ATRIOVENTRICULAR SEPTAL DEFECT: FROM FETUS TO ADULT

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1879

Heart 2006;92:1879–1885. doi: 10.1136/hrt.2006.093344

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The term “atrioventricular septal defect” (AVSD) covers a spectrum of congenital heart malformations characterised by a common atrioventricular junction coexisting with deficient atrioventricular septation. In ostium primum atrial septal defect (ASD) there are separate atrioventricular valvar orifices despite a common junction, while in complete AVSD the valve itself is also shared.

The estimated incidence of the condition in the era of two-dimensional echocardiography varies from 0.24/1000^{w1} live births to 0.31/1000^{w2} live births. There is a strong association with Down's syndrome, with half of patients with AVSD in the population of Bohemia also complicated by Down's syndrome.^{w3} Conversely in a Toronto study about one third of patients with Down's syndrome had a complete AVSD and 5% had an ostium primum ASD.^{w4} In a prospective screening study within the relatively static Northern Irish population the incidence of AVSD in Down's syndrome was 17% and the overall incidence of congenital heart disease in Down's syndrome was 42%.¹

Three different genetic patterns are described in AVSD: the association with Down's syndrome, as an autosomal dominant trait, and isolated. Molecular studies of patients with congenital heart disease and partial duplications of chromosome 21 have proposed DSCAM (Down's syndrome cell adhesion molecule) as a candidate gene causing congenital heart disease in Down's syndrome.² Cases with autosomal dominant inheritance, however, are not linked to chromosome 21.^{w5}

Over the past four decades the management of the complete form of the condition has evolved from palliative pulmonary artery banding in infancy with later repair to primary repair in early infancy to prevent the development of pulmonary vascular obstructive disease. Decreasing operative mortality has significantly altered the prognosis of these patients with and without Down's syndrome. The improved prognosis for patients with Down's syndrome and AVSD has implications for the management of patients diagnosed antenatally. The postnatal and longer term outcomes are influenced by the presence of associated defects, such as ventricular hypoplasia, and the management of these may be both difficult and controversial.

SPECTRUM OF ANATOMY

The essential morphological hallmark of an AVSD is the presence of a common atrioventricular junction as compared to the separate right and left atrioventricular junction in the normal heart. Other morphological features include defects of the muscular and membranous atrioventricular septum and an ovoid shape of the common atrioventricular junction with unwedging of the left ventricular outflow tract from the normal position between the tricuspid and mitral valve. There is disproportion of outlet and inlet dimensions of the left ventricle, with the former greater than the latter as compared to the normal heart where both dimensions are similar.

The valve leaflet morphology in AVSD bears little resemblance to the arrangement of the leaflets of normal mitral and tricuspid valves. There are essentially five leaflets, two of which are bridging leaflets across the crest of the interventricular septum (fig 1). In ostium primum ASD there are separate right and left valve orifices due to a tongue of valvar tissue joining the bridging leaflets, but the atrioventricular junction remains a common structure (fig 2). In complete AVSD there is a space between the bridging leaflets and therefore the atrioventricular valvar orifice is common.

The potential for left to right shunting is related to the bridging leaflets being attached to the atrial septum, to the ventricular septum, or floating within the AVSD (fig 3). In Rastelli's classification the superior bridging leaflet was mostly contained within the left ventricle in type A, with increased extension into the right ventricle in types B and C.^{w6} The potential for ventricular shunting tended to increase from type A to type C. This classification has been superseded by a simpler independent description of the two aspects of the anatomy which determine the variability and clinical presentation.³ The first is the individual leaflet arrangement within the common atrioventricular junction. The second is the relationship of the bridging leaflets of the common valve to the atrial and ventricular septal structures.

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Figure 1 The arrangement of the atrioventricular valve leaflets in complete atrioventricular septal defect (AVSD) with a common atrioventricular junction and common valve.

