

Septal Defects

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Abstract

Cardiac septal defects are the most common form of congenital heart disease if bicuspid aortic valve is excluded. This category includes atrial septal defect (ASD), atrioventricular septal defect, and ventricular septal defect (VSD). The different types of ASD are: primum, secundum, sinus venosus, and unroofed coronary sinus. The most common forms of atrioventricular septal defect are complete and partial whereas the four main groups of VSDs are inlet, muscular, membranous, and outlet. A good knowledge of the anatomy is necessary to classify adequately the ASDs and VSDs. Transthoracic echocardiography is the primary and most important imaging modality and MRI can be useful when echocardiography is not feasible or not diagnostic.

1 Introduction

Septal defects, in general, are the most common form of congenital heart disease. In the adult population, they represent most of the new congenital heart disease cases. The interventricular septum and the interatrial septum separate the pulmonary

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circulation and the systemic circulation. When either the interventricular or the interatrial septum is deficient, systemic and pulmonary circulation can become in contact and create a shunt lesion. There are several types of septal defects that will be described in this chapter. Usually, septal defects are classified in three types: atrial septal defects (ASD), ventricular septal defects (VSD), and atrioventricular septal defects (AVSD). The primary imaging modality for those kinds of pathologies remains the transthoracic echocardiography, but MRI and to a lesser degree CT can precise the diagnosis and its repercussion on the cardiac chambers. In the first section of this chapter, several types of ASD will be described, the most common being the ostium secundum type. Atrioventricular septal defects will be discussed in the ASD section since an ostium primum type ASD is an inherent part of this pathology. The different types of VSDs will be described and discussed in the second section.

2 Atrial Septal Defects

Atrial septal defects (ASD) are common congenital heart defects. The incidence has been estimated to be approximately 100 per 100.000 live births. They constitute 8–10% of congenital cardiac malformations in children (Hugh et al. 2016; Hoffman and Kaplan 2002; Wang et al. 2003; Botto et al. 2001).

The majority of ASDs occur sporadically with no identifiable cause. This anomaly can be isolated or occur in association with other congenital cardiovascular malformations. In some congenital heart defects, the ASD may even be crucial for survival of the patient, such as D-transposition of the great arteries, total anomalous pulmonary venous return (TAPVR), tricuspid atresia, and hypoplastic left heart syndrome (Hugh et al. 2016; Saremi 2014).

The vast majority of children with isolated ASD, even if a large left-to-right shunt is present, are asymptomatic and the defects are usually an incidental finding on imaging studies. Only 1% becomes symptomatic in the first year (Rajiah and Kanne 2010). In the second decade, patients

might still be asymptomatic or present with palpitations or shortness of breath during strenuous activities. By contrast, almost all adult patients with a large defect are symptomatic (Geva et al. 2014; Campbell 1970). Untreated large atrial septal defect is associated with a reduced life expectancy mainly because of the changes induced in the pulmonary vasculature bed and in the myocardium (Campbell 1970). In the first two decades, the annualized mortality rate is low (0.6% and 0.7%, respectively), then increases to 4.5% in the fourth decade to reach 7.5% in the sixth decade (Geva et al. 2014; Campbell 1970).

The evaluation of an interatrial septal defect usually begins with a transthoracic echocardiography (TTE). However, this method is operator dependent and can be limited by acoustic window (Rajiah and Kanne 2010). If the findings on echocardiography are uncertain, computed tomography angiography (CTA) and/or magnetic resonance imaging (MRI) are valuable tools for further evaluation. Both can be used to define the anatomy of an ASD, its impact on the cardiac chambers and associated anomalies (mainly the pulmonary venous return) (Sadler 2015; Hagen et al. 1984). However, cardiac MRI stays the cross-section modality of choice because in addition to provide a comprehensive assessment of the cardiac anatomy, it can accurately quantify shunts, ventricular size, as well as cardiac and valve functions. Computed tomography angiography (CTA) should be reserved for patients with known contraindications to MRI or when the ability to tolerate MRI is limited by claustrophobia. The role of chest radiographs is limited. In children with an isolated ASD, it is almost always normal. Enlargement of the right cardiac chambers and the pulmonary artery may be seen only with a hemodynamically significant ASD. Hemodynamically significant are generally the ASDs with a pulmonic-to-systemic flow ratio (QP/QS) > 1.5:1.

ASD can be found at several sites. Knowledge of the development of interatrial septum helps understanding the different types of ASD. So, the next sections review briefly the embryologic development of the interatrial and the different types of ASD. Then, the indications and options of treatment are presented.

2.1 Embryologic Development of the Interatrial Septum

The interatrial and interventricular septa are formed between the 27th and 37th days of development. At the beginning, a pulsatile tube is transformed into a hollow, single-chambered pump (Rajiah and Kanne 2010). Then, tissue masses, the so-called *endocardial cushions*, appear in the atrioventricular region, separate the heart into right and left upper and lower chambers, and take part in the formation of the interatrial septum, the inlet portion of the interventricular septum, and the atrioventricular valves (Rajiah and Kanne 2010; Sadler 2015).

From the roof of the common atrium arises a septum, the *septum primum*, and grows caudally toward the endocardial cushions (Sadler 2015). The orifice between the septum primum and the endocardial cushions is the *ostium primum*. During further growth of the septum primum, perforations in the upper portion appear, forming the secondary interatrial communication, the *ostium secundum*. Then a second fold begins to grow in the atrium, the *septum secundum*. By further growth, it overlaps the ostium secundum, but an oblique cleft between the two atria is maintained, the *foramen ovale* (Fig. 1).

The foramen ovale represents a normal interatrial communication that is present throughout fetal life. This normal fetal interatrial communication is

commonly encountered in the neonatal period. After birth, left atrial pressure normally exceeds right atrial pressure, leading to apposition and fusion of the septum primum and septum secundum (Saremi 2014; Hagen et al. 1984).

In order to complete the development, the endocardial cushions fused with the anterior and posterior walls of the heart chamber dividing the atrioventricular canal into the mitral and tricuspid inlets. The position of the tricuspid annulus is normally more apical than the mitral annulus (Rajiah and Kanne 2010).

2.2 Locations (Types) of ASD

Interatrial communications can be found at several sites (Fig. 2). Although they are often summarized as atrial septal defects (ASDs), they do not necessarily involve the interatrial septum. They are classified according to their location and the anatomical structure that is involved:

- Septum primum and atrioventricular septum → Ostium primum ASD (20%)
- Fossa ovalis → Ostium secundum ASD (70%)
- Sinus venosus (embryologic structure) → Sinus venosus defect (5–10%)
- Coronary sinus → Unroofed coronary sinus (1%)
- Foramen ovale → Patent foramen ovale

A proper classification is crucial for an optimal therapeutic decision-making.

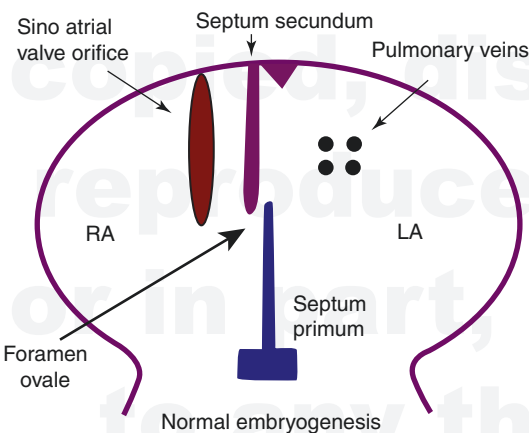


Fig. 1 Coronal view of the atrial septa. RA right atrium, LA left atrium

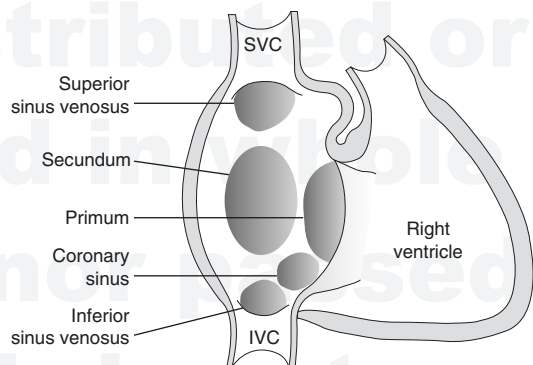


Fig. 2 View from the right atrium showing the different types of ASDs. IVC inferior vena cava, SVC superior vena cava. Reprinted with permission from Slovis et al. (2008)

2.2.1 Ostium Primum Defect and Atrioventricular Septum Defect (AVSD)

The ostium primum ASD is part of the spectrum of the atrioventricular septum defects (AVSD) or endocardial cushion defects. Therefore, we group them in the same section. The defect occurs in the region of endocardial cushions. The fusion of the endocardial cushions not only divides the embryonic atrioventricular canal into a right and a left orifice, but it is also crucial for the closure of the ostium primum and the formation of the interventricular septum. A failure of fusion of the endocardial cushions results in a persistent atrioventricular canal and a defect in the interatrial and interventricular septa. It also results in a common atrioventricular valve with one valve ring and five leaflets (Saremi 2014; Sadler 2015; Anderson et al. 1998).

A partial, a transitional, an intermediate, and a complete form are described (Fig. 3). The complete form includes an ostium primum ASD, a large ventricular septal defect of the inlet type, and a common AV valve with one single orifice consisting of the five leaflets (Fig. 4) (Anderson et al. 1998; Prasad et al. 2004). In the intermediate form, there is also common valve ring, but the free margins of the bridging leaflets are fused, creating two separate valve orifices. The resulting left atrioventricular valve has three leaflets, contrary to a normal mitral valve with only two leaflets. This abnormality of the mitral valve is also called “cleft mitral valve” (Fig. 5) (Van Praagh et al. 1994). The cleft mitral valve is usually regurgitant. With time, the valve becomes thickened and similar to mitral valve prolapse. In partial form, the mitral and tricuspid annuli are separate but always located at the

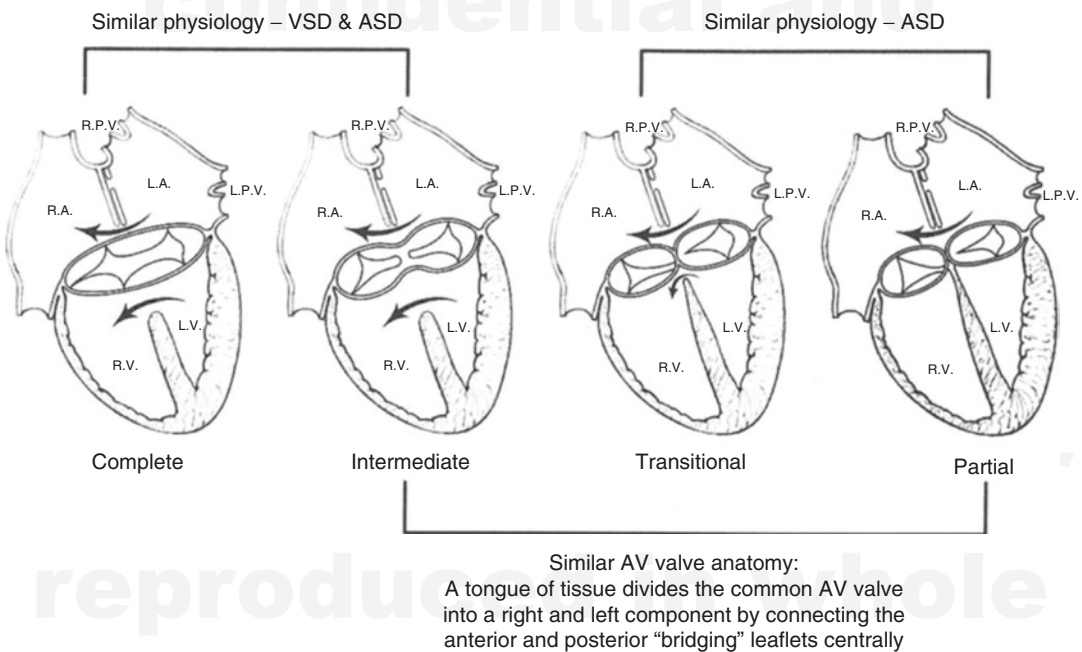


Fig. 3 Summary of AVSD. Physiologic and anatomic similarities between the different forms of atrioventricular septal defects are illustrated. *Reprinted with permission from Allen HD et al.*

