CONGENITAL HEART DISEASE

Can atrioventricular septal defects exist with intact septal structures?

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The hallmark of an atrioventricular septal defect (AVSD) is a common atrioventricular junction, giving rise to a trileaflet left atrioventricular valve. AVSDs have the potential for interatrial shunting alone, interventricular shunting alone, or both. AVSDs without interatrial or interventricular communications have been identified at postmortem examination, but there are no reports of AVSDs with intact septal structures diagnosed in life. Six patients are described with AVSD and intact atrial and ventricular septa diagnosed echocardiographically. This report shows that AVSDs can exist without interatrial or interventricular communications and that the characteristic feature of this condition, the common atrioventricular junction with a trileaflet left atrioventricular valve, can be diagnosed in life by using cross sectional echocardiography. AVSDs with intact septal structures may be more common than previously described.

Atrioventricular septal defects (AVSDs) are characterised morphologically by the presence of a common atrioventricular junction guarded by a five leaflet common valve.[1] It is well recognised that AVSDs have the potential for interatrial shunting alone (ostium primum atrial septal defect (ASD)), for interventricular shunting alone (inlet ventricular septal defect (VSD)), or for both (most complete AVSDs) depending on the presence or absence of attachment of the bridging leaflets to the septal structures. There have also been two case reports of AVSDs with intact ventricular and atrial septal structures, diagnosed at postmortem examination.[2 3] However, AVSDs with intact septa diagnosed in life have not previously been reported. Cross sectional echocardiography has been shown to be effective in the diagnosis of an AVSD and in defining the characteristic morphological features.[4] A common atrioventricular junction is considered to be the key to the echocardiographic recognition of an AVSD.[5] Here we describe a series of six patients with the echocardiographic hallmarks of an AVSD but without interatrial or interventricular communications.

PATIENTS

Case 1

A 4 month old girl with Down's syndrome was referred for cardiovascular assessment in May 1981. The antenatal history was unremarkable. Clinically, she was acyanotic, with normal peripheral pulses and a normal cardiac impulse. Auscultation showed a normal first heart sound and a widely split second heart sound with an accentuated pulmonary component. There was a grade 2/6 ejection systolic murmur loudest at the third left intercostal space. There were no signs of cardiac failure.

Her chest radiograph showed laevocardia, a left sided aortic arch, minimal cardiac enlargement, and slightly increased pulmonary vascular markings. ECG showed sinus rhythm with a superior mean frontal QRS axis (−60°) and upright right sided T waves indicating right ventricular hypertrophy.

Cross sectional echocardiography showed a trileaflet left atrioventricular valve and a small VSD. There was no ASD.

She has remained asymptomatic from a cardiac point of view and was last reviewed at the age of 22 years in March 2002, when she had a normal cardiovascular clinical examination. Echocardiography at the time showed a common atrioventricular valve with no interatrial or interventricular communications, her small VSD having closed spontaneously. There was also trivial atrioventricular valve regurgitation.

Case 2

A 2 day old boy was referred for assessment because of an abnormal ECG in September 1986. He was asymptomatic and clinically pink in air, with normal peripheral pulses and a normal cardiac impulse. On auscultation, he had normal heart sounds and a grade 2/6 ejection systolic murmur at the third left intercostal space. The rest of the physical examination was normal. His ECG showed sinus rhythm with a superior QRS axis and left ventricular dominance. On review at the age of 4 months, he remained asymptomatic and his ECG had become more normal, showing sinus rhythm with a slightly superior QRS axis of −10° and a normal infant RS progression pattern.

He has remained asymptomatic. On review at the age of 9 years, his clinical examination was normal. Cross sectional echocardiography in 1995 showed a trileaflet left atrioventricular valve typical of an AVSD, with very trivial regurgitation. The atrial septum was intact, with no ostium primum ASD, and likewise the ventricular septum was intact.

At last review, this patient remains asymptomatic and leads a normal life.

