17

Intracardiac shunt malformations

Einat Birk and Norman H Silverman

Intracardiac malformations leading to a cardiac left-to-right shunt postnatally include atrial septal defects (ASD), atrioventricular septal defects (AVSD) and ventricular septal defects (VSD). These lesions comprise the largest group of cardiac defects detected during fetal life, the most common being VSD and AVSD.[1,2]

Defects of the atrial septum

Anatomy

An ASD is a common congenital defect seen in children, occurring in 1 in 1500 live births.[3] It can present as an isolated defect or in association with complex congenital heart defects. Several mechanisms cause the formation of an atrial communication, leading to several defect types (Figure 17.1):

- Secundum ASD
- Primum ASD (also named AVSD – partial or transitional type)
- Sinus venosus ASD (superior and inferior types)
- Coronary sinus ASD

Secundum ASD is the most common atrial communication in children. It occurs when the septum primum fails to cover the oval fossa, which is patent and allows right-to-left flow during fetal life. This failure of the septum primum can result in a single defect or fenestrated defect, as well as a wide range of defect sizes.

A primum ASD involves the lower part of the atrial septum and is part of the AVSD spectrum, which will be discussed separately.

A sinus venosus ASD is located in the posterosuperior or posteroinferior portion of the atrial septum. The superior defect is the more common, lying at the junction of the superior vena cava, right upper pulmonary vein and

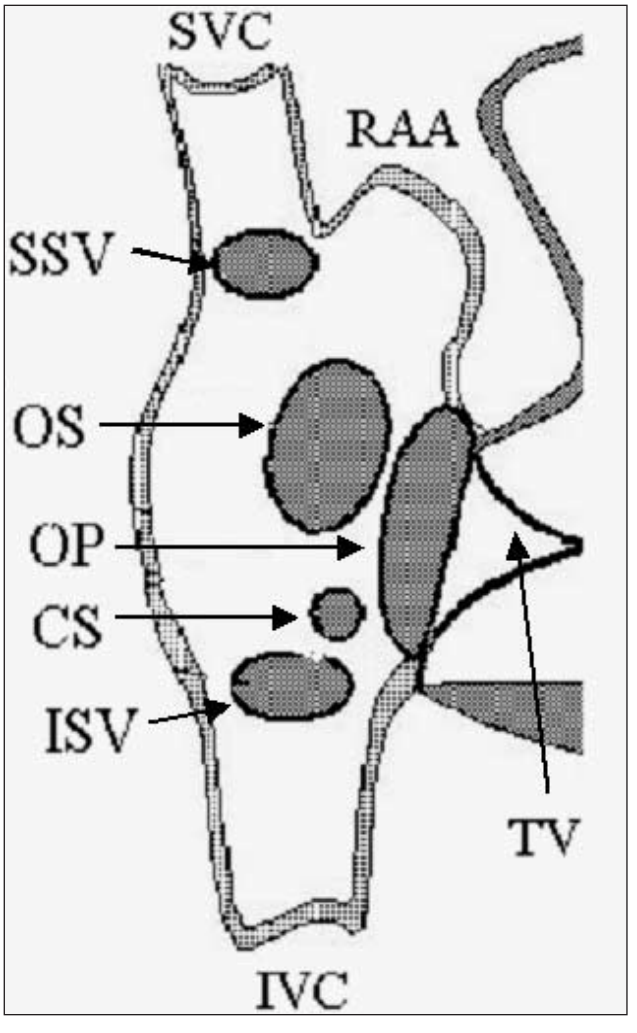


Figure 17.1
This drawing shows the different types of interatrial septal communications as seen from the right aspect of the atrial septum. CS, coronary sinus; IVC, inferior vena cava; OP, ostium primum; OS, ostium secundum; RA, right atrium; RAA, right atrial appendage; SV, sinus venosus; SVC, superior vena cava.

atrial septum. The superior vena cava appears to override the defect, which tends to be large. The inferior defect occurs at the junction of the inferior vena cava and the atrial septum. It is a less common defect and also tends to be of significant size. These defects result from a developmental malformation in the sinus venosus or from a primary failure in the partitioning of the true embryonic septum secundum.[4]

A coronary sinus ASD is a rare defect believed to occur when the atriosinus venosus fold fails to form. Therefore, instead of the normal draining of the coronary sinus into the right atrium via its usual orifice, there is a persistence of the wide communication between the sinus venosus and both atria.[4] Persistent left superior vena cava terminating in the left atrium is almost always present. Unlike a large coronary sinus receiving a persistent left superior vena cava that eventually drains into the right atrium, this defect allows a communication between the two atria, and the left superior vena cava drains directly into the left atrium.

During fetal life, the normal atrial communication at the oval fossa allows right-to-left atrial flow, allowing oxygen-rich blood to flow to the left heart and, consequently, to the brain and the heart. After birth, following the normal decrease in pulmonary pressure and resistance, and immediate increase of systemic vascular resistance caused by the loss of the placenta, the foramen should close and prevent intra-atrial shunting. The foraminal mechanism has been found to be substantially less efficient than was previously believed, since the advent of modern postnatal cardiac ultrasound has shown that a substantial number of infants under 6 months of age have intra-atrial left-to-right shunts. This is due to incomplete closure of the fossa ovalis by the septum primum.

When the septum primum is deficient, the right atrial and ventricular pressure decreases gradually as the compliance increases, leading to a predominant left-to-right shunting across the atrial communication. The amount of left-to-right shunting is determined by the size of the atrial defect and by the relative atrial and ventricular diastolic pressure differences.