- Complete AVSDs have one annulus with large interatrial and interventricular communications. Intermediate defects (one annulus, two orifices) are a subtype of complete AVSD.
- Complete AVSDs have physiology of ventricular septal defects (VSD) and atrial septal defects (ASD).
- Transitional AVSDs are a form of partial AVSD in which a small inlet VSD is also present.
- Partial defects and the intermediate form of complete AVSD share a similar anatomic feature: a tongue of tissue divides the common atrioventricular valve into distinct right and left orifices.
- Partial and transitional AVSDs have physiology of ASDs.
- LA left atrium, LPV left pulmonary vein, LV left ventricle, RA right atrium, RPV right pulmonary vein, RV right ventricle



Fig. 4 Complete atrioventricular septal defect. Four-chamber view image obtained from cine FLASH MRI sequence demonstrates a common atrioventricular valve, a large ostium primum ASD, and an inlet VSD

same level. A cleft mitral valve is present and the bridging leaflets fuse to the crest of the interventricular septum, leaving only an ostium primum atrial septal defect (Sadler 2015). Transitional AVSD is a subtype of partial AVSD. This term is used when a partial AVSD also has a small inlet VSD that is partially occluded by dense chordal attachments to the ventricular septum. Whatever the employed imaging modalities, this type of ASD are well demonstrated on the four-chamber view.

Atrioventricular septal defects constitute approximately 20% of ASDs and are particularly common in Down syndrome (Hugh et al. 2016). In fact, it accounts for 40% of cardiac defects in patients with Down syndrome. Other associated syndromes are DiGeorge syndrome and Ellis-Van Creveld syndrome. It may also be associated with a persistent left SVC and a secundum ASD (Webb and Gatzoulis 2006). Ostium primum defects are usually larger than ostium secundum defects and are symptomatic at a younger age (Rajiah and Kanne 2010).

2.2.2 Ostium Secundum Defect

This atrial septal defect occurs in the fossa ovalis, the central part of the atrial septum as a result of either excessive resorption of the septum pri-

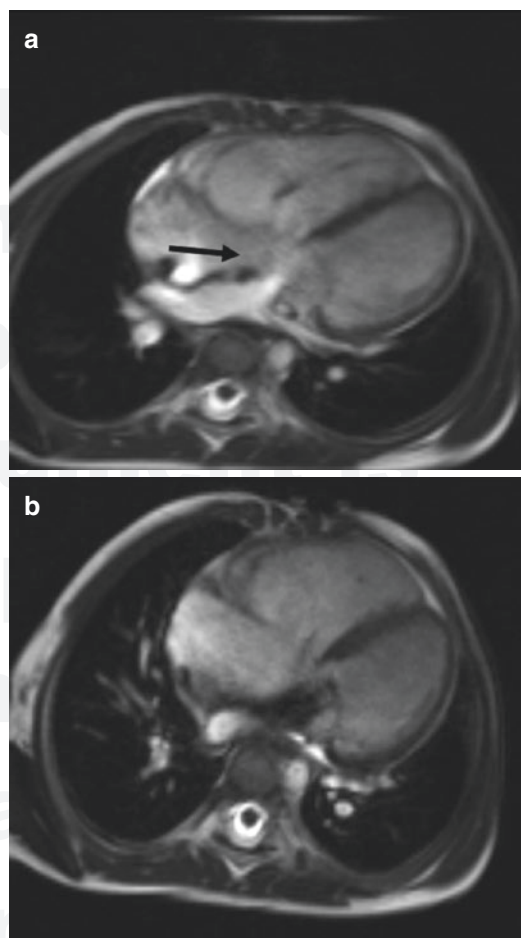


Fig. 5 Partial atrioventricular septal defect. (a), (b) Four-chamber view images obtained from cine FLASH MRI sequence demonstrate mitral and tricuspid valves at the same level due to the common ring, ostium primum ASD (black arrow) and mitral regurgitation

um or deficient growth of septum secundum (Figs. 6 and 7) (Prasad et al. 2004). In case of excessive resorption of the septum primum, the defect is central with one or multiple holes. In case of abnormal development of the septum secundum, the defect is superiorly located and larger (Blom et al. 2005). The size of ostium secundum defects varies from several millimeters to 2–3 cm (Geva et al. 2014).

Secundum ASDs are by far the most common ASDs, accounting for almost 70% of all ASDs. Associations with several syndromes have been described, such as Down, Holt-Oram, Klinefelter, Ellis-van Creveld, Noonan, Treacher-Collins, and others.

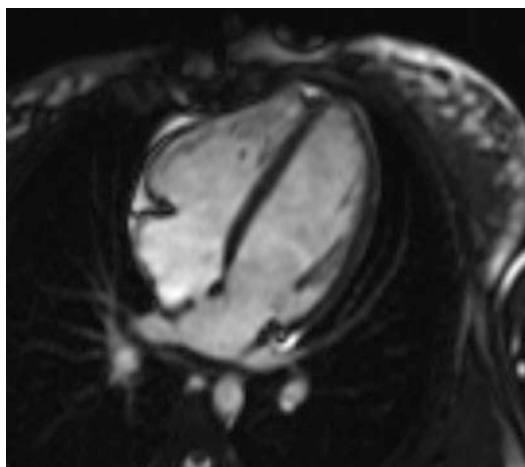


Fig. 6 Isolated secundum ASD. Four-chamber view image obtained from cine TRUFISP MRI sequence demonstrates an ostium secundum defect of less than 10 mm in diameter. The QP/QS ratio was estimated to be 1.2:1

2.2.3 Sinus Venosus Defect

Embryologically, this defect results of an incomplete resorption of the embryonic sinus venosus leading to a communication between the right pulmonary veins and the SVC, IVC, or the right atrium. The anomaly is a deficient separation of the pulmonary venous connection to one of the caval veins located at the junction of the right atrium rather than a true defect in the interatrial septum (Webb and Gatzoulis 2006; Van Praagh et al. 1994). Therefore, blood with high oxygen content from the pulmonary veins is directed into the vena cava/right atrium instead of the left atrium. Most commonly, this type of ASD is related to the superior vena cava where blood from the right superior and middle pulmonary veins flows in the right atrium (Fig. 8). In rare cases, it occurs in relation with the inferior vena cava where blood from the right lower pulmonary vein flows in (Fig. 9) (Van Praagh et al. 1994; Brickner et al. 2000; al Zaghal et al. 1997; Kafka and Mohiaddin 2009).

This produces a larger left-to-right shunt than ostium secundum defects and leads to a threefold higher risk of developing pulmonary artery hypertension (Rajiah and Kanne 2010; Vogel et al. 1999). It is a relatively uncommon type of ASD, constituting approximately 5–10% of all ASDs (Webb and Gatzoulis 2006; Davia et al. 1973).

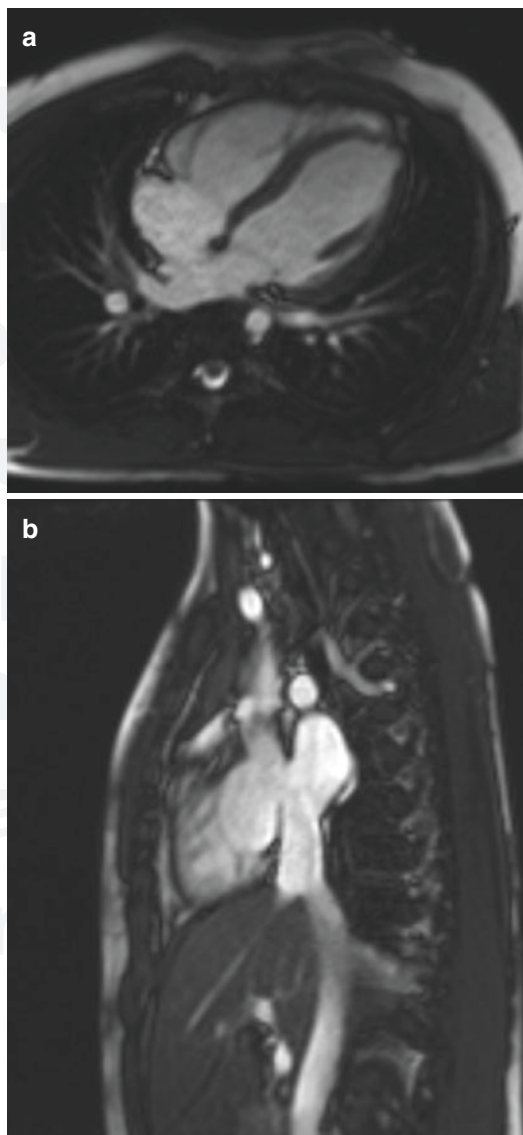


Fig. 7 Secundum ASD. (a) Four-chamber view and (b) perpendicular view of the interauricular septum images obtained from cine TRUFISP MRI sequence demonstrate an isolated, high-riding ostium secundum defect. The QP/QS ratio was estimated to be 1.4:1

2.2.4 Fenestrated (Unroofed) Coronary Sinus

The term fenestrated or unroofed coronary sinus corresponds to a defect of the wall between the coronary sinus and the left atrium allowing a left-to-right shunt (Van Praagh et al. 1994). It is a spectrum, ranging from partial fenestration to complete absence of the