Case 3

A neonate with Down's syndrome was referred for assessment in March 1999. He was asymptomatic, not cyanosed, and had normal femoral pulses and a normal cardiac impulse. On auscultation, he had normal heart sounds and no murmurs. An echocardiogram showed a trileaflet left atrioventricular valve with no regurgitation. There was no ASD and no VSD.

Abbreviations: ASD, atrial septal defect; AVSD, atrioventricular septal defect; VSD, ventricular septal defect

On review at the age of 5 years, he remains well and asymptomatic. Clinically, he has normal heart sounds and a very soft systolic murmur audible midway between the lower left sternal edge and apex. Cross sectional echocardiography confirmed an AVSD with a trileaflet left atrioventricular valve with trivial regurgitation. The atrial and ventricular septa were intact.

Case 4

A 2 year old boy underwent resection of subaortic stenosis and suturing of the zone of apposition of the left aspect of the superior and inferior bridging leaflets in August 2002. On review at the age of 4 years, he was asymptomatic. Clinically, he was not cyanosed and had normal peripheral pulses and a normal cardiac impulse. On auscultation, he had normal first and second heart sounds and a grade 3/6 ejection systolic murmur loudest at the right upper sternal edge. There was no carotid thrill.

The ECG showed sinus rhythm with a QRS axis of 15° and voltage criteria for left ventricular hypertrophy.

Cross sectional echocardiography showed a common atrioventricular junction with trivial regurgitation through the (repaired) trileaflet left atrioventricular valve. There was also a bicuspid aortic valve and subaortic stenosis with a Doppler derived peak instantaneous pressure gradient of 30 mm Hg and no left ventricular hypertrophy. The atrial and ventricular septa were intact.

Case 5

A 5 week old girl with Down's syndrome was referred for cardiovascular assessment in April 2004. She was asymptomatic, not cyanosed, had normal peripheral pulses, and had a normal cardiac impulse. Auscultation found fixed splitting of the second heart sound with a slightly accentuated pulmonary component and no murmurs.

Cross sectional echocardiography showed the presence of an ostium secundum ASD with left to right shunting and mild right heart volume overload. There was also a trileaflet left atrioventricular valve and a common atrioventricular junction, with no primum ASD or VSD.

On review in January 2005 at the age of 20 months, she remained asymptomatic and had a normal cardiovascular examination. Echocardiography confirmed that the secundum ASD had closed. The atrial and ventricular septa were intact. A common atrioventricular junction with a trileaflet atrioventricular valve was not regurgitant (fig 1–4). In addition, unwedging of the aorta was seen. Because of the common atrioventricular junction, the aorta was displaced anteriorly and to the right instead of its normal position wedged between the mitral and tricuspid valve rings.

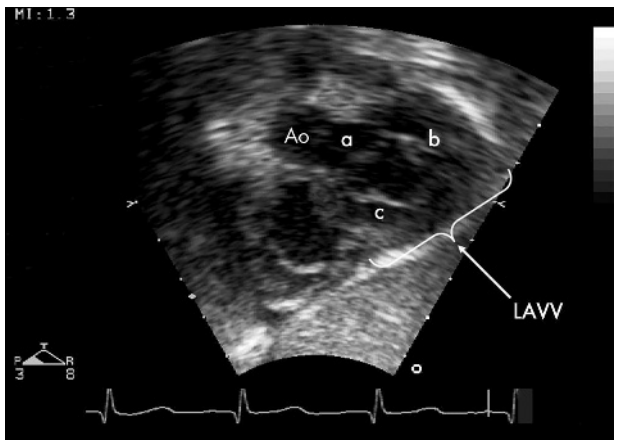

Figure 1 Case 5: subcostal view of the common atrioventricular junction with the trileaflet atrioventricular valve open in atrial systole. The aortic valve is displaced anteriorly and to the right in an unwedged position, sitting above the superior bridging leaflet. a, superior bridging leaflet; b, left mural leaflet; c, inferior bridging leaflet. AO, aorta; LAVV, left atrioventricular valve.