Fetal diagnosis

Ostium secundum atrial septal defect

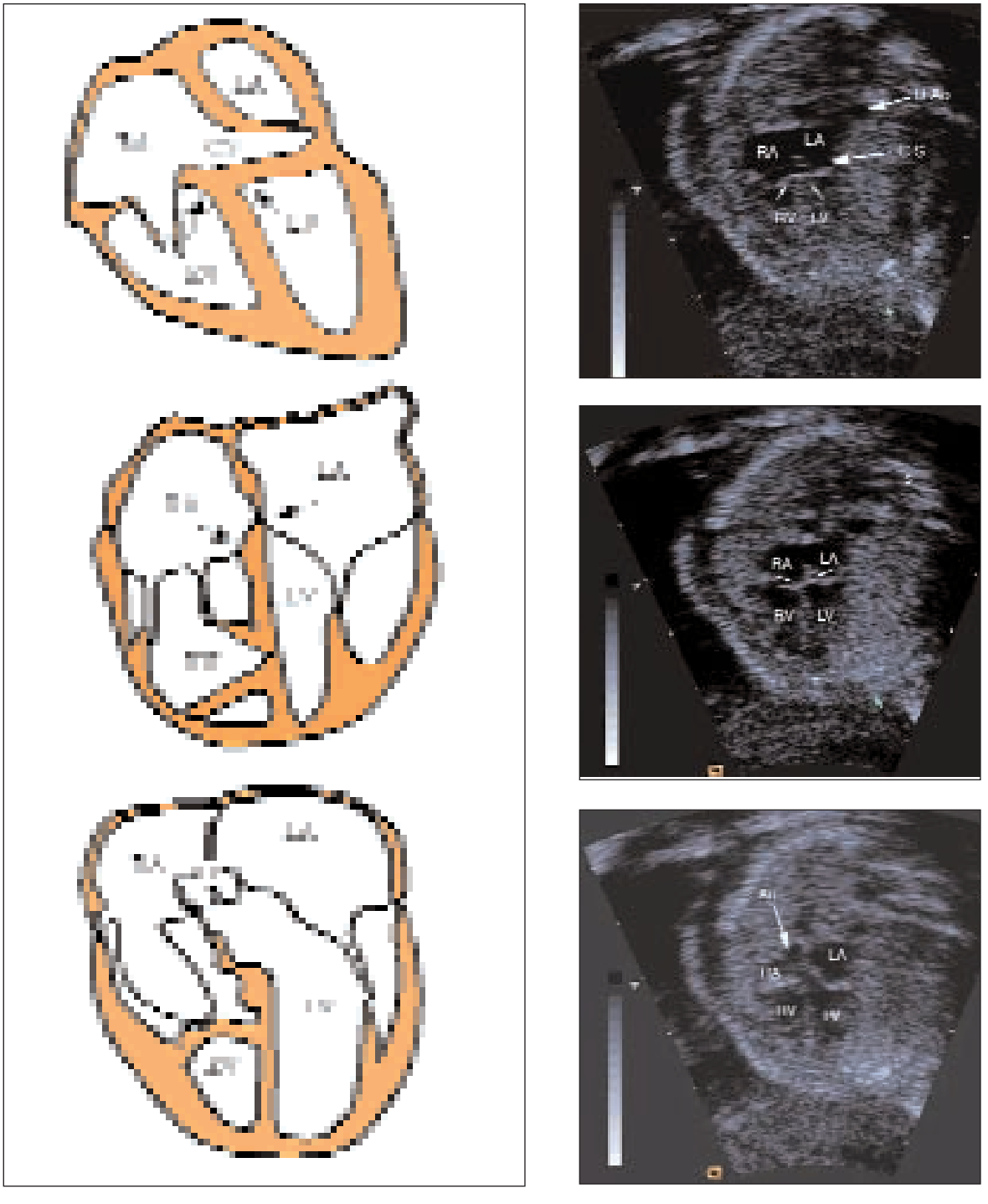
During fetal life, there is normally a communication between the right and left atria, which is located at the secundum septum, namely the foramen ovale. Relatively oxygenated blood streams from the ductus venosus via the inferior vena cava and into the left atrium by diverting the septum primum flap, which lies on the left side on the septum. This flap of atrial tissue is pushed open during most of the cardiac cycle. The foramen ovale lies in the middle third of the atrial septum and grows with gestation. The normal foramen size is similar to the aortic diameter[5] and grows from approximately 3 mm at 20 weeks of gestation to 8 mm at term.[6]

After birth, placental flow is eliminated and two processes occur: pulmonary venous flow increases and left atrial pressure rises above right atrial pressure. The flap of the septum primum is then pushed against the foramen ovale, leading to a functional closure of the atrial communication. Only when this flap valve mechanism fails to close the foramen ovale, is the communication termed a secundum defect. Since the size of the fetal foramen ovale and the primum flap varies widely in the normal fetus, it is impossible to predict the failure of the closure process during fetal life. Therefore, in contrast with postnatal life, when a secundum ASD is the commonest atrial defect, the fetal diagnosis of such an ASD possesses inherent difficulty and is rarely possible.[1]

Ostium secundum ASD can be a part of many forms of complex congenital heart defects, from anomalies of pulmonary venous return through to coarctation of the aorta. It is an essential part of defects such as tricuspid atresia. As in an isolated defect, it is rarely associated with extracardiac anomalies or genetic disorders.