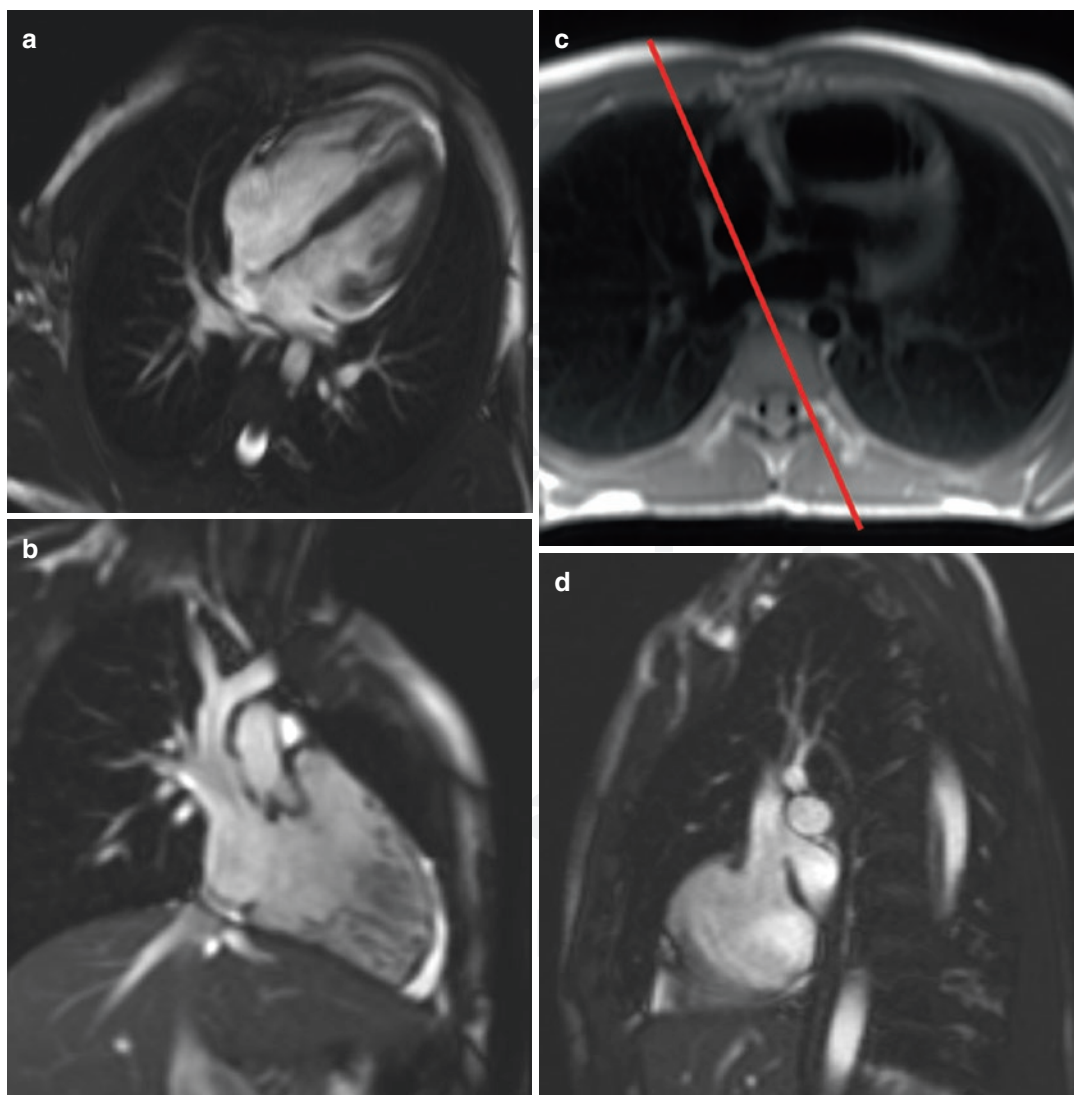


Fig. 8 Superior sinus venosus atrial septal defect. (a) Four-chamber view image obtained from cine TRUFISP MRI sequence shows superior type sinus venosus defect located in the posterior and superior aspect of atrial septum at the opening of right superior vena cava. (b) Coronal-view image obtained from cine TRUFISP MRI sequence demonstrates a partial anomalous pulmonary venous return. Blood from the right superior and middle pulmonary veins

is drained into the right superior vena cava instead of the left atrium. (c) Axial HASTE MRI image. The red line indicates the best orientation to depict sinus venosus ASD. (d) Sagittal oblique image obtained from cine TRUFISP MRI sequence (the orientation displayed by the red line in Fig. 8c) demonstrates a defect in the posterior and inferior aspects of right SVC like that found in the superior sinus venosus atrial septal defect ASD

wall between the left atrium and the coronary sinus. In the majority of cases, the communication is in the mid-portion of the coronary sinus. It is a rare cardiac anomaly accounting for only 1% of all ASDs (Ootaki et al. 2003a). It may occur as an isolated finding, often of little functional importance, or as part of

another congenital heart disease (Saremi 2014). An unroofed coronary sinus is almost always associated with a left SVC (Fig. 10) and sometimes with a secundum ASD (Ootaki et al. 2003a). It is relatively common in left isomerism heterotaxy syndromes (Matsuwaka et al. 1987).

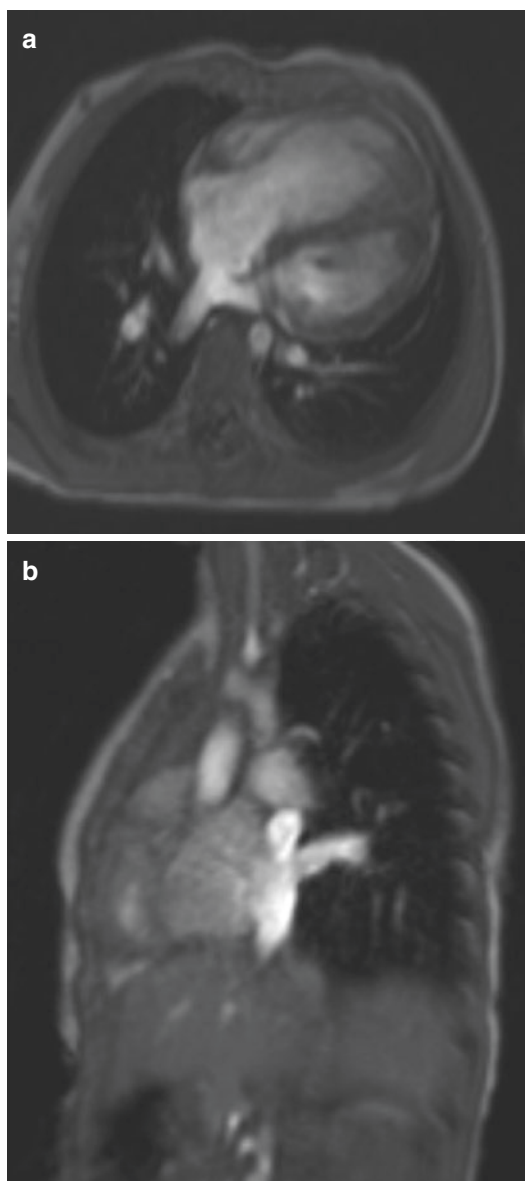


Fig. 9 Inferior sinus venous atrial septal defect. (a) Four-chamber view image obtained from cine FLASH MRI sequence shows a communication between the posterior aspect of inferior vena cava and the left atrium. Blood from the right inferior pulmonary vein is directed into the inferior vena cava/right atrium instead of the left atrium. The QP/QS ratio was 2.8:1. (b) Sagittal oblique view image obtained from cine FLASH MRI sequence demonstrates the right inferior pulmonary vein draining into the right atrium near the opening of inferior vena cava

2.2.5 Patent Foramen Ovale

Complete closure of foramen ovale occurs in almost 75% within the first two years of life. In 25% of people, anatomic closure does not

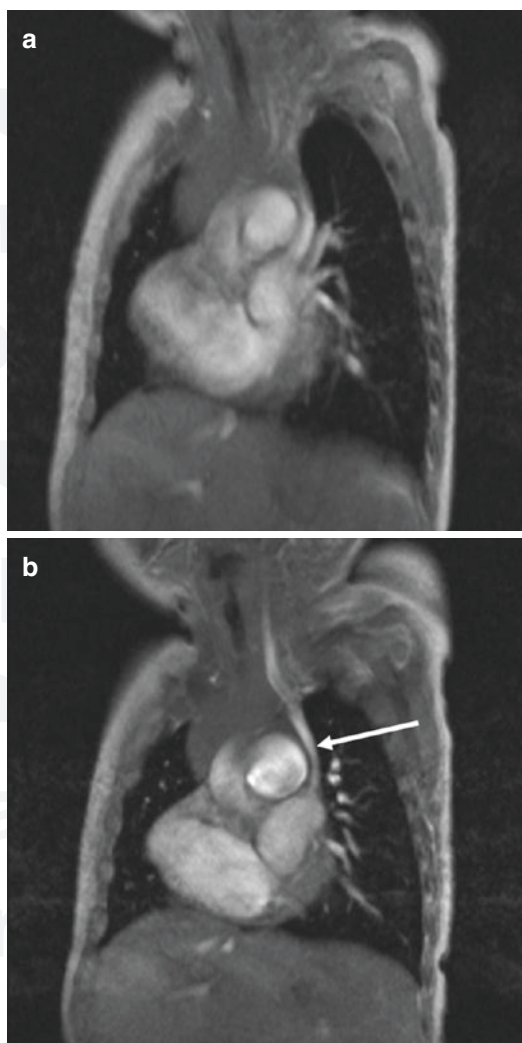


Fig. 10 (a), (b) Totally unroofed coronary sinus defect. Sagittal oblique images of cine FLASH MRI sequence show a persistent left superior vena cava (white arrow) draining directly into the left atrium due to a complete absence of common wall between the left atrium and the coronary sinus and interatrial communication corresponding to the opening of coronary sinus

occur. In case of persistent foramen ovale and patent valve of fossa ovalis, it may be of little or no clinical relevance and is therefore a quite common incidental finding on imaging studies and autopsies. In some cases, the valve is incompetent leading to a left-to-right shunt across the foramen. Therefore, these patients are at risk of right-to-left shunting complications. Also, this communication can be used as approach for angiographic cardiac evaluation or intervention by cardiologists.

2.3 Hemodynamic Consequence of ASD

The amount of shunting through the defect is determined by the size of the defect, by the difference of resistance in the systemic and pulmonary circulation, which relates to the compliance of the left and right ventricles, and by associated cardiac anomalies (Rajiah and Kanne 2010; Geva et al. 2014). In most patients, an ASD results in a left-to-right shunt. Defects smaller than 10 mm in diameter are usually associated with a small shunt and little or no enlargement of the right heart structures. Those with a pulmonic-to-systemic flow ratio (QP/QS) > 1.5:1 are usually hemodynamically significant. This is the magnitude of shunt needed for a right-sided volume overload and pulmonary overcirculation triggering a cascade of changes in the myocardium and in the pulmonary vasculature (Geva et al. 2014; Driscoll et al. 2006). A long-standing significant shunt will result in an impaired right atrial pump function, right ventricular dilatation, and myocardial changes (Sugimoto et al. 2011). It also comes to a remodeling of the pulmonary vascular bed, leading to pulmonary hypertension (Steele et al. 1987). In children with ASD, pulmonary hypertension is rare (Sachweh et al. 2006). In adults with a large ASD, pulmonary artery hypertension is a common finding and tends to increase with age (Geva et al. 2014; Humenberger et al. 2011). Eisenmenger syndrome is present in 5–10% of adults with untreated ASDs (Steele et al. 1987; Sachweh et al. 2006).

Ostium primum/endocardial cushion defects and sinus venosus defects are usually associated with a hemodynamically significant shunt and they do not improve over time (Geva et al. 2014). Therefore, they need surgical repair.

The natural history of secundum ASD is more variable. The probability of spontaneous closure depends on the size of the secundum ASD. Spontaneous closure occurs in approximately 56% of patients with an initial defect size of 4–5 mm, in 30% with a defect of 6–7 mm, and in 12% with a defect of 8–10 mm. If the secundum ASD does not close spontaneously, the size of the defect can decrease or even increase with age (Hanslik et al. 2006; Helgason and Jonsdottir 1999; McMahon et al. 2002; Saxena et al. 2005).