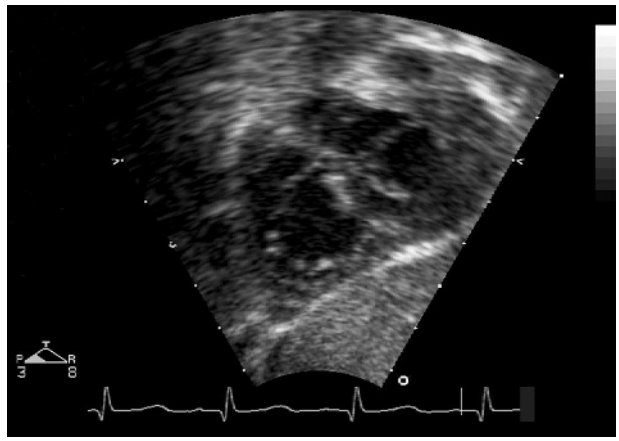

Figure 2 Case 5: subcostal view of the common atrioventricular junction at the end of atrial systole.

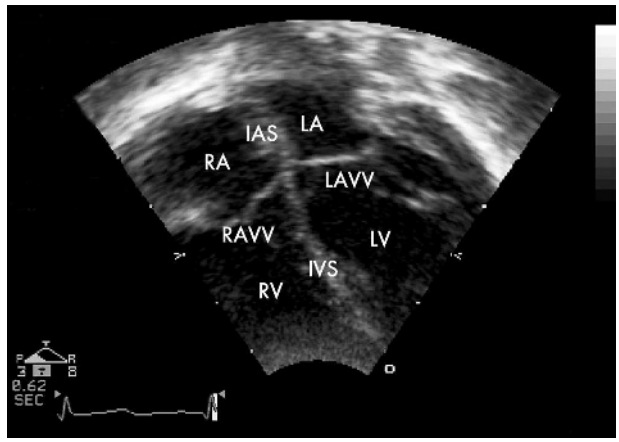

Figure 3 Case 5: apical four chamber view showing the absence of normal offsetting of the atrioventricular valves and intact atrial and ventricular septa in systole. IVS, interventricular septum; LA, left atrium; LV, left ventricle; RA, right atrium; RAVV, right atrioventricular valve; RV, right ventricle.

Case 6

A 2 week old boy was referred for cardiac assessment in April 2004 following the diagnosis of Down's syndrome at birth. He was asymptomatic, with a normal cardiovascular physical examination. Cross sectional echocardiography showed a common atrioventricular junction with a common orifice divided into two separate trileaflet atrioventricular valves. There was no ASD and no VSD.

DISCUSSION

Promoting the term atrioventricular septal defect rather than atrioventricular canal or endocardial cushion defect, Piccoli *et al*[6] described the essential morphological features of an AVSD as, firstly, disproportion between the inlet and outlet portions of the interventricular septum and, secondly, malorientation of the aortic valve relative to the atrioventricular valves. These anatomical features were accompanied by defects in the atrial or ventricular septa and by malformations of the atrioventricular valves, leading to complete defects with a common atrioventricular valve annulus or partial defects

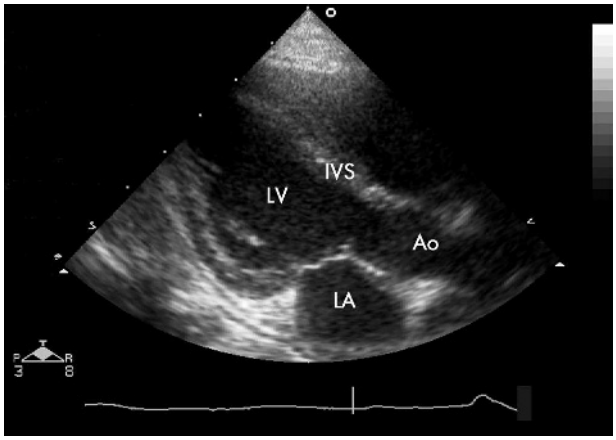

Figure 4 Case 5: parasternal long axis view showing an intact IVS. Ao, aorta.

where tricuspid and mitral valves were separate. It has since become clear, however, that a common atrioventricular junction, guarded by a common atrioventricular valve, is the characteristic feature of all forms of AVSD.[1] This common valve consists of five leaflets: a superior bridging leaflet, an inferior bridging leaflet, a left mural leaflet, a right mural leaflet, and a right anterolateral leaflet. In a complete AVSD, the common valve guards a single, common orifice. In the partial form, a tongue of tissue joins the free margins of the superior and inferior bridging leaflets, creating two separate orifices within a common valve ring.