ASSOCIATED DEFECTS

Subaortic stenosis

The left ventricular outflow tract is elongated and appears relatively narrowed in patients with AVSD. This is mainly due to the anterior unwedged position of the outflow tract and is particularly notable in patients with ostium primum ASD where the superior bridging leaflet is firmly fixed to the crest of the ventricular septum.^{w7} Preoperatively significant obstruction, however, is usually due to additional lesions, such as fibrous subaortic shelf or fibromuscular tunnel,^{w8} fibrous tissue tags^{w9} or anomalous insertion of left ventricular papillary muscles.^{w10}

Ventricular hypoplasia

In most patients with AVSD the right and left components of the common atrioventricular junction are comparable and the

Figure 2 The arrangement of the atrioventricular valve leaflets in ostium primum atrial septal defect (ASD) with a common atrioventricular junction but separate valvar orifices for right and left ventricles.

ventricles are similarly sized (balanced AVSD). In a minority of cases the common atrioventricular junction is committed to the right or left ventricle leading to right or left ventricular dominance and relative hypoplasia of the opposing ventricular chamber.^{w11} Extreme commitment of the common atrioventricular junction to the left ventricle has been termed "double inlet left ventricle with a common valve".^{w12}

Tetralogy of Fallot

The reported incidence of the combination of AVSD and tetralogy of Fallot is 5% of all patients with AVSD, while the latter complicates 16.5% of cases of tetralogy of Fallot.^{w13} The combination is more common in patients with Down's syndrome whereas most other associated lesions complicating AVSD are more common in patients without Down's syndrome.⁴

Atrial isomerism

Complex forms of AVSD are found in the majority of hearts with right atrial isomerism and in around half with left atrial isomerism.^{w14} The former tend to have univentricular hearts, often with a common atrium, while the latter tend to have biventricular hearts. Complete heart block is common in left atrial isomerism associated with AVSD.

Other associated lesions include double-orifice atrioventricular valve,^{w15} parachute left atrioventricular valve,^{w15} and coarctation of the aorta.^{w8} There are occasional case reports of associated truncus arteriosus,^{w16} transposition of the great arteries^{w17} and congenitally corrected transposition of the great arteries.^{w18} Ebstein's anomaly,^{w19} hypertrophic cardiomyopathy,^{w20} cor triatrium,^{w21} and absent pulmonary valve syndrome^{w22} have also been reported.

FETAL DIAGNOSIS

The view of the heart most commonly used in routine antenatal ultrasound anomaly scanning is the four-chamber view. AVSD is one of the lesions potentially detectable on this view.

The key diagnostic feature on the obstetric four-chamber view of the heart is the presence of a common atrioventricular valve. In a good four-chamber view the defects in the atrial and ventricular septum should also be visible (fig 4). Antenatal detection rates of AVSD, however, remain lower than might be expected. In one UK study of 92 consecutive liveborn infants with AVSD, only 29% were detected by routine obstetric ultrasound.⁵ Machlitt has recently reported that antenatal detection using the four-chamber view can be significantly improved by measuring the ratio of atrial to ventricular length (AVL).^{w23} In this study if a cut-off value for the AVL ratio over 0.6 was chosen the detection rate of AVSD was 82.6% with a 5.7% false positive rate.

The spectrum of AVSD in fetal life is different from that diagnosed postnatally. Up to 45% of those diagnosed in utero may have associated heterotaxy syndromes, particularly left atrial isomerism.^{6 w24 w25} Fetal echocardiography in cases of AVSD therefore should include determination of atrial situs, ventriculo-arterial connections, ventricular size and aortic arch calibre. The detection of these associated abnormalities is very important so that when counselling parents an accurate indication of outcome can be given. The mortality is higher in those with other associated cardiac abnormalities; in particular, the combination of left atrial isomerism, AVSD and congenital complete heart block, often with fetal hydrops, has a very poor prognosis with a large fetal loss.⁷

Figure 3 The potential for shunting in AVSD relates to the connections between the bridging leaflets and the atrial and ventricular septum.