Patients with a defect >2 cm are usually symptomatic in childhood (Ko et al. 2009).

2.4 Indications for Defect Closure

A hemodynamically significant shunt causing enlargement of the right heart structures should be closed once the diagnosis is made, independently of symptoms (Geva et al. 2014; Baumgartner et al. 2010; Warnes et al. 2008). As mentioned above, a hemodynamically significant shunt is defined by a pulmonary-to-systemic flow ratio (QP/QS) > 1.5:1. Patients with an ASD and unexplained paradoxical embolism should be treated as well (Warnes et al. 2008). Studies have shown that even in patients older than 40 years, closure of ASDs increases long-term survival and limits the deterioration of function due to heart failure (Attie et al. 2001; Konstantinides et al. 1995).

Hemodynamically insignificant defects without other indications can be followed conservatively, keeping in mind the possibility of increased shunting in later life (Geva et al. 2014).

2.5 Treatment Strategies and Outcomes

Despite the fact that surgical and interventional approaches are available with the same efficacy, persistent foramen ovale and ostium secundum defects are usually closed with transcatheter septal occluder (Figs. 11 and 12) (Ko et al. 2009). Complications are rare. The most common major periprocedural complications are device embolization (0.7%) and pericardial tamponade (0.1%) (Abaci et al. 2013). Late complications are device embolization (0.1%), device thrombosis (0.2%), and device erosion through the atrial wall or aortic root (0.1%) (Abaci et al. 2013).

Large atrial septal occluders, especially in small pediatric patients, can become in contact or protrude with adjacent structures, especially the roof of the left atrium and the aortic root. In a study of Lapierre et al. (Lapierre et al. 2012), it has been demonstrated that with the growth of the children, the distance between the septal

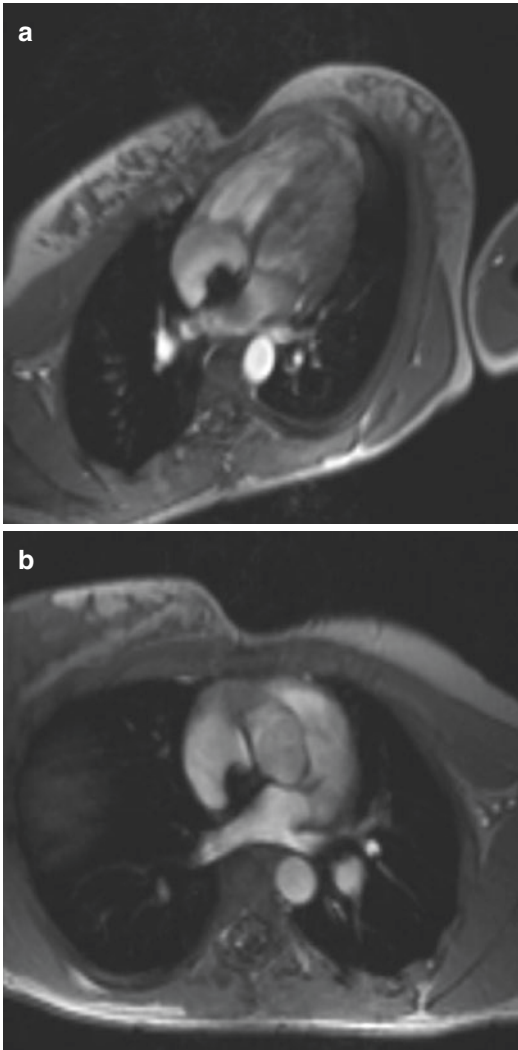


Fig. 11 Secundum ASD treated with septal device occluder. MRI obtained 2 years after procedure. Four-chamber view (a) and axial view (b) images of cineFLASH MRI sequence demonstrated an adequate position of septal device occluder for closure of ASD without residual shunt. Note the normal size of the right cardiac chambers

occluder device and the adjacent structures increases. In this study, there were no long-term complications with large atrial septal occluder devices. MRI remains the modality of choice to evaluate the effect of the ASD closure on the right cardiac chambers. MRI can also evaluate the atrial septal occluder itself and its relationship with other cardiac structures without significant blooming artifacts (Lapierre et al. 2012; Weber et al. 2008).

Sinus venosus defects as well as unroofed coronary sinus defects need surgical closure (Geva et al. 2014). Treatment of a sinus venosus defect also consists of diversion of the anomalous pulmonary vein into the left atrium (Rajiah and Kanne 2010).

In patients with AVSD (complete or partial), the treatment of choice remains surgical. The overall 10-year survival rate is variable in the literature, ranging from 70% to 100%, depending on the complexity of the anatomy and the era of surgery (Calkoen et al. 2016). Arrhythmias, including complete heart block that necessitates pacemaker placement, range from 0.5% to 7.5%. The most frequent indication for reoperation is left atrioventricular valve regurgitation. Other indications for reoperation are: subaortic stenosis, residual ASD or VSD, left ventricular outflow tract obstruction, left atrioventricular valve stenosis, and right atrioventricular valve regurgitation (Figs. 13 and 14). So, patients with repaired AVSD need echocardiographic follow-up exams on regular basis since these various complications can occur over time. Cross-sectional imaging, such as CT or MRI, can be required depending on the echocardiographic findings.

3 Ventricular Septal Defects

Ventricular septal defect (VSD) is the most common congenital heart disease if the bicuspid aortic valve is excluded. VSD is defined as a communication between the right and left ventricles through an opening or hole in the inter-ventricular septum. Since many ventricular septal defects close with time and many patients are asymptomatic, the prevalence varies between studies and examination techniques. A screening study with highly sensitive color Doppler study reported prevalence in newborns of up to 5%, much higher than in postmortem investigations in adults (Hoffman and Kaplan 2002; Roguin et al. 1995; Hoffman et al. 2004; Penny and Vick 2011).

VSD can occur as an isolated cardiac malformation, but it is frequently found as a part of complex cardiac malformations, including

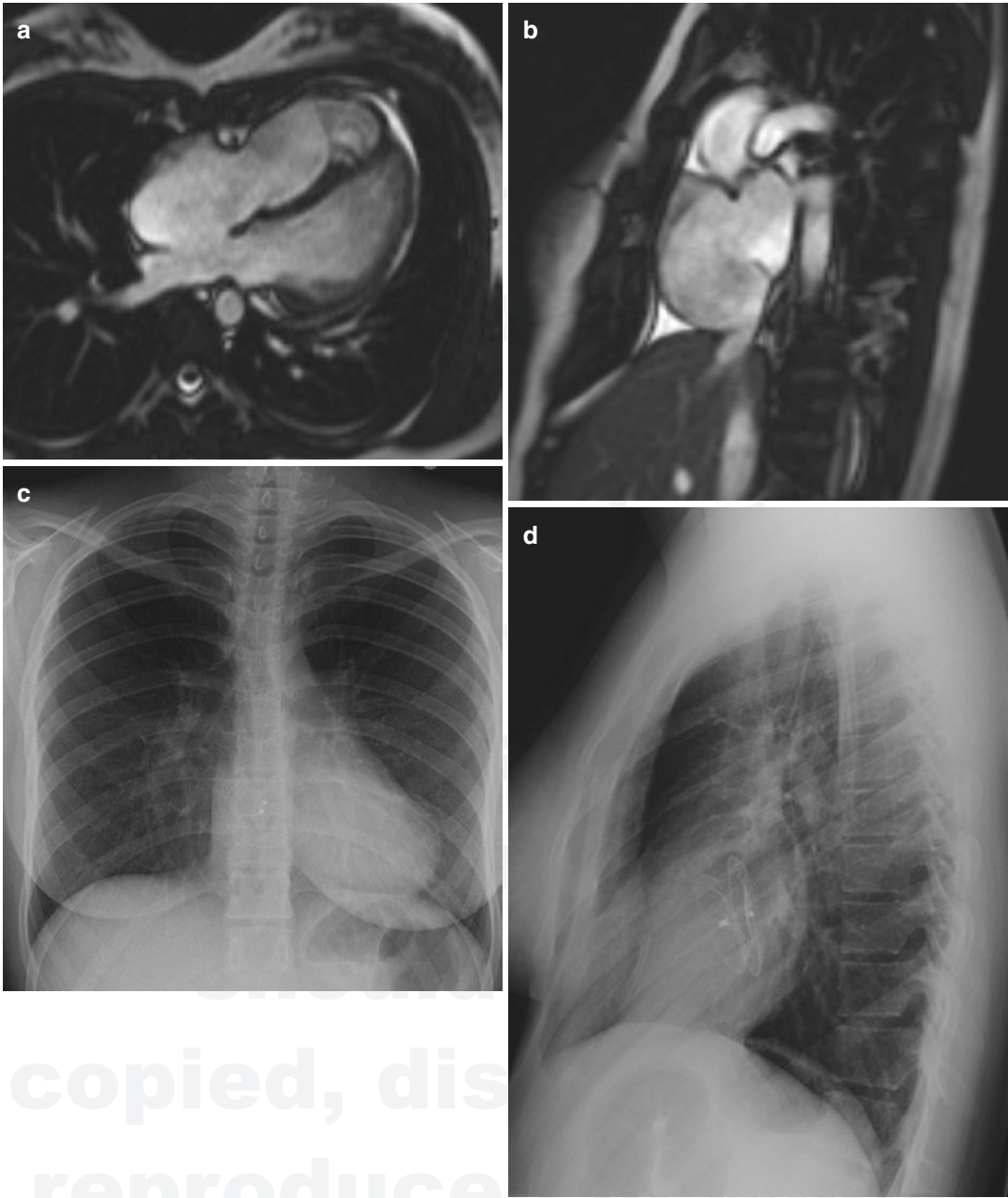


Fig. 12 Secundum ASD treated by septal device occluder. Four-chamber view (a) and perpendicular view (b) images of cine TRUFISP MRI sequence demonstrate large secundum ASD. Frontal (c) and lateral (d) radiographs obtained

four years after percutaneous treatment show septal device occluder in a good place, normal cardiac size, and normal arterial pulmonary vascularization

double outlet right ventricle, tetralogy of Fallot, univentricular atrioventricular connection, transposition of the great arteries, congenitally corrected transposition, or coarctation of the aorta

(Penny and Vick 2011; Wald and Powell 2006). Also, VSD represents the most common cardiac lesion found with chromosomal syndromes as trisomy 13, 18, and 21 (Allen et al. 2008).

