was diagnosed with a

VIDEO HIGHLIGHTS

Video 1: Two-dimensional TEE with color Doppler, mid-esophageal image, apical 4-chamber view (0°) demonstrates the primum ASD with a bilateral (predominantly left-to-right) shunt, inlet VSD, and left and right AV valve regurgitations. *LA*, Left atrium; *LV*, left ventricle; *RA*, right atrium; *RV*, right ventricle.

Video 2: CMR SSFP images, 4-chamber view, demonstrate an ASD, right AV valve regurgitation (*yellow arrow*), and dilated RA and RV. A small VSD with left-to-right shunting is also seen (*white arrow*). *LA*, Left atrium; *LV*, left ventricle; *RA*, right atrium; *RV*, right ventricle.

Video 3: CMR SSFP images, basal short-axis view, demonstrate a cleft anterior leaflet of the LAVV at the A2 position (*white arrow*), a trileaflet RAVV, and a bridging band extending between the septal leaflet of the RAVV and the anterior leaflet of the LAVV (*yellow arrow*). *PA*, Pulmonary artery.

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Table 1 Clinical and multimodality investigational characteristics

Clinical characteristics	Value
Body mass index, kg/m^2	43
Body surface area, m^2	2.6
New York Heart Association class	III
Electrocardiogram:	
PR interval, ms	198
QRS, ms	121
QTc, ms	439
Heart catheterization:	
Right atrial pressure, mm Hg	11
Right ventricular pressure (systolic/diastolic), mm Hg	60/11
Pulmonary artery pressure (systolic/diastolic/mean), mm Hg	60/20/35
Pulmonary artery wedge pressure, mm Hg	20
Superior vena cava O_2 saturation, %	63
Inferior vena cava O_2 saturation, %	74
Right atrium O_2 saturation (high-low), %	74-66
Right ventricle O_2 saturation, %	81
Main pulmonary artery O_2 saturation, %	86
Pulmonary vascular resistance, Wood unit	2.3
Qp/Qs	2.1
Coronary angiography	Normal, right dominant
Cardiac computed tomography:	
Situs	Solitus
Coronary arteries origin	Normal
Pulmonary veins connection	Left atrium
Systemic venous return	Right atrium
ASD diameter, cm	1.4 × 2.2
CMR:	
LV ejection fraction, %	52
LV end-diastolic volume indexed, mL/m^2	57
LV end-systolic volume indexed, mL/m^2	27
LV mass indexed, gm/m^2	52
Right ventricular ejection fraction, %	42
Right ventricular end-diastolic volume indexed, mL/m^2	125
Right ventricular end-systolic volume indexed, mL/m^2	72
Right ventricular outflow tract diameter, cm	2.9
Left atrial volume indexed, mL/m^2	41
Right atrial volume indexed, mL/m^2	67
Aortic root diameter, cm	3.2
Aortic arch	Left arch, normal anatomy
Aortic flow (Qs), mL/min	79
Pulmonary flow (Qp), mL/min	185
Qp/Qs	2.3
LV late gadolinium enhancement	No

transitional AVSD composed of a primum ASD with a bidirectional (predominantly left-to-right) shunt (Video 1), a restrictive inlet ventricular septal defect (VSD; Figure 2) with a small residual left-to-right shunt, which was only visualized in the CMR study (Video 2), and 2 separate AV valves. There was a cleft at the A_2 position of the anterior leaflet of the LAVV (Video 3) with moderate eccentric regurgitation. The right AV valve (RAVV) had severe regurgitation due to malcoaptation of the leaflets. The septal leaflet's chordae were attached to the crest of the scooped AV septum (Figure 2), forming an aneurysmal membrane. Cardiac catheterization showed mildly elevated pulmonary vascular resistance, nonobstructed coronary arteries, and a gooseneck deformity of the LV outflow tract (Figure 3). A pulmonary function study showed mild restrictive lung disease.

The patient demonstrated clinical improvement with medical treatment and inpatient cardiac rehabilitation. They were discharged after a hospital stay of 10 days and referred to outpatient cardiac rehabilitation. The patient continued to follow up at our congestive heart failure and cardiac surgery clinics and was offered a referral to a center specializing in congenital heart surgeries but declined again for the previously mentioned reasons.