Antenatal diagnosis of this lesion is particularly important because of the strong association with chromosomal problems, especially trisomy 21. Abnormal karyotype is more common in isolated AVSD (48–58%).^{5–7 w25} The relative risk of trisomy 21 in a pregnancy in which a diagnosis of AVSD is the only cardiac defect has been reported to be 107.^{w26}

CLINICAL PRESENTATION

The clinical presentation (table 1) and course in AVSD relate to the specific morphology of the defect and the presence of associated defects. In infants with complete AVSD and a sizeable interventricular component, congestive cardiac failure is likely to develop within the first few months of life as pulmonary vascular resistance falls. If there is significant regurgitation of the common atrioventricular valve, associated coarctation of the aorta or ventricular imbalance, cardiac failure may occur much earlier and often within the

first week of life. Without surgery many of these patients will die in infancy and those who survive will develop pulmonary vascular disease and eventually die with Eisenmenger’s syndrome.⁸ There is a subgroup of patients with complete AVSD and a large interventricular component, often with Down’s syndrome, where cardiac failure does not occur, and this phenomenon relates to persisting elevation of pulmonary vascular resistance from birth.^{w27}

The newborn with AVSD may present with a mild degree of central cyanosis. This finding relates to bidirectional shunting at both the atrial and ventricular level in the presence of elevated pulmonary vascular resistance at birth. The only positive precordial findings of a congenital heart defect at this time may be a right ventricular impulse and accentuated pulmonary component of the second heart sound. A precordial murmur may, at this stage, be much abbreviated or absent.¹

Figure 4 Fetal cardiac ultrasound at 23 weeks gestation demonstrating complete AVSD with a common atrioventricular valve and large atrial and ventricular septal defects. LV, left ventricle; RV, right ventricle.

Table 1 Postnatal diagnosis of AVSD	
▶ Clinical	
–	cyanosis mild or absent
–	congestive cardiac failure
–	right ventricular impulse
–	increased pulmonic component second heart sound
–	variable ejection systolic murmur, apical mid-diastolic murmur (in large left to right shunt), pansystolic murmur (with atrioventricular valve regurgitation)
▶ ECG	
–	prolonged PR interval
–	superior frontal QRS axis
–	partial right bundle branch block
–	superior p wave axis (in associated left atrial isomerism)
▶ Chest radiograph	
–	cardiomegaly
–	prominent pulmonary conus
–	increased pulmonary vascularity
–	left aortic arch
–	left bronchial isomerism (in associated left atrial isomerism)

In patients with complete AVSD and a small interventricular component, or in patients with ostium primum ASD where atrioventricular valve regurgitation is minimal, cardiac failure is rare and clinical symptoms may be minimal or absent in infancy and childhood. Without surgery, however, there is considerable longer term morbidity and mortality with only 25% survival beyond 40 years of age.^{w28}

DIAGNOSIS

Chest radiograph

The chest x-ray in AVSD normally demonstrates levocardia and a left-sided aortic arch. There is usually cardiomegaly and pulmonary plethora, the former more pronounced with associated atrioventricular valve regurgitation. Abnormalities in bronchial branching may indicate associated atrial isomerism.^{w14}

Electrocardiogram

The most characteristic feature of AVSD on the ECG is superior orientation of the frontal QRS loop.^{w29} The p wave axis may also be superior in associated left atrial isomerism.^{w30} First degree heart block is present in the majority and prolongation of the QRS complex in over half of patients with AVSD.^{w30} There is an increased risk of complete heart block due to displacement of the atrioventricular node. The precordial voltages may indicate relative ventricular hypoplasia.^{w31}

Echocardiogram

The echocardiographic features of AVSD are listed in table 2. Highly accurate evaluation and diagnosis of AVSD can be achieved by two-dimensional echocardiography^{9 w32} (fig 5). AVSD can be distinguished from inlet ventricular septal defect and isolated mitral valve cleft by estimation of the left ventricular inlet/outlet ratio and per cent left atrioventricular valve guarded by the posterior leaflet.¹⁰ Associated defects such as obstructed inflow and outflow lesions¹¹ or unbalanced AVSD¹² can be readily identified and assessed. When two-dimensional echocardiography is combined with Doppler and colour-flow studies, patients can be appropriately selected as suitable for primary surgical repair.^{w33} In an overview of echocardiography in AVSD Smallhorn details the basic morphological features and preoperative risk factors.¹³ He states that in the current era it is rarely necessary to perform other investigations before surgical repair. Three-dimensional echocardiography has provided similar information with some additional detail in the assessment of dynamic atrioventricular valve morphology and the mechanism of valve incompetence.^{w34} Transoesophageal echocardiography is probably best utilised intraoperatively to improve surgical outcomes and decrease the incidence of reoperation^{w35} (fig 6). Intraoperative epicardial echocardiography provides similar information to enable the surgeon to minimise residual defects.^{w36}