In a complete AVSD, there is usually an interatrial communication (ostium primum ASD) between the inferior margin of the atrial septum and the superior aspect of the common atrioventricular valve, and a large interventricular communication between the crest of the ventricular septum and the inferior margin of the common valve. In a partial AVSD, the bridging leaflets fuse to the crest of the ventricular septum, leaving only an ostium primum ASD. They may also fuse with the atrial septum, leaving only a VSD.

A much rarer form of AVSD occurs when the common leaflet is fused to both the atrial septum and the crest of the ventricular septum, creating an AVSD with no interatrial or interventricular communication. There have been two case reports of such an AVSD, where the diagnosis was made at necropsy.[2 3] In addition, one of the pathological specimens from the morphological study of Piccoli *et al*[6] in 1979 was also found to have only an isolated cleft between the anterior and posterior components of the left atrioventricular valve, and intact atrial and ventricular septa. This is the first report of patients with an AVSD and intact septal structures diagnosed in life.

Cross sectional echocardiography has been shown to be a useful non-invasive tool in the assessment of AVSDs.[4] A combination of echocardiographic views is required to analyse the defect, both to define the anatomical potential for interatrial or interventricular shunting and to examine the atrioventricular junction.[4 5] It is this common atrioventricular junction that is key to the echocardiographic diagnosis of an AVSD. As discussed previously, in a partial AVSD, a rim of tissue divides the common valve into two separate orifices. As a result, the left atrioventricular valve has three leaflets (comprising the left halves of each bridging leaflet and the left mural leaflet) and does not resemble a normal mitral valve. In all our patients, AVSD was diagnosed echocardiographically based on the finding of a common atrioventricular junction and a trileaflet left sided atrioventricular valve. This is consistent with the findings of previous studies of AVSDs with echocardiography.[4 5 7–9] Furthermore, cross sectional echocardiography has been shown to be useful in identifying another characteristic feature of AVSDs, the anterior and rightward displacement of the aorta in relation to the common atrioventricular junction.[4 6] This "unwedging" of the aorta (fig 1) was identified in all our patients. In addition, four of our patients had Down's syndrome and all but one of the ECGs available to us showed a superior mean frontal QRS axis, consistent with a diagnosis of AVSD.

In the context of a trileaflet left atrioventricular valve and intact septal structures, the presence of a common atrioventricular junction is important in distinguishing an AVSD from an isolated cleft of the mitral valve. Previously, isolated cleft of the mitral valve had been described as a forme fruste of AVSD, but a mitral valve with a cleft in the anterior leaflet and the trileaflet left atrioventricular valve of an AVSD have now been shown to be morphologically distinct.[8 10] The fundamental difference is that the cleft in an otherwise normal mitral valve is supported by a discrete left atrioventricular junction or valve ring separated from the right side of the heart by well formed atrioventricular septal structures.[8] In contrast, in an AVSD with intact septa, the left atrioventricular valve is supported by a valve ring common to both atrioventricular valves. Furthermore, the valves are at the same level, unlike the normal offsetting of the mitral and tricuspid valves, where the tricuspid is nearer to the apex of the ventricle. In addition, echocardiographic identification of papillary muscle position and mural leaflet size have been shown to be useful in the differentiation of AVSD from cleft mitral valve.[9] Further support for the distinction between an isolated cleft in the anterior leaflet of the mitral valve and the trileaflet nature of the left atrioventricular valve in an AVSD comes from the recent study of Van Praagh *et al*[11] in cleft mitral valve without ostium primum defect. They found that in patients with normally connected great arteries and a VSD the inlet dimension of the left ventricular septal surface was shorter than its outlet dimension, characteristic of atrioventricular canal malformations (AVSD). In line with abnormal ventriculoarterial connections there was no inlet-outlet disproportion of the ventricular septum and by implication no similarities with an AVSD. Furthermore the "cleft" in the AVSD type of VSD with normally connected great arteries pointed to the inlet part of the interventricular septum, whereas in patients with abnormal ventriculoarterial connections the cleft pointed to the outlet of the ventricles.