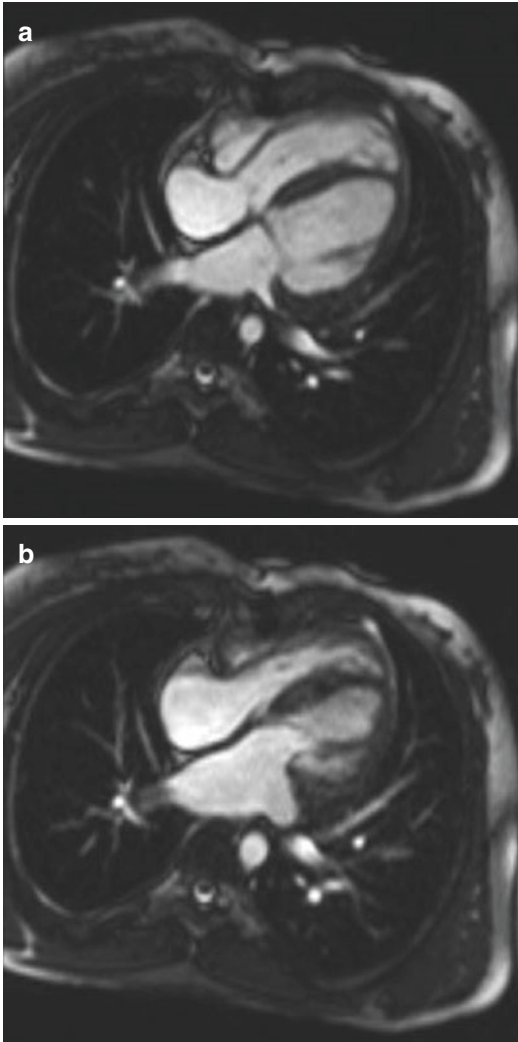


Fig. 13 Postoperative complete atrioventricular septal defect with stenosis of the mitral valve. (a), (b) Four-chamber view images of cine TRUFISP MRI sequence demonstrate no residual ASD or VSD but a limited opening of mitral valve creating a stenosis with secondary enlargement of the left atrium

In patients in which ventricular septal defects are the main pathology, echocardiography is usually sufficient and another imaging modality is rarely needed. Nowadays, the anomalies can be studied using CT or MRI with well-known advantages and limitations. Both can be used to define the anatomy of VSDs, its influence on the cardiac chambers, and associated anomalies. However, cardiac MRI stays the cross-section modality of choice because, in addition to provide a comprehensive assessment of the cardiac

anatomy, it can accurately quantify shunts, ventricular size, as well as cardiac and valve functions. Computed tomography angiography (CTA) should be reserved for patients for whom MRI is almost impossible or difficult to perform. However, VSDs can be difficult to analyze at cardiac MRI for radiologist or cardiologist unfamiliar with this subject.

In this section, a review of the classification of ventricular septal defects and the associated abnormalities is presented. Also, a MRI approach for the evaluation of VSD is proposed and the data that should be obtained by cardiac MRI prior to surgery or intervention are outlined.

3.1 Classification of Ventricular Septal Defect

Ventricular septal defects can occur in any portion of the ventricular septum (Allen et al. 2008). Several nomenclature schemes of ventricular septal defects are in use. Jacobs et al. (Jacobs et al. 2000) summarized the different nomenclatures in a paper published in the year 2000. The choice of the classification system to use for ventricular septal defects is not especially important. What is important is that all of the cardiologists, radiologists, and heart surgeons in a clinic use the same nomenclature as much as possible, as a way to avoid misunderstandings.

For a more precise description and classification, knowledge of the anatomy of the ventricular septum is necessary. The ventricular septum is made of four basic components (Fig. 15): the *inlet* septum, which is contiguous with the atrioventricular valves; the *muscular* component, which represents the main portion of the ventricular septum; the *outlet* septum between the aortic and the pulmonary valve, also referred to as the infundibular, conal, supracristal, or subarterial septum; and the *membranous* component, which is found at the base of the heart where the three other components converge and below the right and non-coronary cusps of the aortic valve (Hugh et al. 2016; Gersony 2001; Minette and Sahn 2006). Ventricular septal defects can also be divided into these four main categories, according to their location: inlet, muscular, outlet, and

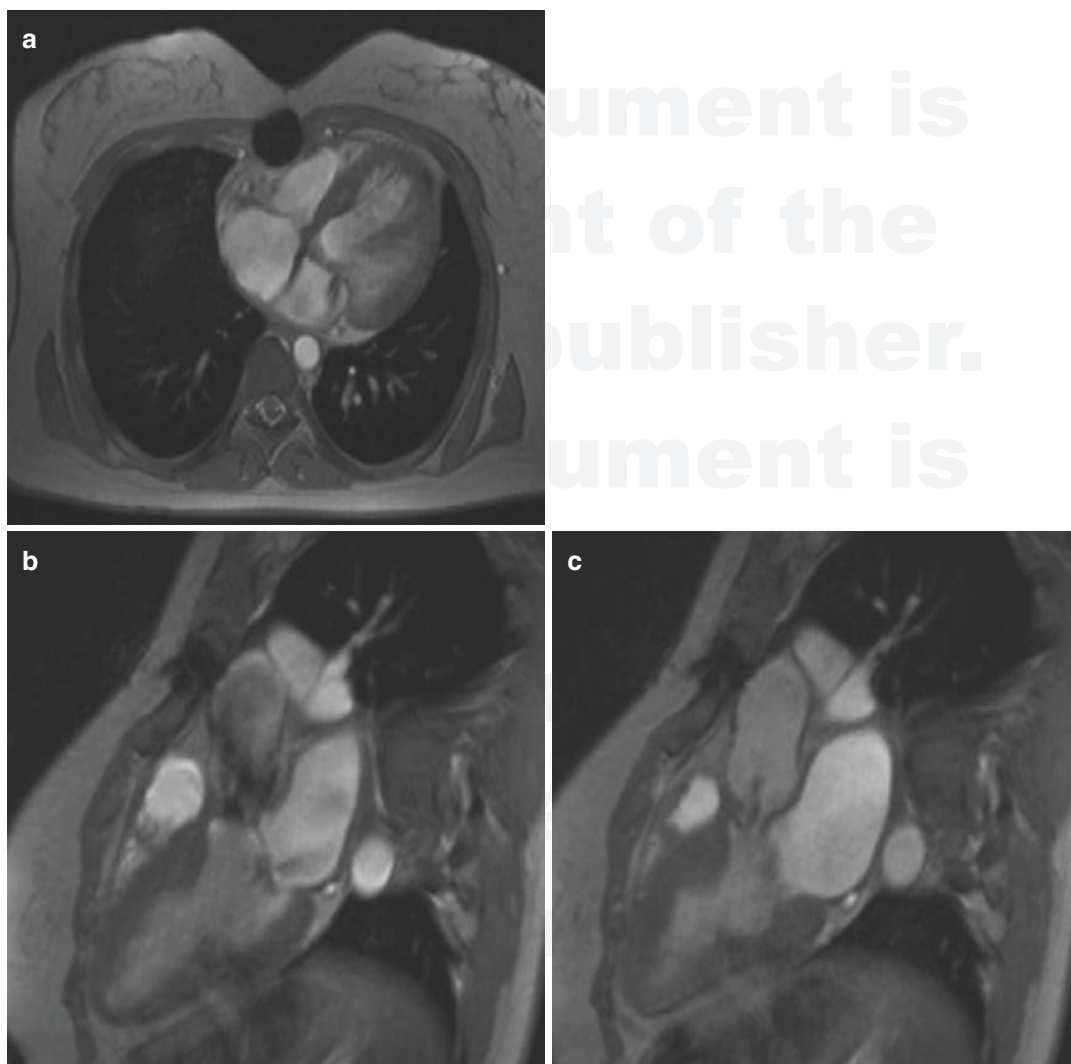


Fig. 14 Postoperative complete atrioventricular septal defect with aortic and mitral regurgitation and subaortic stenosis. (a) Four-chamber view image of cine FLASH MRI sequence shows correction of complete atrioventricular septal defect without any residual shunt but with left atrioventricular valve regurgitation. (b), (c) Three-

chamber view images of cine FLASH MRI sequence demonstrate a reduction in caliber of the left ventricular outflow tract associated with a membrane in the subaortic region creating a subaortic stenosis. Aortic regurgitation is also visualized

membranous (Soto et al. 1980). Defects can be limited to one component of the ventricular septum or can include several components. Thus, accordingly, more subtypes of the four main types may be mentioned.

3.1.1 Inlet Septum Defects

Isolated inlet septum defects are uncommon. This type accounts for 5–8% of all VSDs. Inlet defects are located posterior and inferior to the membranous

defect, beneath the septal leaflet of the tricuspid valve (Fig. 16). They are often small, but rarely close spontaneously (Gersony 2001).

Inlet septum defects can also be found in association with atrial septal defects and abnormalities of the atrioventricular valves, forming a so-called atrioventricular canal defect, more precisely described in the section about atrial septal defects (Gersony 2001). This anomaly is frequently seen in patients with trisomy 21.

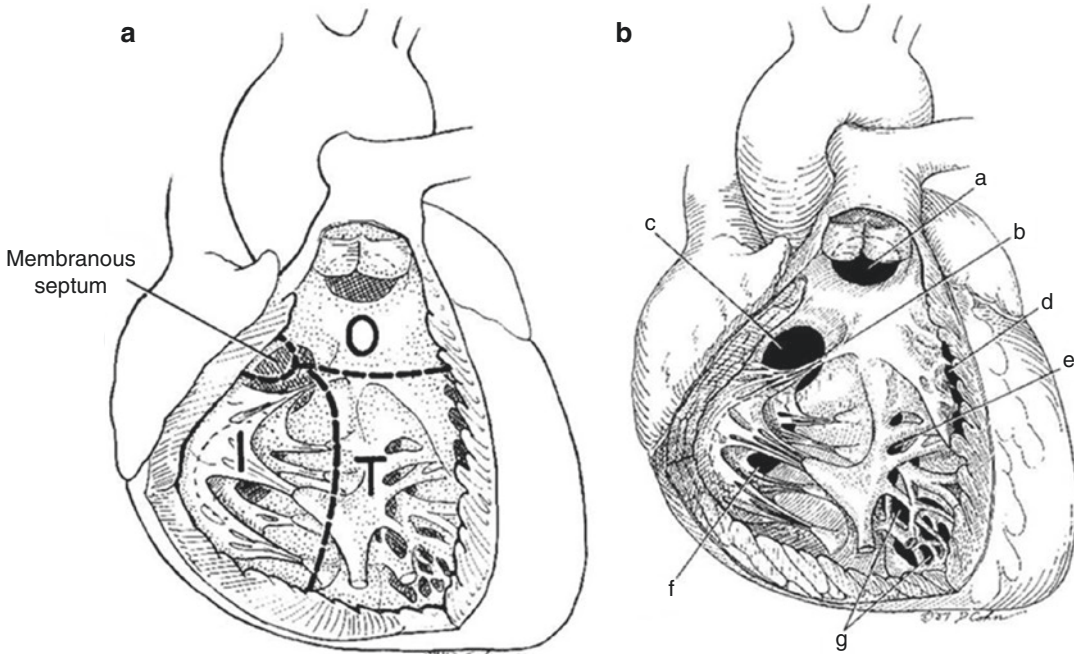


Fig. 15 (a) Components of the ventricular septum as viewed from the right ventricle. I: inlet septum; T: trabecular septum; O: outlet septum. (b) Anatomic position of defects: a, outlet defect; b, papillary muscle of the conus; c,

perimembranous defect; d, marginal muscular defects; e, central muscular defects; f, inlet defect; g, apical muscular defects. *Reprinted with permission from Allen et al. (2008)*



Fig. 16 Four-chamber view cine FLASH MRI sequence image showing an inlet ventricular septal defect. Note that both atrioventricular valves are located at the same level

marginal (Wang et al. 2003; Allen et al. 2008). Central muscular VSD is located posterior to the septal band and in the mid-portion of the septum (Fig. 17). Apical defects, the most common, are found in the apical portion of the ventricles (Fig. 18). Small muscular defects near the septal-free margins have been called marginal defects. Muscular VSDs are frequently multiple and may give a “Swiss cheese” appearance (Allen et al. 2008).