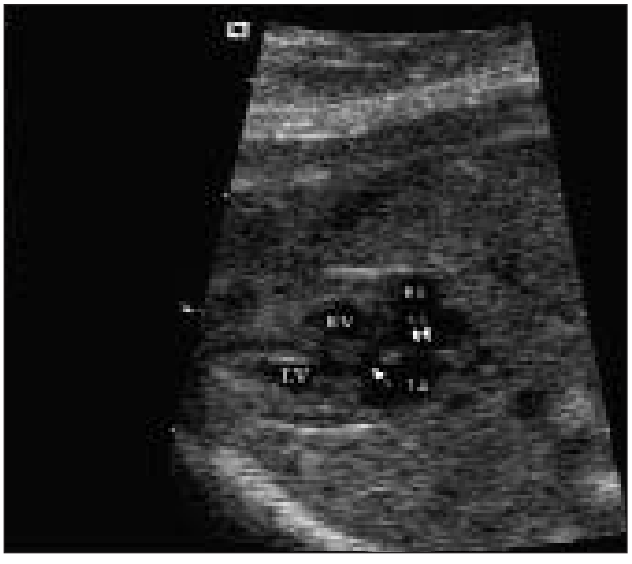
Ostium primum atrial septal defect (partial atrioventricular septal defect)

One of the most commonly diagnosed atrial communications in utero is the primum ASD.[1] This lesion is a form of common atrioventricular canal defect without a ventricular component. In this defect, the lower portion of the atrial septal fusion to the underlying atrioventricular valve junction is absent; both atrioventricular valves are attached to the crest of the ventricular septum and lose their normal differential appearance. The left atrioventricular valve is referred to as being 'cleft', showing a commissure between the primitive anterosuperior and posteroinferior bridging leaflets. Mild degrees of valvar insufficiency from this site can be detected in some fetuses. The four-chamber view is a useful plane in detecting this lesion. With this plane, a series of scans should be looked at in a sequential fashion in order to define appropriate morphological information (Figure 17.2). The posterior cross-sectional cut reveals the coronary sinus emptying into the right atrium just above the tricuspid valve. This view may lead to the impression that both atrioventricular valves insert into the ventricular septum at the same level. However, a more anterior coronal cut will show that both atrioventricular valves open and close with their normal differential insertion. This cut is also ideal for color Doppler interrogation of both valves. A more anterior angulation will reveal the left ventricular

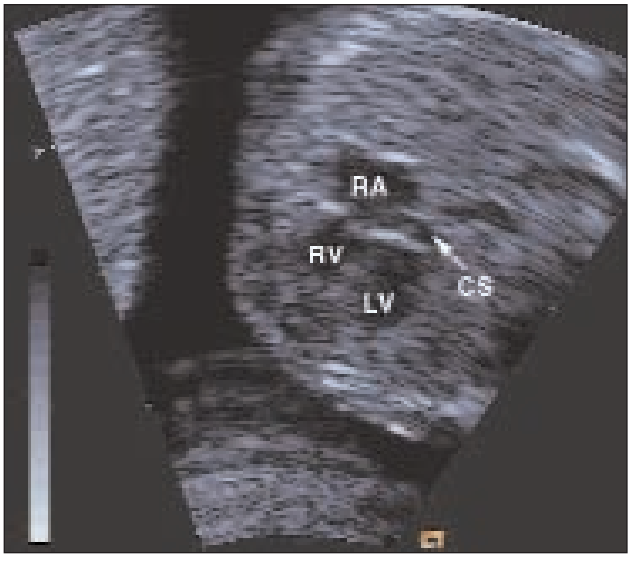


a b

Figure 17.2
(a) These drawings show a series of four-chamber cuts as scanned from the back to the front of the heart. (i) Posterior cross-sectional cut reveals the coronary sinus (CS) emptying into the right atrium (RA) just above the tricuspid valve. This view may lead to a false impression that both atrioventricular valves insert into the ventricular septum at the same level (arrows). (ii) A more anterior conventional four-chamber cut will show that both atrioventricular valves open and close with their normal differential insertion (arrows). This cut is also ideal for color Doppler interrogation of both valves. (iii) A more anterior cut will reveal the left ventricular (LV) outflow tract including the aortic valve (AO) and ascending aorta. The coronal cut where both atrioventricular valves open is probably the most useful in the diagnosis of a primum atrial septal defect. (b) (i–iii) This series of ultrasonic cuts in the four-chamber plane corresponds to the series shown in Figure 17.1a. D Ao, descending aorta.



a



b

Figure 17.3
(a) The heart is imaged in the four-chamber view showing both atrioventricular valves inserting into the ventricular septum at the same level. The single arrow points to the missing lower atrial septum, indicating the presence of a primum atrial septal defect. The double arrow points to the foramen ovale, the site of a normal fetal atrial communication. (b) This frame is a four-chamber cut showing the enlarged coronary sinus (CS, arrow) entering the right atrium (RA), which reflects the increased flow from a persistent left superior vena cava entering the upper portion of the coronary sinus. The coronary sinus enlargement may give the false impression of an ostium primum type of interatrial communication.

outflow tract including the aortic valve and ascending aorta. The coronal cut where both atrioventricular valves open is probably the most useful in the diagnosis of a primum ASD (Figure 17.3a). In this view the lower atrial septum is missing and both atrioventricular valves insert into the ventricular septum at the same level. A mild degree of left atrioventricular valve insufficiency may be detected as well as occasional right atrioventricular valve insufficiency. The more posterior cut, where the coronary sinus is displayed together with the tricuspid valve, can lead to the false impression that both atrioventricular valves are at the same level, leading to the erroneous diagnosis of a primum ASD. This error is even more likely to occur when the coronary sinus is enlarged (Figure 17.3b). This is the case when a persistent left superior vena cava drains into the coronary sinus; however, this vessel can usually be detected from other views. The cleft in the left atrioventricular valve is best seen in a cross-section from the parasternal or subcostal short-axis views.

When a primum ASD is detected, a complete sequential analysis of the heart is mandatory. Primum ASD is associated with situs anomalies such as right or left atrial isomerism, when the atrial septum tends to be small, leading to the appearance of a common atrium. Left heart hypoplasia, subaortic narrowing and coarctation of the aorta are known associated anomalies. A secundum ASD is a common associated finding. Primum ASD is associated with extracardiac anomalies, the most common being trisomy 21. It has also been rarely associated with Di George syndrome and Ellis–Van Creveld syndrome. Karyotyping should be performed when the diagnosis of primum ASD is made.