DISCUSSION

AVSDs are caused by abnormal development of the endocardial cushions during intrauterine development, which results in a central defect in the heart.[8] In the mild form (partial), there are typically a combined septum primum ASD and anterior leaflet cleft of the LAVV. Whereas transitional AVSD is considered a subtype of the partial form, it has a restrictive inlet VSD that is closed by the chordal attachment of the RAVV. Both partial and transitional AVSDs have a single AV ring but 2 orifices. Intermediate AVSD is differentiated from transitional by having a wider nonrestrictive inlet VSD. The complete form is characterized by the existence of 1 AV ring and a single wide orifice. All AVSDs share common features, which should get the reader's attention in regular cardiac imaging, including (1) loss of apical displacement of the tricuspid valve, placing it at the same level as the mitral valve; (2) the papillary muscles appear closer to each other due to counterclockwise rotation of the position of the inferomedial papillary muscle (Figure 4); (3) elongation of the distance from the LV apex to the aortic valve because of unwedging and anterior displacement of the LV outflow tract; and (4) absence of the AV septum. The LAVV anterior leaflet cleft is typically directed to the center of the ventricular septum (Video 3) and is different from the isolated mitral valve anterior leaflet cleft, which is directed anteriorly toward the aortic valve. Cardiac imaging plays an important role in every stage of care for these patients. It is mandatory to establish the diagnosis and differentiate AVSD from simpler lesions like isolated

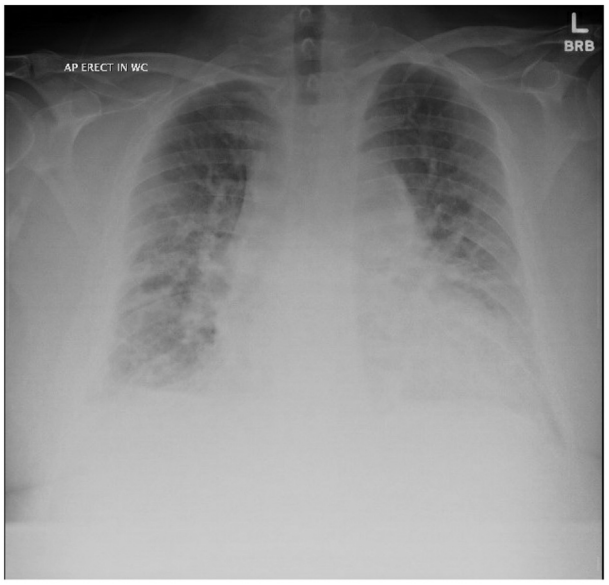

Figure 1 Chest x-ray. Anteroposterior view shows cardiomegaly and bilateral pulmonary congestion.

ASD or anterior mitral leaflet cleft. AVSDs may have associations with other congenital heart diseases, including but not limited to LV outflow tract obstruction, tetralogy of Fallot, heterotaxy, aortic coarctation, and anomalous pulmonary venous connection. Therefore, thorough multimodality cardiac imaging to define the hemodynamics and concomitant pathologies is crucial. Patients will need regular follow-up imaging even after having the definitive intervention as it is common to have postsurgical complications such as residual defects, LAVV and/or RAVV regurgitation, and subaortic stenosis.[9]

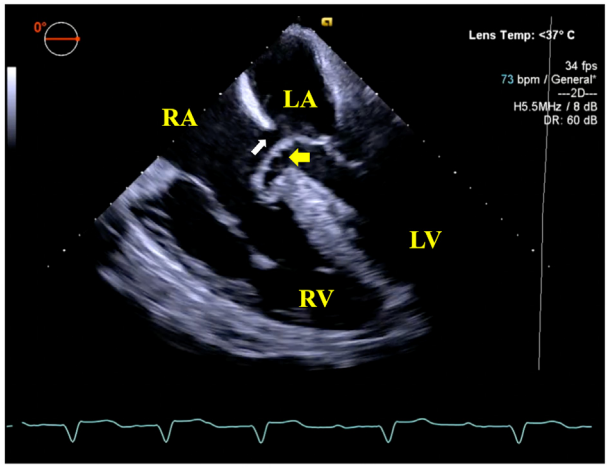

Figure 2 Two-dimensional transesophageal echocardiography, midesophageal image, apical 4-chamber view (0°), demonstrates a transitional AVSD composed of loss of normal apical displacement of the right AV valve, a primum ASD (*white arrow*), and a restrictive inlet VSD (*yellow arrow*) covered by an aneurysmal membrane formed by the right AV valve chordae attached to the scooped interventricular septum. *LA*, Left atrium; *LV*, left ventricle; *RA*, right atrium; *RV*, right ventricle.