Table 2 Echocardiographic features of AVSD

- ▶ Loss of normal offsetting of atrioventricular valves (absence of the atrioventricular muscular septum)
- ▶ Abnormal configuration of the atrioventricular valves
- ▶ Trifoliate appearance of the left atrioventricular valve (with "cleft")
- ▶ Left ventricular inlet/outlet disproportion
- ▶ Abnormal position of the left ventricular papillary muscles

Figure 5 Transthoracic echocardiogram in an infant with AVSD. The precordial four-chamber view demonstrates a large atrial component with chordal attachments from the superior bridging leaflet to the crest of the interventricular septum. CT, chordae tendinae; IVS, interventricular septum; LA, left atrium; RA, right atrium.

Magnetic resonance imaging

Magnetic resonance imaging can provide images similar to those obtained by echocardiography.^{w37} This mode of imaging may be more accurate than echocardiography in predicting the size of the ventricular component of the defect and identifying ventricular hypoplasia.^{w38} It is possible to measure accurately the regurgitant fraction in valve regurgitation and this may be of value not only in preoperative assessment but in longer term follow-up.^{w39}

Angiography

Since angiography is no longer part of the normal preoperative assessment of patients with AVSD the main indication for cardiac catheterisation is to measure pulmonary vascular resistance. This will be necessary in borderline cases before surgical correction.^{w40} If the resistance is found to be elevated, reversibility with oxygen, inhaled nitric oxide and other potent pulmonary vasodilators can be assessed.

Figure 6 Transoesophageal echocardiogram in an infant with AVSD. Transgastric short-axis view of the left ventricle demonstrates trifoliate structure of the left atrioventricular valve in diastole. IBL, inferior bridging leaflet; IVS, interventricular septum; ML, mural leaflet; SBL, superior bridging leaflet.

Screening

In the clinical setting of Down's syndrome, Tubman *et al* have advocated screening for congenital heart disease by echocardiography shortly after birth.¹ The rationale is the high incidence of congenital heart disease in Down's syndrome and the variable clinical presentation. Early diagnosis of structural cardiac defects and in particular complete AVSD allows institution of medical and supportive treatment at an early stage and a plan for timely surgical intervention should this be indicated.^{w41}

TREATMENT

Medical treatment

Medical treatment in complete AVSD includes the management of congestive cardiac failure with diuretics and a vasodilator such as captopril. The use of digoxin is more controversial. Associated feeding difficulties and failure to thrive are managed with nasogastric tube feeds and added calories. In most cases medical treatment is short-term and directed at optimising the patient's condition for corrective surgery.

There is no role for catheter intervention in complete AVSD or ostium primum ASD as the position of the septal defects renders them unsuitable for device occlusion.

Surgery

The mainstay of management in AVSD is surgical correction of the defect. The objectives of surgical correction are to close all septal defects and to repair the atrioventricular valve. In the current era the aim is to provide surgical correction in complete AVSD within the first few months of life and certainly before six months in order to avoid the development of pulmonary vascular disease.¹⁴ Low operative mortalities have been reported and are generally < 10%¹⁵ and in some series < 5%.^{w42} In a recent report from Leiden the operative mortality for complete AVSD repair in Down's syndrome has fallen to 0%.^{w41} Prifti *et al*, however, urge a note of caution in the trend for earlier repair in that although there was no difference in operative mortality in their reported study between infants weighing < 5 kg body weight and those > 5 kg, the former group had a higher incidence of late reoperation for left atrioventricular valve regurgitation.¹⁵ By contrast a study in San Francisco concludes that with meticulous techniques the age at repair for AVSD from neonates to older infants does not influence outcome or valve function.^{w43}