Sinus venosus and coronary sinus atrial septal defect

To the best of our knowledge, both sinus venosus and coronary sinus ASDs have not yet been reported in the fetus. These lesions are rarely associated with cardiac, extracardiac or chromosomal anomalies.

Natural history and outcome

Small secundum ASDs usually close spontaneously during the first 2 years of life. Defects persisting beyond 2 years of age tend to stay open and lead to a left-to-right atrial shunting of variable amount.

Primum ASDs (or partial AVSDs) do not close spontaneously and usually lead to a significant left-to-right shunting as well as the risk of pulmonary hypertension and pulmonary vascular disease. Some will develop substantial insufficiency of the left atrioventricular valve.

Sinus venosus ASDs never close spontaneously and almost always have a large concomitant left-to-right atrial shunt.

Even patients with a large left-to-right atrial shunt may be asymptomatic for many years. Some will develop right ventricular dysfunction and atrial arrhythmias during late adult life. A serious but rare complication is the development of secondary pulmonary hypertension and pulmonary artery thrombosis. All complications can be prevented by closing the defect. For most secundum defects, transcatheter closure has become available in many centers. Larger defects as well as all primum and sinus venosus atrial septal defects have to be closed surgically. The closure of a primum ASD involves repair of the left atrioventricular valve. Some of these patients will eventually need additional surgery for left atrioventricular valve repair or replacement. A smaller group will require further surgery to alleviate progressive obstruction of the left ventricular outflow tract. While life quality and expectancy after secundum ASD repair during childhood are similar to those of the general population,[7] patients following primum ASD repair have somewhat poorer results. Approximately 10% will need repeated surgery and life expectancy is shorter than that of the normal population.[8] When a primum defect is associated with left heart anomalies such as hypoplasia of the left atrioventricular valve, hypoplasia of the left ventricle, subaortic obstruction or coarctation of the aorta, the overall prognosis is guarded. In rare cases the left heart hypoplasia will not allow a biventricular repair, leading to palliative solutions such as the Fontan-type repair.

Restrictive foramen ovale

As discussed previously, the normal fetal foramen ovale has a wide range of what is considered to be normal size. The normal flow across it is of low velocity, ranging between 20 and 40 cm/s on pulsed Doppler.[6] Restrictive flow across it corresponds to an increase in flow velocity, usually above 100 cm/s. Most case reports of a restrictive flow across the foramen oval are associated with various forms of hypoplasia of the left heart.[9] In these cases the expected increase in left atrial pressure results in the opposition of the primum septum to the atrial septum and therefore a decrease in the flap valve diameter. Some authors believe that a restrictive foramen may lead to the development of hypoplastic left heart, since foraminal flow provides most flow into the left ventricle.

Complete atrioventricular septal defect

Anatomy

A complete AVSD is one of the more common cardiac defects detected prenatally.[1,10,11] This lesion is also known by the terms endocardial cushion defect or atrioventricular canal defect. In this lesion the atrial and ventricular septation is not complete and the separation between mitral and tricuspid orifices does not occur. Instead, there is a common atrioventricular junction. This lesion can be found as a spectrum of anomalies, ranging from a complete form (when both atrial and ventricular septation is incomplete, leading to a communication at both atrial and ventricular level) to a partial, or incomplete, form (when only the atrial or ventricular communication persists). All forms involve an intrinsic abnormality of the atrioventricular valves.

In most cases of AVSD, the atrioventricular junction is connected to the right and left ventricles so that the blood flows relatively evenly into each ventricle. This relationship is also described as a balanced AVSD. When the atrioventricular junction is predominantly connected to one of the ventricles, there is usually hypoplasia of the ventricle receiving the smaller portion of the atrioventricular orifice. This relationship is also described as an unbalanced AVSD, and right or left dominance can be identified.

Fetal diagnosis

The goals of the fetal cardiac ultrasound examination are:

1. Identify the presence and extent of the AVSD
2. Assess the relationship of the atrioventricular junction to the underlying ventricles
3. Assess the size of both ventricles
4. Assess the degree of atrioventricular valve regurgitation
5. Identify associated anomalies

The apical four-chamber view is the most commonly used cut to identify an AVSD (Figure 17.4). In the normal heart, the tricuspid septal leaflet is attached to the ventricular septum, while the mitral valve has no septal attachment and inserts into the crux of the heart at a slightly more cranial position. In an AVSD the left atrioventricular valve is attached to the ventricular septum at the same level as is the right atrioventricular valve. Therefore, both atrioventricular valves are at the same level, losing the normal differential insertion. The primum septum that

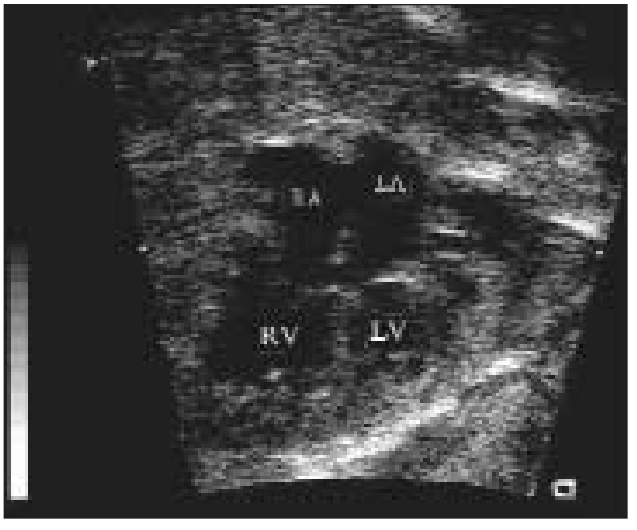


Figure 17.4
The heart is imaged in the apical equivalent four-chamber view, showing a typical picture of an atrioventricular septal defect. Both atrioventricular valves insert into the ventricular septum at the same level. Atrial and ventricular septal defects are seen above and below this insertion.