Frequently, this type of VSD is difficult to analyze from the right ventricle because they are usually multiple bordering and overlying trabeculae, giving the appearance of multiple channels whereas on the left ventricular side, fewer overlying trabeculae are present and the multiple channels seen from the right ventricular side, coalesce to a single defect.

Small, isolated muscular defects often close spontaneously due to muscular growth at the margins of the defect. Muscular ventricular septal defects seen in fetal echocardiography may not be visible anymore at birth (Gersony 2001).

3.1.2 Muscular Ventricular Septal Defects

Muscular VSD accounts for 5–20% of all VSDs. They are located entirely within the muscular septum. The location of VSD can be central, apical, or

3.1.3 Membranous Ventricular Septal Defects

Membranous ventricular septal defects are the most common VSDs, accounting for 80% of all

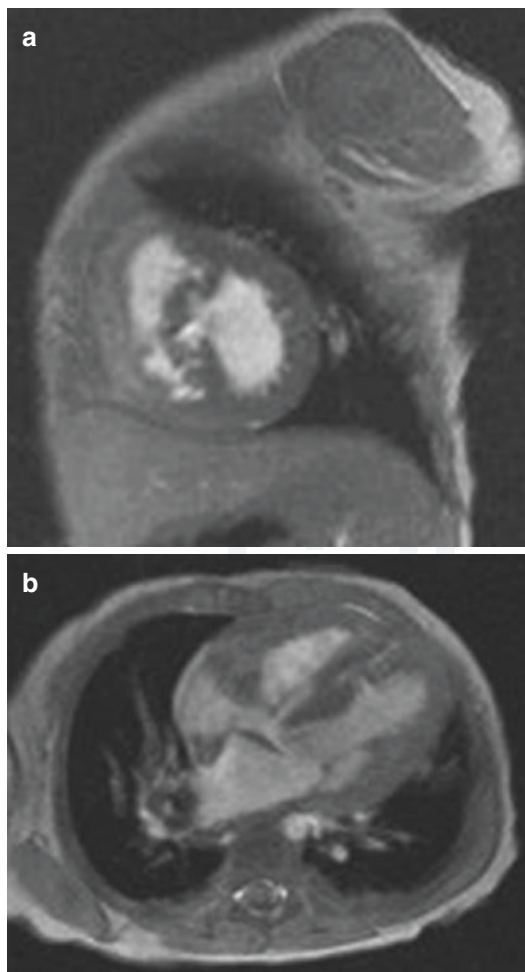


Fig. 17 Central muscular VSD. (a) Short-axis view and (b) four-chamber view cine FLASH MRI sequence images showing a central muscular ventricular septal defect. Note that on the right ventricular side, multiple channels are apparent and coalesce to a single defect on the left side

VSDs (Allen et al. 2008). They lie in the membranous part of the septum at the base of the heart, in the outflow tract of the left ventricle beneath the aortic valve (Fig. 19). Through this defect, there is a fibrous continuity between leaflets of the tricuspid and aortic valves (Penny and Vick 2011).

Pure membranous septal defects surrounded by fibrous tissue are rare. Most of the time, defects involve the membranous septum and an adjacent component. So the term “perimembranous” is more appropriate and can be subclassified as perimembranous inlet, perimembranous muscular, and perimembranous outlet, depending

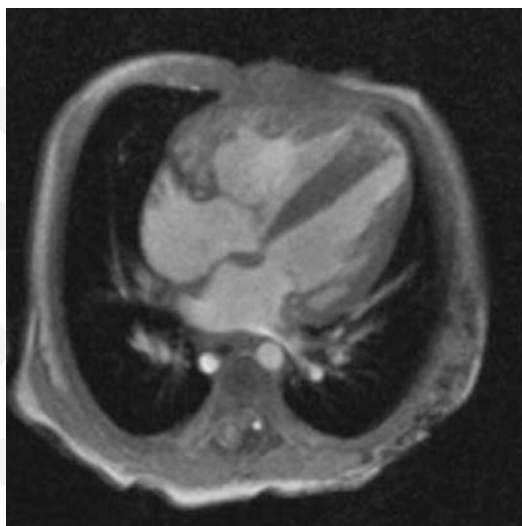


Fig. 18 Apical muscular VSD. Four-chamber view cine FLASH MRI sequence image showing a small apical muscular ventricular septal defect

on the extension of the defect (Allen et al. 2008; Minette and Sahn 2006). When the defect opens all part of the ventricle, it is called confluent defect (Penny and Vick 2011).

Small membranous defects may become smaller or close spontaneously as will be described in more detail in the section entitled Aneurysm of the membranous septum (Gersony 2001).

3.1.4 Outlet Ventricular Septal Defects

Outlet ventricular septal defects account for 5–7% of all VSDs, except for some Asiatic countries as Japan, where the incidence is approximately 30% (Allen et al. 2008). They are found in the muscular infundibulum, an area that, in the normal heart, constitutes a free-standing tube of muscular tissue which supports the pulmonary valve and separates the right and left ventricular outflow tract (Penny and Vick 2011; Minette and Sahn 2006). Defects of the outlet ventricular septum are also called subarterial, subpulmonary, conal, supra-cristal (superior to the crista interventricularis), or infundibular.

Anatomically, the defects in this region can be in the inferior or middle muscular part of the infundibulum (called also subaortic VSD) with a muscular rim always present under the pulmonary valve or in the subpulmonary region

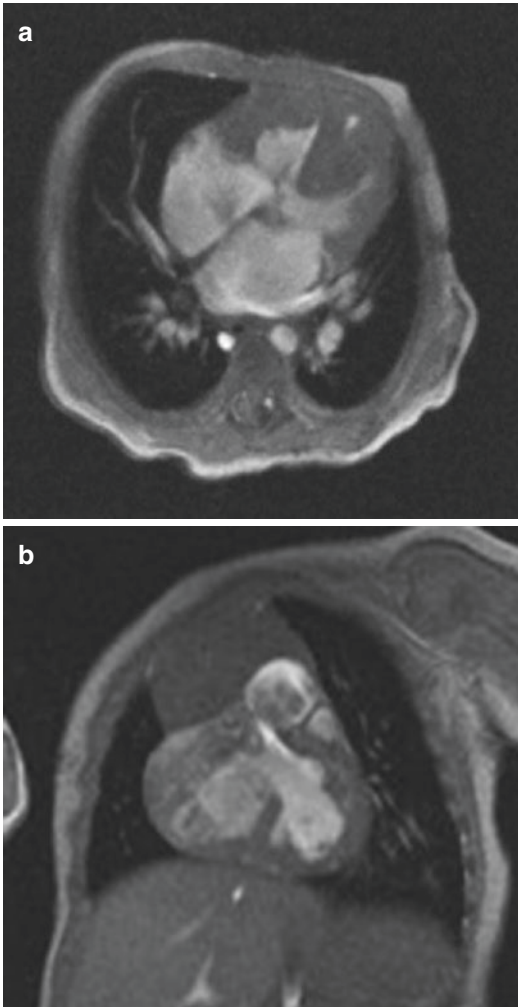


Fig. 19 Perimembranous VSD. (a) Four-chamber view and (b) short-axis view cine FLASH MRI sequence images showing a perimembranous ventricular septal defect with inlet extension

(directly under the pulmonary valve). Defects in this region therefore result in continuity between aortic and pulmonary valves (Penny and Vick 2011). These defects do not close spontaneously, but may become smaller over time. As outlet ventricular septal defects can involve the aortic sinuses, they may even close on the basis of prolapse of aortic valve leaflet tissue and/or sinus into the ventricular septal defect. This will be

described in more detail in the section entitled Prolapse of the aortic valve and aortic regurgitation (Gersony 2001).

3.2 Associated Findings

3.2.1 Prolapse of the Aortic Valve and Aortic Regurgitation

In perimembranous and outlet septum ventricular septal defects, the defects are located near the annulus of the aortic valve, and therefore the anatomical support of the aortic valve can be insufficient (Tatsuno et al. 1973). As a result, prolapse of the unsupported aortic valve leaflet and sinus of Valsalva through the ventricular septal defect can occur (Wald and Powell 2006). Hemodynamic factors may promote and augment prolapse, as described by Tatsuno et al. (Tatsuno et al. 1973) (Fig. 20). The blood shunting through the ventricular septal defect in early systole forces the aortic valve to prolapse through the defect. Thereby the opening of the ventricular septal defect becomes narrower and the velocity of the blood shunting through the defect increases. This in turn pulls the aortic cusps into the right ventricular cavity. During diastole, the intra-aortic pressure forces the cusps of the aortic valve to close. The unsupported cusp is shifted toward the right ventricle and is separated from the other two cusps, resulting in incompetency of the aortic valve with subsequent regurgitation (Tatsuno et al. 1973). That means that although the leaflet prolapse reduces the orifice size of the ventricular septal defect, it may lead to an important impairment of the blood flow by causing aortic insufficiency. Usually, the right coronary cusp of the aortic valve is involved in patient with outlet VSD. In contrast, patients with perimembranous VSD can have herniation of the right coronary cusp or much less common the non-coronary cusps (Wald and Powell 2006). The prevalence of this complication is higher in patients with outlet VSD and the associated aortic regurgitation increases with age.

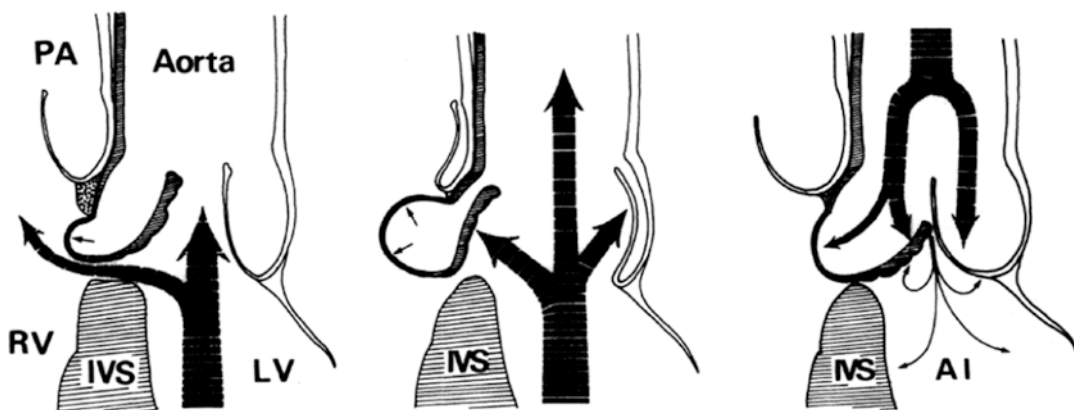


Fig. 20 Prolapse of the aortic valve and sinus of Valsalva and subsequent regurgitation: (Left) The flow of the shunting blood during early systole pulls the anatomically unsupported aortic valve and sinus through the VSD into the right ventricular lumen. (Center) In mid systole, the blood ejected from the left ventricle pushes unsupported sinus outward, producing a large bulge. (Right) In

diastolic phase, the intra-aortic pressure separates the unsupported cusp from the other two cusps, resulting in insufficiency of the aortic valve with subsequent regurgitation. LV left ventricle, RV right ventricle, IVS interventricular septum, PA pulmonary artery, AI aortic incompetence. Reprinted with permission from Tatsuno *et al.* (1973)

3.2.2 Aneurysm of the Membranous Septum

The septal leaflet of the tricuspid valve arises from the membranous septum. The membranous septum has two portions, the atrioventricular portion and the interventricular portion (Fig. 21). The atrioventricular portion lies superior to the attachment of the septal leaflet of the tricuspid valve and separates the left ventricle from the right atrium. The interventricular portion lies inferior to the attachment of the septal leaflet of the tricuspid valve and separates the two ventricles (Tandon and Edwards 1973).