Five of the patients we have described were asymptomatic. The condition was detected in four as part of routine cardiovascular assessment for patients with Down's syndrome and, in the fifth, as a result of an abnormal ECG. Two patients had additional congenital cardiac malformations. In one patient this was clinically significant and he required surgery for subaortic stenosis and left atrioventricular valve regurgitation. He has remained asymptomatic since. Four of the patients do have trivial atrioventricular valve regurgitation that is not clinically significant. However, because of the trileaflet left atrioventricular valve, atrioventricular valve regurgitation has the potential to be more significant in cases of AVSD with intact septa than in isolated mitral valve clefts.

Down's syndrome is a well recognised association of AVSD but less so with isolated mitral valve clefts.[10] In the comparison by Kohl and Silverman[9] between cleft mitral valve and AVSD, none of the 11 patients with cleft mitral valve had Down's syndrome, whereas six of 11 with AVSD did. A recent study examining possible aetiological differences between complete and partial AVSDs has suggested that patients with a complete AVSD or partial AVSD with an interventricular communication have higher rates of Down's syndrome than do patient with a partial AVSD and interatrial communication alone.[12] This may imply that Down's syndrome does not have a particularly strong association with AVSD with intact septal structures, but this has not been our

experience. In our unit and many others, however, all patients with Down's syndrome are screened by cross sectional echocardiography, even in the absence of abnormal cardiac signs. Thus, given an awareness of the possibility of AVSD with intact septal structures, the condition will be picked up in the Down's syndrome group, whereas chromosomally normal children with no abnormal cardiac signs would not normally have a two dimensional echocardiogram and would therefore not be identified.

This is the first report of patients with AVSD and intact atrial and ventricular septal structures diagnosed echocardiographically. These cases show that AVSDs can exist without interatrial or interventricular communications and that the hallmark of this condition, the common atrioventricular junction together with a trileaflet left atrioventricular valve, can reliably be detected by cross sectional echocardiography.

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REFERENCES

1 **Becker AE**, Anderson RH. Atrioventricular septal defects. What's in a name? *J Thorac Cardiovasc Surg* 1982;**83**:461–9.
2 **Silverman NH**, Ho SY, Anderson RH, *et al*. Atrioventricular septal defect with intact atrial and ventricular septal structures. *Int J Cardiol* 1984;**5**:567–72.
3 **Ho SY**, Russell G, Gerlis LM. Atrioventricular septal defect with intact septal structures in a 74-year-old. *Int J Cardiol* 1990;**26**:371–3.
4 **Silverman NH**, Zuberbuhler JR, Anderson RH. Atrioventricular septal defects: cross-sectional echocardiographic and morphologic comparisons. *Int J Cardiol* 1986;**13**:309–31.
5 **Falcão S**, Daliento L, Ho SY, *et al*. Cross sectional echocardiographic assessment of the extent of the atrial septum relative to the atrioventricular junction in atrioventricular septal defect. *Heart* 1999;**81**:199–205.
6 **Piccoli GP**, Gerlis LM, Wilkinson JL, *et al*. Morphology and classification of atrioventricular defects. *Br Heart J* 1979;**42**:621–32.
7 **Smallhorn JF**, de Leval M, Stark J, *et al*. Isolated anterior mitral cleft: two dimensional echocardiographic assessment and differentiation from ''clefts'' associated with atrioventricular septal defect. *Br Heart J* 1982;**48**:109–16.
8 **Sigfusson G**, Ettedgui JA, Silverman NH, *et al*. Is a cleft in the anterior leaflet of an otherwise normal mitral valve an atrioventricular canal malformation? *J Am Coll Cardiol* 1995;**26**:508–15.
9 **Kohl T**, Silverman NH. Comparison of cleft and papillary muscle position in cleft mitral valve and atrioventricular septal defect. *Am J Cardiol* 1996;**77**:164–9.
10 **Di Segni E**, Edwards JE. Cleft anterior leaflet of the mitral valve with intact septa: a study of 20 cases. *Am J Cardiol* 1983;**51**:919–26.
11 **Van Praagh S**, Porras D, Oppido G, *et al*. Cleft mitral valve without ostium primum defect: anatomic data and surgical considerations based on 41 cases. *Ann Thorac Surg* 2003;**75**:1752–62.
12 **Loffredo CA**, Hirata J, Wilson PD, *et al*. Atrioventricular septal defects: possible etiologic differences between complete and partial defects. *Teratology* 2001;**63**:87–93.