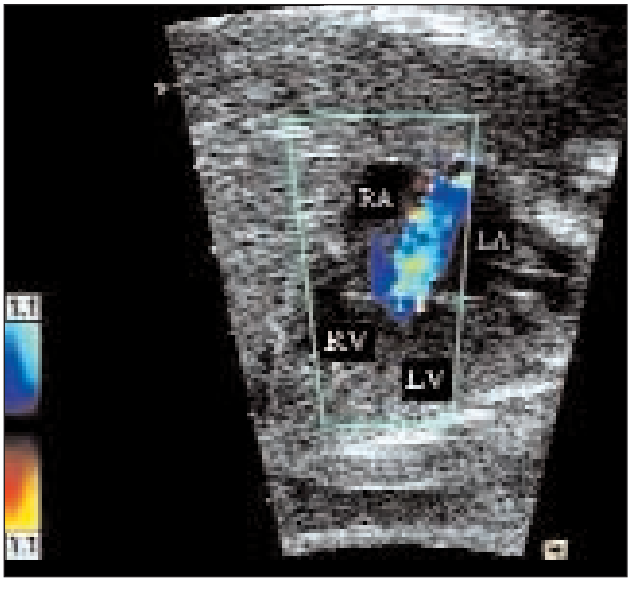


Figure 17.5
Color Doppler interrogation of the atrioventricular valves demonstrate a systolic jet of valve insufficiency, as shown in this four-chamber cut.

can be easily identified in the normal four-chamber view is absent. A ventricular communication can usually be identified in this view; in most cases this defect is large, although smaller defects can exist and are usually more difficult to identify. The apical four-chamber view is ideal for assessing the relationship of the atrioventricular junction to the underlying ventricles as well as the size of both ventricles. In this plane the ventricles should be of similar size. The atrial septum can be malaligned with the ventricular septum: when the atrial septum is deviated to the left, the right atrium drains to both ventricles (also described as double-outlet right atrium). When the atrial septum is shifted to the right, the left atrium drains to both ventricles. Such an anomaly can be corrected by surgery. A less favorable variation is when one ventricle is significantly smaller than the other. This is also known as an unbalanced AVSD and in extreme situations will lead to a single ventricle solution. The apical four-chamber view is also ideal for flow interrogation of the atrioventricular valves. Insufficiency of both atrioventricular valves, which is common in the newborn with an AVSD, is less commonly seen during fetal life (Figure 17.5). The short-axis views obtained from the parasternal or subcostal equivalent angles provide a detailed picture of the atrioventricular valve anatomy. In rare cases the insertions of the right or left atrioventricular valves is on the other side of the ventricular septum. These are more difficult to repair and may lead to a worse prognosis. To the best of our knowledge, this has not yet been reported in the fetus. As in the case of primum ASD, the presence of a large coronary sinus may be misdiagnosed as an AVSD.[12] The coronary sinus lies behind the left atrium and is usually enlarged by additional flow from a persistent left superior vena cava draining into the coronary sinus. A posterior coronal four-chamber view can create the illusion of both tricuspid and mitral valves inserting into the ventricular septum at the same level. However, a more anterior cut (Figure 17.2) will reveal the real relationship between the valve insertions where they can be demonstrated in both open and closed position. The normal offset can then be demonstrated, avoiding the false diagnosis of an AVSD. One should keep in mind that persistent left superior vena cava and dilated coronary sinus can coexist with an AVSD.

Associated lesions

When an AVSD is detected, a complete sequential analysis of the heart is mandatory. An AVSD is associated with situs anomalies such as right and left isomerism. Tetralogy of Fallot and double-outlet right ventricle are well-known associated lesions, more common in fetuses with trisomy 21. Left heart hypoplasia, subaortic narrowing and coarctation of the aorta are all known associated cardiac anomalies, usually in fetuses with normal chromosomes.

Another common association is a secundum ASD. The most common extracardiac anomaly associated with an AVSD is trisomy 21. An AVSD is also associated with other chromosomal anomalies such as trisomies 18 and 13. The fetal karyotype, therefore, should be examined whenever this diagnosis is made. It can also be a part of other syndromes such as Ellis–Van Creveld, VACTRL, CHARGE, Cornelia de Lange and Goldenhar syndromes.[13] In a recent study,[14] out of 301 fetuses with an AVSD, only 51% had isolated AVSD. Right isomerism occurred in 12% and left isomerism in 20%. Extracardiac abnormalities and nonkaryotypic syndromes were evident in 13%; 39% had trisomy 21 and 10% had other chromosomal abnormalities. Similar findings were found by other groups.[15,16]

Natural history and outcome

During fetal life AVSD is usually well tolerated, and most fetuses reach term and are delivered according to routine obstetric practice. A few will develop congestive heart failure and nonimmune hydrops because of severe insufficiency of the atrioventricular valve or heart block, especially in left atrial isomerism.[14,17,18] In those cases the odds for fetal or neonatal demise are high. According to a recent study, 15% of fetuses diagnosed with this lesion whose parents opted to continue with the pregnancy died in utero.[14]

The infant with an isolated AVSD will remain asymptomatic for a few weeks. Most will develop signs of congestive heart failure during the first 4–8 weeks of life as pulmonary vascular resistance falls. All will require surgical repair, which is usually carried out during the first 6 months of life. Survival after AVSD repair is high, exceeding 90%, although some patients (especially the group with normal chromosomes) will require additional surgery because of the development of left atrioventricular valve insufficiency or narrowing of the left ventricular outflow tract. The odds for a successful repair decline when additional cardiac anomalies are present. Again, the chance of survival to 3 years is significantly lower in the group followed since intrauterine life when compared to the surgical literature, and is quoted as low as 38%.[14]

Ventricular septal defect

Anatomy

VSD is the most common congenital heart defect diagnosed during the first year of life.[19,20] VSD is also a common cardiac defect detected prenatally, at a rate lower that that of prenatal detection of an AVSD.[1,2]

The ventricular septum is arbitrarily divided into four sections: the inlet, membranous, trabecular and outlet components, which have different embryological origins.