As mentioned above, small membranous ventricular septal defects may become smaller over time or even close spontaneously. There are two contributing factors for the closure of the defect: growth of fibrous tissue at the margins of the defect and adhesion of the septal tricuspid leaflet to the edges of the ventricular septal defect. Due to the higher pressure in the left ventricle, a pouch protruding toward the right ventricle may form, referred to as aneurysm of the membranous septum (AMS) (Gersony 2001; Tandon and Edwards 1973). AMS is a rare disease, mostly

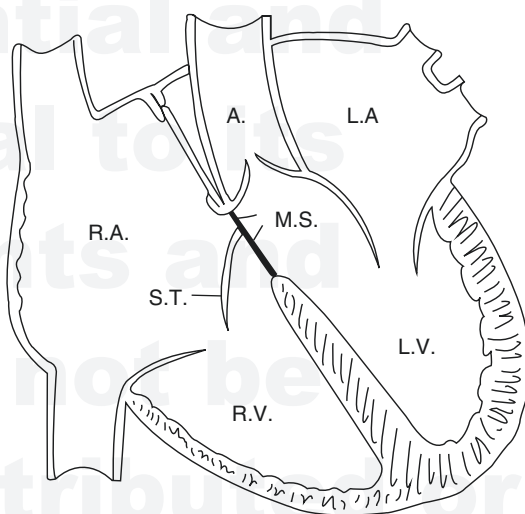


Fig. 21 Anatomy of the membranous septum. The membranous portion of the ventricular septum (M.S.) lies beneath the origin of the aorta (A). The septal leaflet of the tricuspid valve (S.T.) originates from the membranous septum. The atrioventricular portion lies superiorly to the attachment of the septal leaflet of the tricuspid valve and separates the left ventricle (L.V.) from the right atrium (R.A.). The interventricular portion lies inferiorly and separates the two ventricles. Right ventricle (R.V.). L.A. = left atrium. Reprinted with permission from Tandon *et al.* (Tandon and Edwards 1973)

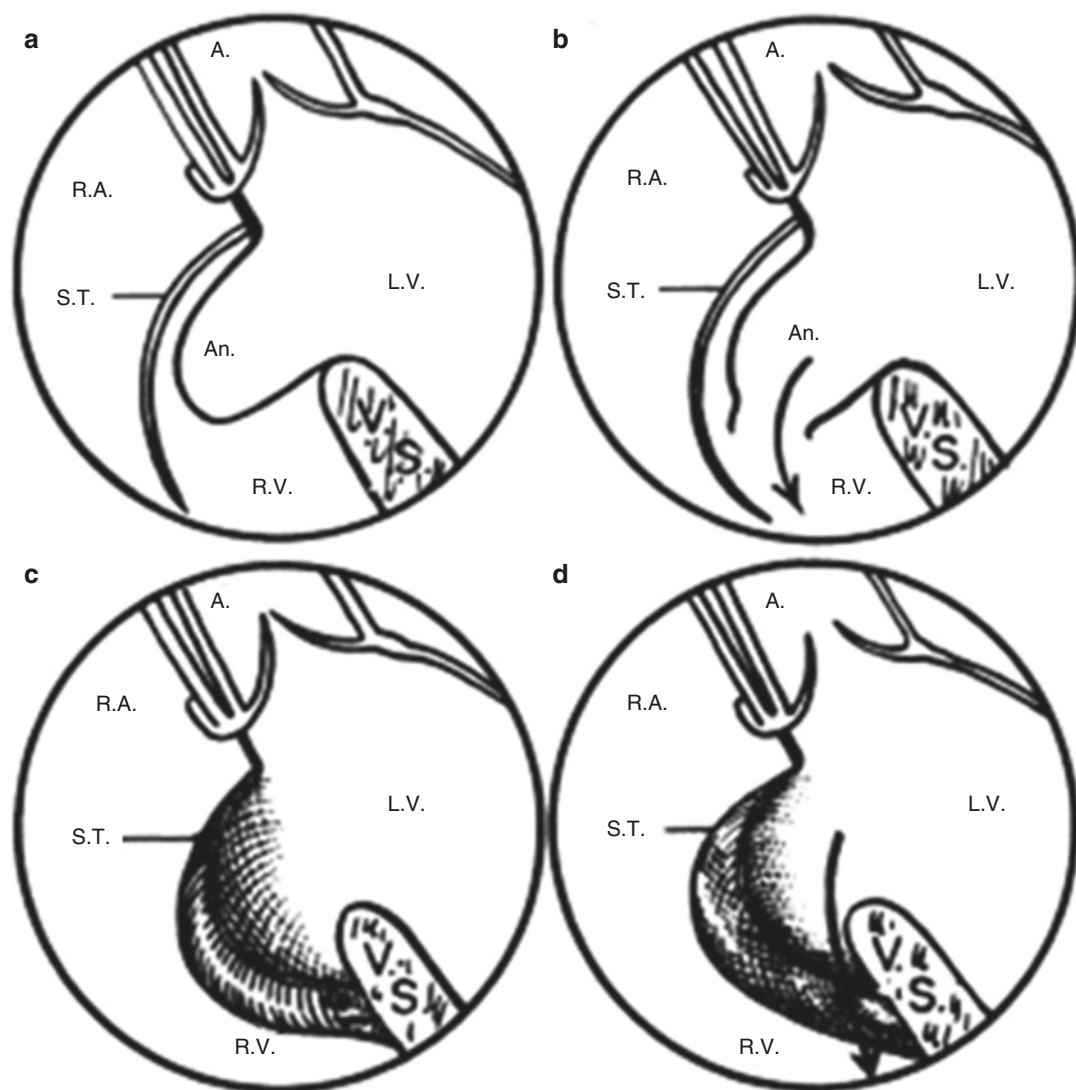


Fig. 22 Different aspects of the aneurysm of the membranous septum: (a) Congenital aneurysm of membranous septum. It is possible to distinguish the aneurysm from the septal leaflet of the tricuspid valve. (b) Ruptured congenital aneurysm of the septum. (c) Spontaneous closure of membranous

ventricular septal defect by adhesion of septal tricuspid leaflet to the ventricular septal defect. (d) Incomplete closure of the membranous ventricular septal defect by the septal leaflet of the tricuspid valve. Reprinted with permission from Tandon et al. (Tandon and Edwards 1973)

associated with other anomalies but occurring in up to 19–22.4% with VSD and in 20% with perimembranous VSD (Langer et al. 2007; Yavuz et al. 2010; Loukas et al. 2006). Basically, AMS may be congenital or acquired. Most congenital aneurysms are intact so that there is no inter-ventricular communication (Fig. 22). Rarely, a congenital aneurysm of the membranous septum may rupture leading to an inter-ventricular

communication. Congenital aneurysms are characterized by the identification of a septal leaflet of the tricuspid valve distinct from the wall of the aneurysm. In contrast, in aneurysms after spontaneous closure of ventricular septal defects, the septal leaflet forms an integral part of the aneurysm (Fig. 23). The closure may be complete or incomplete allowing some communication between the ventricles. Anomalies of the tricuspid

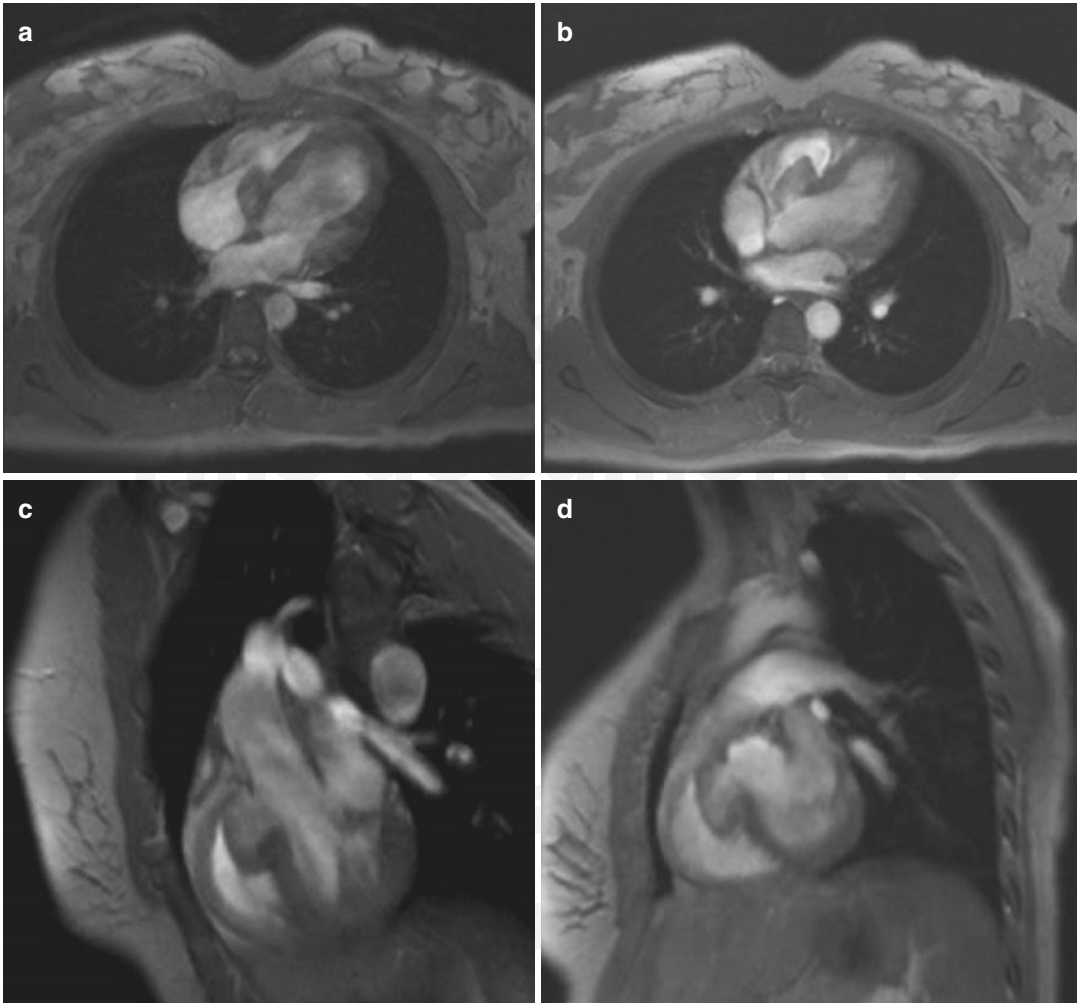


Fig. 23 Aneurysm of the membranous interventricular septum at cine FLASH MRI sequence. (a), (b) Four-chamber view, (c) three-chamber view, and (d) short-axis view images show a large aneurysm of the membranous interventricular septum protruding into the right ventricle

valve, which may be acquired for the left-to-right shunting, are frequently associated and result in tricuspid regurgitation.