IMAGES IN CARDIOLOGY

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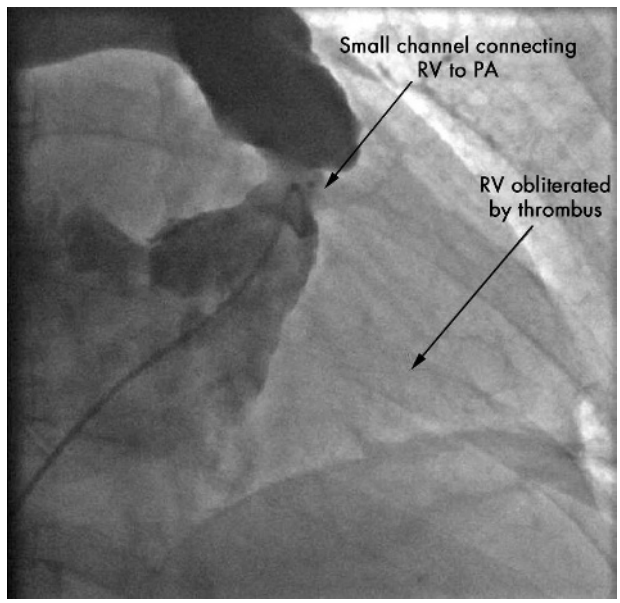
Thrombotic obliteration of the right ventricular cavity

A 54 year old woman presented with intermittent dull central chest pain, which occurred at rest and was not exacerbated by exertion. She also complained of shortness of breath and reduction of her exercise tolerance to a few yards only over the previous four weeks. She smoked 15 cigarettes per day and drank alcohol only occasionally. She was not hypertensive or diabetic. Physical examination showed that her vital signs were normal, jugular venous pressure was raised, heart sounds were normal and lung auscultation was unremarkable. Full blood count, creatinine, and troponin I were within normal range, erythrocyte sedimentation rate was 22 mm/h, C-reactive protein was 93 mg/l, and the autoimmune screen was negative. The ECG showed sinus rhythm with T wave inversion anteriorly, the chest radiograph showed mild cardiomegaly, and echocardiography showed a large space-occupying lesion involving most of the right ventricle with reduced right ventricular function. Right ventriculography showed that the right ventricle was completely obliterated by the mass, with only a slit-like opening connecting the right ventricle to the pulmonary artery (panel). Biopsy of the mass showed clots and thrombus only and no myocardial tissue. Subsequently, the patient's condition suddenly deteriorated and her blood pressure became unrecordable. Emergency cardiac surgery was performed and a large right ventricular thrombus, which was obliterating the ventricle, was removed. The patient made a good recovery.



Obliteration of the right ventricular cavity by thrombus. This right ventriculogram shows almost complete obliteration of the right ventricular cavity by the thrombus, leaving only a small channel between the right ventricle and pulmonary artery.

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