Septal closure

In complete AVSD the surgical options for closing the septal defects are with a one-patch or two-patch technique. Differing views are held with some surgeons opting for a single patch technique in all patients^{w43 w44} or a modification with patch closure of the atrial defect and direct suture of the common atrioventricular valve leaflets to the crest of the ventricular septum.^{w45} Others opt for a two-patch repair in all patients with similar outcome for operative mortality and residual left atrioventricular valve regurgitation.^{w46} Other centres utilise both techniques depending on the individual valve morphology.¹⁶ Ebels postulates that the two-patch technique is only warranted when the superior and inferior leaflets bridge the septum extensively, seen most often in patients with Down's syndrome.^{w36}

Left atrioventricular valve repair

A second area of surgical controversy relates to repair of the left atrioventricular valve. Some argue that the zone of apposition

Practical point

Clinical examination alone is unreliable in postnatal screening for congenital heart disease in neonates with Down's syndrome. An ECG demonstrating a superior frontal QRS axis is strongly suggestive of AVSD, but the "gold standard" investigation in confirming or excluding the diagnosis is transthoracic echocardiography

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(confusingly called cleft) between the bridging leaflets should be routinely closed in order to restore a bifoliate valve.^{w47–50} Others take the view that the trifoliate arrangement of the left atrioventricular valve should be respected and only repaired if regurgitant.^{w51 w52} A recent study from Germany attempts to answer the question by comparing outcomes from each approach in the same centre.^{w53} This study concludes that the zone of apposition should be surgically addressed whenever the morphology of the left atrioventricular valve allows for closure without producing stenosis.

Associated defects

Associated defects such as subaortic stenosis may have to be corrected at the time of AVSD repair. Careful preoperative assessment and intraoperative echocardiography have been shown to optimise outcome in these patients.^{w35} Patients with a combination of AVSD and tetralogy of Fallot were historically managed with a systemic to pulmonary shunt and repair later in childhood.^{w54} A number of recent series now report acceptable outcomes from primary repair in infancy.^{w55–57}

Unbalanced ventricles

The management of AVSD associated with unbalanced ventricles remains difficult. Where the right or the left ventricle is grossly underdeveloped, single ventricle palliation may be considered.^{w58} Modest right ventricular hypoplasia may be better tolerated and in some patients may permit a biventricular repair and in some a one-and-a-half ventricle repair (biventricular repair with bidirectional cavopulmonary anastomosis).^{w59 w60} The most difficult cases to refer appropriately for surgical repair are those with mild to moderate left ventricular hypoplasia. Cohen *et al* have suggested that the calculation of an atrioventricular valve index (AVVI) echocardiographically may be helpful.¹² In the subcostal left anterior oblique view the area of the atrioventricular valve apportioned over each ventricle is measured and expressed as left/right valve area. They postulate that if the AVVI is < 0.67 in the presence of a large ventricular septal defect, a single ventricle surgical approach should be considered.

Ostium primum ASD

In ostium primum ASD the timing of surgery is less critical. In patients with minimal atrioventricular valve regurgitation there may be few symptoms throughout infancy and early childhood and the timing of surgery is often elective with a low operative mortality.^{17 w61 w62} The prognosis is less good for patients presenting in infancy with cardiac failure as early presentation is often a predictor of severe left atrioventricular valve regurgitation or associated defects such as subaortic stenosis, coarctation of the aorta or dominant right ventricle.^{w63} In those patients with significant left heart obstruction or left ventricular hypoplasia a Norwood operation may be considered.^{w64}

Down's syndrome

In patients with AVSD the onset of pulmonary vascular disease is earlier in patients with associated Down's syndrome dictating early surgical repair.^{w65} The spectrum of anatomy in AVSD also varies in patients with coexisting Down's syndrome where extensive bridging of both superior and inferior bridging leaflets is more common and left ventricular hypoplasia and left heart obstructive lesions less common.^{w66} This anatomy may in fact be more favourable for surgical repair and Rizzoli has demonstrated that Down's syndrome per se is not a risk factor for surgery.^{w67} A Japanese study concludes that Down's syndrome does not affect the long term results of complete AVSD when the defect is repaired during the first year of life.^{w68} A recent report from Italy involving over 200 patients demonstrates a decreased risk for biventricular repair in Down's syndrome and lower mortality in cases with associated defects.^{w69} Twenty years ago there was considerable debate regarding cardiac surgery in patients with Down's syndrome versus a non-interventional approach.¹⁸ These recent studies together with quality of life issues have swung the pendulum towards surgical intervention in most patients.