When viewed from the right ventricle, the inlet septum has a lightly trabecular surface and is bounded by the tricuspid annulus and the attachments of the papillary muscles to the ventricular septum.

The membranous septum is a relatively small segment lying beneath the septal leaflet of the tricuspid valve and adjacent to the aortic and mitral valves. Viewed from the left ventricle, the membranous septum lies adjacent to the right fibrous trigone just beneath the aortic valve. The membranous septum is a thin translucent structure and therefore cannot be well imaged in all planes, and is often difficult to image even after birth. This may result in the false impression of a VSD in some views, especially in the fetus.

The trabecular septum, which derives its name from its heavily trabeculated appearance, extends from the inlet septum to the region of the outlet septum just proximal to the pulmonary valve, and does not lie in a single plane. It contains the moderator band (or septomarginal trabeculation, which is also called the septal band of the crista supraventricularis), which extends in a Y-shaped fashion below the pulmonary valve. Its anterior portion abuts the outlet septum, while its posterior portion is the papillary muscle of the conus (muscle of Lancisi). Inferiorly, the septomarginal trabeculation extends as a broad muscle bundle. This portion of the muscle, the septomarginal trabeculation, is also referred to as the septal limb of the crista supraventricularis. The ventricular infundibular fold, which is the right ventricular muscle lying between the tricuspid and the pulmonary valves, is also called the parietal band of the crista supraventricularis. The outlet septum (also named conus septum) is a small segment extending from the septomarginal trabecula to the pulmonary valve.

VSDs can occur in any of the septal locations, but also occur at the sites of fusion between them; for example, defects found around the membranous septum are termed perimembranous. They can also be named for their area of extension, such as perimembranous-inlet, perimembranous-trabecular and perimembranous-outlet defects. Perimembranous defects comprise about 75% of all VSDs.[21] When the defects are surrounded entirely by muscle, they are termed muscular-inlet, muscular-trabecular and muscular-outlet defects. Muscular defects comprise 10–15% of all ventricular septal defects. Defects of the outlet septum most commonly are adjacent to the pulmonary and aortic valves and are termed subarterial doubly committed or supracristal defects. Such defects comprise about 5% of the VSDs but are more common in Asian populations.[21]

When the different portions of the septum adjacent to the VSD are malaligned, they are termed a malalignment

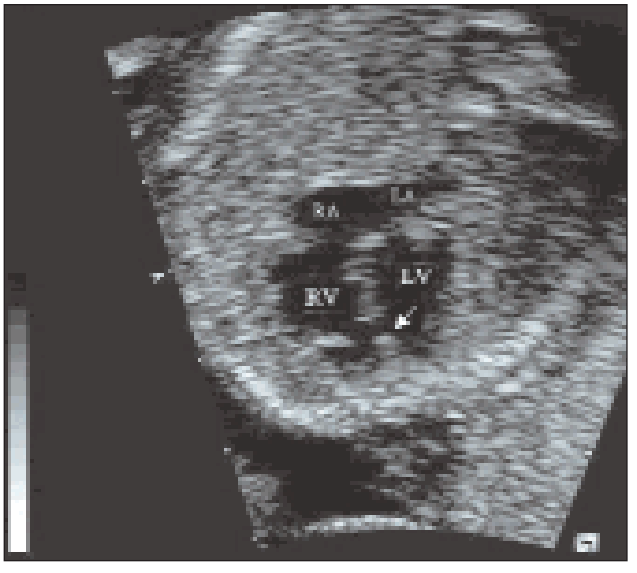


Figure 17.6
The ventricular septum of a 20-week fetus is visualized in a four-chamber view. A muscular septal defect is identified at the lower portion of the septum (arrow). Note the "T" artifact at the defect edges.

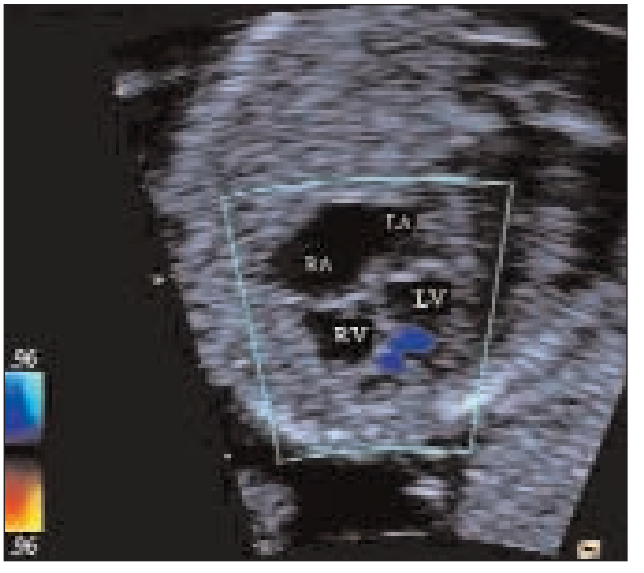


Figure 17.7
Flow across this muscular ventricular septal defect can be detected using color flow mapping.

VSD. The VSD can be malaligned between the outlet and trabecular septum or with respect to the atrioventricular valves, associated with straddling of an atrioventricular valve.