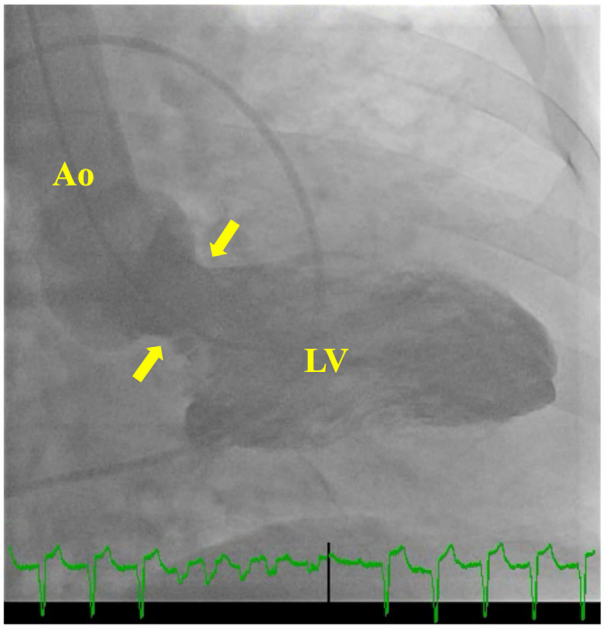

Figure 3 Catheter-guided left ventriculography. Right anterior oblique 30° view shows a gooseneck deformity of the LV outflow tract (*arrows*), which is formed by anterior displacement and unwedging of the aortic root and the LV outflow tract from the AV valve annuli. *Ao*, Aorta; *LV*, left ventricle.

Due to the absence of the AV membrane, the AV node is displaced posteriorly and His bundle is displaced inferiorly, resulting in left QRS axis deviation, and a PR interval prolongation can coexist as well.[10] Severe subtypes of AVSD (complete and intermediate) typically manifest in early childhood; however, the partial and transitional subtypes

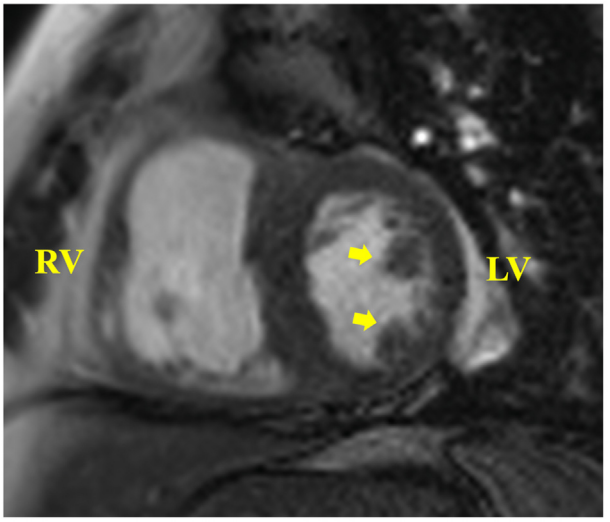

Figure 4 CMR steady-state free precession (SSFP) images, midventricular short-axis display, demonstrate the papillary muscles (*arrows*). The inferomedial papillary muscle, normally positioned in the inferior segment, is displaced counterclockwise to the inferolateral segment and is closer to the anterolateral papillary muscle than normal. *LV*, Left ventricle; *RV*, right ventricle.

can be asymptomatic until adulthood. The persistence of unrepaired AVSD carries the risk of development of Eisenmenger syndrome, paradoxical emboli, and infective endocarditis.

Although partial and transitional AVSDs have a physiology that resembles ASDs, management is still challenging given that surgery is the only definitive treatment and interventional closure is not feasible because of the proximity of the defect to the cardiac crux and the absence of an adequate rim to hold a closure device. However, Castro Rodriguez *et al*[11] reported a transcatheter closure for a small iatrogenic VSD in a patient who had a partial AVSD but had undergone a surgical repair for the ASD and a prior mitral replacement, which could provide support to hold the closure device.

With aging, these patients may develop other cardiac and noncardiac comorbidities that pose additional burdens to them and make management more difficult, in addition to their need for special centers with expertise in treating adult congenital heart diseases.[2,12] The available data in the literature about the outcome of repair in adulthood are still scarce, although a few publications ensure good short- and long-term outcomes.[6,13,14]

CONCLUSION

Congenital heart diseases should be included in the differential diagnoses of cardiopulmonary-related presentations in adulthood as some of them (including mild forms of AVSD) can remain clinically silent until the fourth or fifth decade of life.[5] The advancements in cardiac imaging modalities can help precisely determine the anatomy and hemodynamics that guide the surgical/procedural indication and subsequent planning. Multidisciplinary approaches involving adult congenital heart disease cardiologists, cardiac imagers, congenital heart disease surgeons, and social workers focusing on patients' clinical and social aspects are warranted. Ultimately, more data about the natural history of uncorrected AVSD at an advanced age and the outcomes of surgical and procedural corrections are encouraged to enrich the literature.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at https://doi.org/10.1016/j.case.2022.06.010.

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