FOLLOW-UP Complete AVSD

Without surgery the natural history of complete AVSD has been well documented by Berger *et al* with only 4% survival beyond 5 years old.⁸ In surviving patients following surgery the most common reason for reoperation in the large Toronto series was left atrioventricular valve regurgitation followed by subaortic stenosis, residual ventricular septal defect and late onset complete heart block.¹⁶ The overall 10 year survival was 83%. Al-Hay reported an increased incidence of postoperative left atrioventricular valve regurgitation in the chromosomally normal patients related to a higher incidence of dysplastic atrioventricular valve.^{w70} Rhodes reports a mild increase in the degree of residual left atrioventricular valve regurgitation in the initial 30 months postoperatively with little further deterioration thereafter.¹⁹ Table 3 lists the long-term complications following surgical repair in AVSD.

Atrioventricular septal defect (AVSD): key points

- ▶ The spectrum of AVSD diagnosed antenatally is different from that diagnosed postnatally. Up to 45% of those diagnosed antenatally may have associated heterotaxy syndromes
- ▶ Early postnatal diagnosis is important in planning timely surgical intervention
- ▶ A detailed transthoracic echocardiogram with Doppler is essential preoperatively to assess atrioventricular valve morphology and function and the relationship of the bridging leaflets to atrial and ventricular septum. Associated defects such as outflow tract obstruction or ventricular imbalance must be identified
- ▶ Surgical correction should attempt to minimise residual left atrioventricular valve regurgitation as this is the most common reason for reoperation and the most important cause of long term morbidity
- ▶ The operative mortality and outcome in AVSD is not significantly adversely affected in patients with Down's syndrome

Table 3 Long-term complications following surgical repair in AVSD

- ▶ Left atrioventricular valve regurgitation
- ▶ Left ventricular outflow tract obstruction
- ▶ Late onset complete heart block
- ▶ Pulmonary vascular disease
- ▶ Atrial or ventricular dysrhythmias
- ▶ Left atrioventricular valve stenosis
- ▶ Right atrioventricular valve stenosis/regurgitation
- ▶ Residual ventricular septal defect
- ▶ Aortic incompetence

Ostium primum ASD

The natural history of ostium primum ASD without surgery carries a 50% mortality before 20 years of age with atrial fibrillation an important cause of morbidity.^{w28} Following surgery the Mayo Clinic reports a 40 year survival of 76%.¹⁷ The reoperation rate in this large series was 11%. The most common cause for reoperation was left atrioventricular valve regurgitation followed by subaortic stenosis and complete heart block. Supraventricular dysrhythmias were observed in 16%. These increased with increasing age at primary operation. Bergin reports low mortality and good long-term survival in patients with ostium primum ASD presenting for surgical repair in later life (> 40 years old).²⁰

CONCLUSION

Early diagnosis of AVSD is important in order to institute appropriate medical and supportive treatment and to plan timely surgical intervention. In the current era the operative mortality is low with a good long term outlook for most patients, with or without Down's syndrome. As the most common cause for reoperation is left atrioventricular valve regurgitation, detailed attention should be given to atrioventricular valve repair at the primary operation with the aid of transoesophageal echocardiography. A minority of patients with associated defects such as ventricular hypoplasia will require alternative surgical strategies. Long term follow-up should focus on atrioventricular valve function, left ventricular outflow tract obstruction and dysrhythmia including late-onset heart block.

Additional references appear on the *Heart* website—<http://www.heartjnl.com/supplemental>

ACKNOWLEDGEMENTS

I am grateful to Dr F Casey for contributing the section on Fetal diagnosis. I am grateful to Professor R Anderson and G Price at the Institute for Child Health, UCL, for provision of figs 1–3, Mrs L Davidson for fig 5 and Dr A Sands for fig 6.

In compliance with EBAC/EACCME guidelines, all authors participating in *Education in Heart* have disclosed potential conflicts of interest that might cause a bias in the article

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