The size of ventricular septal defects varies from very small to large, involving a third or more of the ventricular septum. Since the septum does not lie in a single plane, it can be difficult to assess the VSD size. Defects may be isolated or multiple, and are commonly a part of, or associated with, other cardiac lesions.

Fetal diagnosis

The goals of the ultrasound examination are:

1. Identify the presence of a VSD
2. Define which segment of the septum is involved
3. Identify additional anomalies

The fetal ventricular septum is easily seen in the four-chamber view, which can be visualized from an apical equivalent or from a lateral orientation. In the apical equivalent four-chamber view the ultrasound beam is parallel to the ventricular septum using lateral resolution (Figure 17.6). Since the membranous part of the septum is thin, the ventricular septum 'disappears' toward the internal crux of the heart. This may result in the appearance of a dropout in the septum, leading to a false diagnosis of a VSD.[22] In order to achieve a different angle, the transducer can be moved to a different location on the maternal abdomen so that the ultrasound beam will be perpendicular to the ventricular septum, using axial resolution. The parasternal short-axis equivalent cut is also useful, when the ultrasound beam is perpendicular to the ventricular septum; the different parts of the septum can be visualized in great detail. A useful physical sign of a septal defect is the so-called "T" artifact: high impedance exists at the blood–tissue interface producing ballooning of echoes at the rim of a defect, creating bright spots at the defect edges.[22] The "T" artifact is not created when the septum is thin, leading to a simple dropout. The use of color flow Doppler imaging may augment the ability to identify a VSD in the fetus (Figures 17.7 and 17.8). Once again, when this is performed from the apical equivalent four-chamber view, the ultrasound beam is parallel to the ventricular septum so that the color tends to "cover" the thin part of the septum, leading to the impression of flow across a VSD. However, when the ultrasound beam is perpendicular to the ventricular septum, color Doppler flow can be detected more accurately when flow is crossing the septum, usually in a bidirectional fashion.[23,24] Since the pressure is similar in the fetal right and left ventricles, the potential pressure gradient across a VSD is small. Color flow velocity should

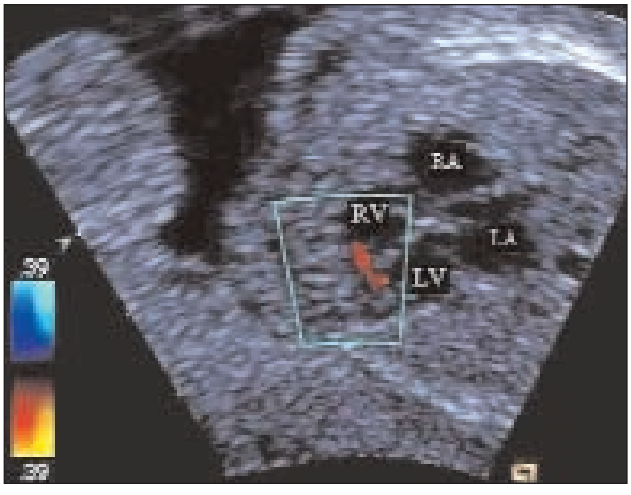


Figure 17.8
The same ventricular septal defect as in Figure 17.7 can be shown by color flow Doppler when the ventricular septum is imaged with the ultrasound beam perpendicular to the defect.

therefore be reduced to a low Niquest limit in order to detect low-velocity jets.

Associated lesions

When a VSD is detected, a complete sequential analysis of the heart is mandatory. An isolated VSD is rarely associated with situs anomalies. However, it is commonly found as part of complex cardiac lesions, some of which are not obvious when the study is performed during early pregnancy. When a VSD is identified, both right and left ventricular outflow tracts should be examined in detail. Since a VSD can be a part of tetralogy of Fallot, the size of the right ventricular outflow tract, main and branch pulmonary arteries, as well as the flow across these structures should be examined. The usual malalignment of the outflow septum in tetralogy of Fallot is not as easy to identify during fetal life as it is in postnatal life. This diagnosis should be suspected whenever the pulmonary artery is smaller than expected or a pressure gradient is deducted across the right ventricular outflow tract. The associations of other lesions with ventricular septal defects are numerous, and include left heart obstructions such as subaortic narrowing, aortic valve stenosis, coarctation of the aorta and interrupted aortic arch. Since both right and left outflow obstructions can evolve during pregnancy and postnatal life, reassessment during late gestation is advised. A VSD can also be a part of complex lesions such as transposition of the great arteries and double-outlet right ventricle.

Extracardiac anomalies associated with a VSD include chromosomal anomaly in over 40% according to some series.[1,2] Such anomalies included trisomies 21, 13 and 18. This rate is significantly higher than expected from postnatal series, and may relate to the selection of patients referred for fetal echocardiography as well as to spontaneous fetal loss in chromosomally abnormal fetuses that would not be included in postnatal series. Other extracardiac anomalies associated with a VSD include a deletion in 22q11 and non-chromosomal multiply malformed fetuses.[2]

Natural history and outcome

During fetal life, a VSD is well tolerated and most fetuses reach term and are delivered according to routine obstetric practice. The odds for fetal demise are higher when extracardiac anomalies are present. Isolated perimembranous and muscular ventricular septal defects detected and followed through pregnancy show a high rate of spontaneous closure.[25] No correlation was found between the size of the defect prenatally and its chance for spontaneous closure.[25] We have data to show that smaller VSDs have a greater chance of closure than larger defects (postnatally). Infants with an isolated VSD may remain asymptomatic for a few weeks; those with relatively large defects will develop signs of congestive heart failure after a few weeks of life as pulmonary vascular resistance falls. Over 50% of VSDs located in the perimembranous or muscular septum will close spontaneously, usually during the first year of life; only a minority of such defects will require surgical repair. Defects in the inlet or outlet septum do not close spontaneously and require surgical repair. Surgery is usually carried out during the first year of life and, when isolated, carries a low mortality and complication rate.[26] Multiple defects or a single large apical muscular defect carry a higher risk of surgical repair due to limited access. Such infants may need the placement of pulmonary arterial banding to decrease pulmonary flow and pressure during the first months of life, followed by surgical or transcatheter closure of the defects coupled with pulmonary debanding later in life. When associated cardiac anomalies are present, the odds for a successful repair depend on the severity and nature of the additional cardiac defects.

References

1. Allan LD, Sharland GK, Milburn A, et al. Prospective diagnosis of 1,006 consecutive cases of congenital heart disease in the fetus. J Am Coll Cardiol 1994;23:1452–1458.

2. Stoll C, Garne E, Clementi M, Euroscan study group. Evaluation of prenatal diagnosis of associated congenital heart disease by fetal ultrasonographic examination in Europe. Prenat Diagn 2001;21:243–252.

3. Fyler DC. Report of the New England Regional Infant Cardiac Program. Pediatrics 1980;65:375–461.

4. Goor DA, Lillehei CW. Congenital Malformations of the Heart. New York: Grune & Stratton, 1975.

5. Wilson AD, Rao PS, Aeschlimann S. Normal fetal foramen flap and transatrial Doppler velocity pattern. J Am Soc Echocardiogr 1990;3:491–494.

6. Phillipos EZ, Robertson MA, Still KD. The echocardiographic assessment of the human fetal foramen ovale. J Am Soc Echocardiogr 1994;7:257–263.

7. Nieminen HP, Jokinen EV, Sairanen HI. Late results of pediatric cardiac surgery in Finland: a population-based study with 96% follow-up. Circulation 2001;104:570–575.

8. El-Najdawi EK, Driscoll DJ, Puga FJ, et al. Operation for partial atrioventricular septal defect: a forty year review. J Thorac Cardiovasc Surg 2000;119:880–890.

9. Cohbot V, Hornberger LK, Hagen-Ansert S, Sahn DJ. Prenatal detection of restrictive foramen ovale. J Am Soc Echocardiogr 1990;3:15–19.

10. Allan LD. Atrioventricular septal defect in the fetus. Am J Obstet Gynecol 1999;181:1250–1253.

11. Silverman NH. Pediatric Echocardiography. Baltimore: Williams and Wilkins, 1993.

12. Park JR, Taylor DK, Skeels M, Towner DR. Dilated coronary sinus in the fetus misinterpretation as an atriventricular canal defect. Ultrasond Obstet Gynecol 1997;10:126–129.

13. Feldt RH, Porter CJ, Edwards WD, Puga FJ, Seward JB. Atrioventricular septal defects. In: Moss and Adams. Heart Disease in Infants, Children and Adolescents, 5th edn. Emmanouilides GC, Riemenschneider TA, Allan HD, Gutgesell HP, eds. Baltimore: Williams and Wilkins, 1995:704–724.

14. Huggon IC, Cook AC, Smeeton C, Magee AG, Sharland GK. Atrioventricular septal defects diagnosed in fetal life: associated cardiac and extra-cardiac abnormalities and outcome. J Am Coll Cardiol 2000;36:593–601.

15. Delisle MF, Sandor GG, Tessier F, Farquharson DF. Outcome of fetuses diagnosed with atrioventricular septal defects. Obstet Gynecol 1999;94:763–767.

16. Allan LD. Atrioventricular septal defect in the fetus. Am J Obstet Gynecol 1999;181:1250–1253.

17. Silverman NH, Kleinman CS, Rudolph AM et al. Fetal atrioventricular valve insufficiency associated with nonimmune hydrops: a two-dimensional echocardiographic and pulsed Doppler ultrasound study. Circulation 1985;72:825–832.

18. Schmidt KG, Ulmer HF, Silverman NH, Kleinman CS, Copel JA. Perinatal outcome of fetal complete atrioventricular block: a multicenter experience. J Am Coll Cardiol 1991;17:1360–1366.

19. Anderson RH, Macartney FJ, Shinebourne EA, Tynan M. Ventricular septal defects. In: Paediatric Cardiology. Anderson RH, ed. London, UK: McGraw-Hill, 1987:565–590.

20. Ferenez C, Rubin DJ, Loffredo AC, Magee AC, eds. Epidemiology of Congenital Heart Disease. The Baltimore–Washington Infant Study 1981–89. Perspectives in Pediatric Cardiology 4. Mount Kisco, NY: Futura Publishing, 1993:31–33.

21. Rudolph AM. Congenital disease of the heart: clinical–physiological considerations. Mount Kisko, NY: Futura Publishing, 2001:198–199.

22. Canale JM, Sahn DJ, Allen HD et al. Factors affecting real-time, cross-sectional echocardiographic imaging of perimembranous ventricular septal defects. Circulation 1981;63:689–697.

23. Lethor JP, Maron F, de Moor M, King MEE. Physiology of ventricular septal defect shunt flow in the fetus examined by color Doppler M-mode. Circulation 2000;101:e93.

24. Chao RC, Ho ESC, Hsieh KS. Fluctuations of interventricular shunting in a fetus with an isolated ventricular septal defect. Am Heart J 1994;127:955–958.

25. Paladini D, Palmieri S, Lamberti A, Teodoro A, Martinelli P, Nappi C. Characterization and natural history of ventricular septal defects in the fetus. Ultrasound Obstet Gynecol 2000;16:118–122.

26. Masuda M, Kado H, Kajihara N et al. Early and late results of total correction of congenital cardiac anomalies in infancy. Jpn J Thorac Cardiovasc Surg 2001;49:497–503.