Most AMS appear in the interventricular portion of the membranous septum (Tandon and Edwards 1973). Rarely, a defect can occur in the atrioventricular portion of the membranous septum (Gerbode's defect) producing an isolated left ventricular-to-right atrial shunt.

At imaging, AMS should be distinguished from sinus of Valsalva aneurysm. AMS arises from the right ventricular aspect of the membranous

and limiting the opening of the tricuspid valve. The septal leaflet of the tricuspid valve forms the wall of the aneurysm. The aneurysm closes almost completely the perimembranous VSD with only a small remaining defect

septum, beneath the septal leaflet of the tricuspid valve, and bulging forward into the right ventricle (Yilmaz et al. 1997). The criteria for the diagnosis of Valsalva sinus aneurysm include an origin above the aortic annulus, a saccular shape and normal dimensions of the adjacent aortic root and ascending aorta (Bricker et al. 2010).

3.2.3 Obstruction of the Ventricular Outflow Tract

VSDs can be associated with obstruction of the right or left ventricular outflow tract. The outflow

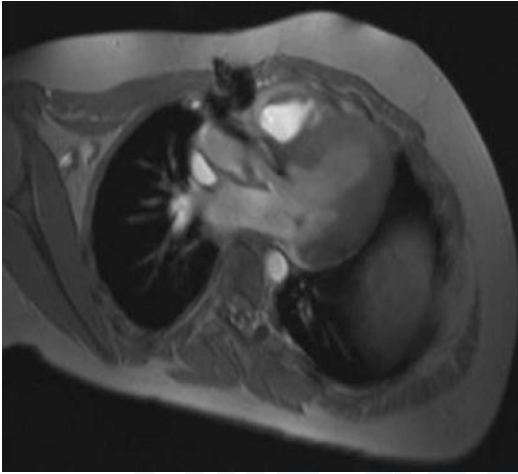


Fig. 24 Posterior deviation of the outlet septum. Three-chamber view of cine FLASH MRI sequence image demonstrates a posterior deviation of the outlet septum creating subaortic stenosis. Note that the associated VSD is not well shown. The anomaly is associated with a left ventricular hypertrophy

tract obstruction can arise from the VSD itself due to a certain malalignment of the outlet septum and the remainder of the ventricular septum. In consequence of anterior and posterior deviation of the outlet septum, either the right or left outflow tract can be obstructed (Fig. 24) (Saremi 2014). An anomalous or hypertrophic muscle bundle crossing the right ventricle from the interventricular septum to the right ventricular free wall, also known as double-chambered right ventricle, can be associated with VSD and cause obstruction of the right ventricular outflow tract/subpulmonary stenosis (Fig. 25) (Pongiglione et al. 1982; Kucher et al. 2001). Myotomy may remove this subpulmonary stenosis (van Son et al. 1993; Newfeld et al. 1976). Likewise, a crescent-shaped ridge of fibro-elastic tissue protruding from the left septal surface into the subaortic region can cause left ventricular outflow tract obstruction in patients with VSD (Newfeld et al. 1976).

3.2.4 Hemodynamic Consequences of VSD

The size of the ventricular septal defect and the pulmonary vascular resistance determine largely the shunt volume. If there is no obstruction of the right ventricular outflow tract and no pulmonary

hypertension, the direction of shunt is left-to-right. Left heart volume overload can occur with large left-to-right shunts but not right ventricular dilatation (Saremi 2014; Wald and Powell 2006). The shunt volume may decrease in the setting of right ventricular outflow tract obstruction or pulmonary vascular hypertension. In extreme cases, the shunt direction may invert, causing a right-to-left shunt (Eisenmenger syndrome).

3.3 Diagnostic Evaluation

In patient in whom ventricular septal defects are the main pathology, echocardiography is usually sufficient and another imaging modality is rarely needed. Nowadays, the anomalies can be studied using CT or MRI with well-known advantages and limitations. Both can be used but cardiac MRI remains the cross-section modality of choice with a reported sensitivity of more than 90% for detection of VSDs (Wang et al. 2003; Didier and Higgins 1986; Didier et al. 1986).

MRI evaluation is mainly indicated in the diagnosis of defects in the outlet septum as echocardiography is often unable to adequately image this region of the ventricular septum (Wang et al. 2003; Wald and Powell 2006; Bremerich et al. 1999). A cardiac MRI is considered to be superior to an ultrasound where there are multiple muscular ventricular septal defects. Furthermore, the ventricular septum is visible on MRI until the apex, which is not always possible when using echocardiography. In addition, MRI can depict complications of outlet septal defects, including secondary right ventricular hypertrophy (Bremerich et al. 1999).

Cardiac MRI can give many important information such as: (a) the location, the number, and the size of the ventricular component of the defect, (b) the relationship between the defects and the position of great vessels, (c) the presence or not of complications (aortic valve prolapse and aortic regurgitation), and (d) the quantification of the hemodynamic burden of the defect by providing reliable quantitative data on the shunt and by quantifying cardiac chamber volumes and function (the most accurate technique). All of these

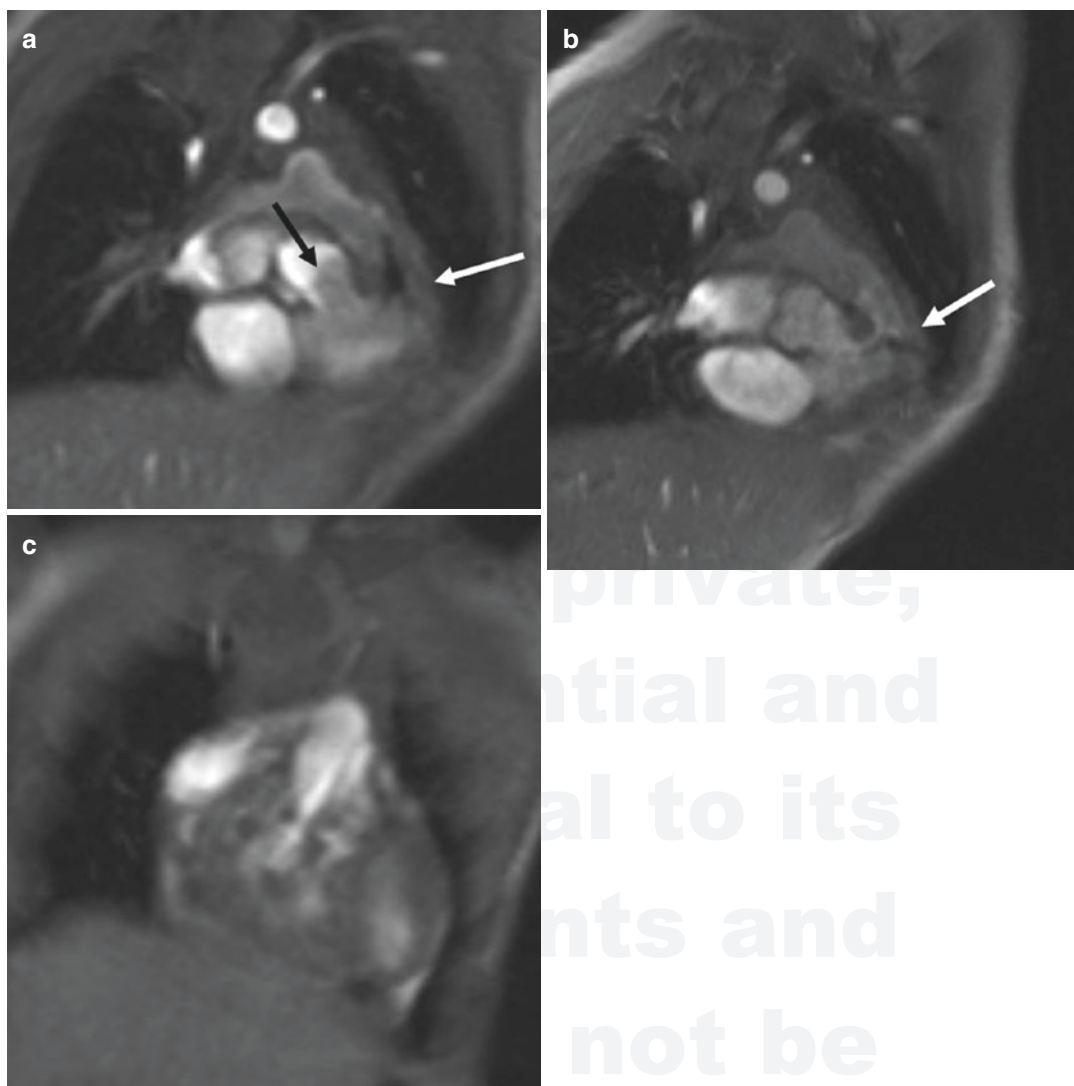


Fig. 25 Double-chambered right ventricle. Cine 2D-Flash sequence. (a), (b) Coronal oblique images show multiple infundibular muscular bands (white arrows) creating a stenosis and a large perimembranous VSD (black arrow). (c) Note that the distal portion is of normal size.

abovementioned criteria can be evaluated using a systematic MRI protocol and may influence the timing and the approach of correction (Table 1) (Parsons et al. 1990; Studer et al. 1982; Pennell et al. 2004; Wald et al. 2015).

However, VSD can be difficult to analyze at cardiac MRI for radiologist or cardiologist unfamiliar with this subject. An approach similar to the echocardiography is helpful.

This feature helps to distinguish a double-chambered right ventricle from a Tetralogy of Fallot. In Tetralogy of Fallot, the outlet septum has a leftward and anterior deviation

The inlet, membranous, and muscular VSD can be evaluated by the coverage of entire interventricular septum in the four-chamber and short-axis views. In using these two different standard plans, it is possible to analyze the anatomy of the interventricular septum and the repercussion on the tricuspid valve and cardiac chambers.

Another acquisition that can be used to clarify membranous and outlet ventricular septal defects

Table 1 Institutional cardiovascular MRI protocol for ventricular septal defects

1. Localizers through the thorax images
2. Morphologic examination with half Fournier shot turbo spin echo (HASTE) or balanced steady-state free precession (SSFP) according to heart rate in axial plane of the thorax
3. Four-chamber SSFP cine stack views to cover all the interventricular septum
4. Short-axis SSFP cine stack views to cover all the interventricular septum
5. Three-chamber SSFP view of the left ventricular outflow tract
6. Sagittal SSFP cine view of the right ventricular outflow tract
7. Axial true plane of the aortic root in SSFP cine view
8. Perpendicular SSFP cine view or gradient echo cine view on the VSD and on the aortic root (planes depend on the site of the VSD)
9. Qp/Qs evaluation by flow mapping on:
(a) Left pulmonary artery
(b) Right pulmonary artery
(c) Main pulmonary artery
(d) Ascending aorta

is an image in the plane of the aortic valve (Fig. 26). This acquisition can be obtained from a coronal image. On the plane of the aortic valve, the membranous VSD appears at the 10 o'clock to 11 o'clock positions (Fig. 27). It is in relationship with the left part of the non-coronary sinus and the right part of right coronary sinus of Valsalva. The infundibular muscular (subaortic) outlet VSD appears at the 11 o'clock to 12 o'clock positions whereas the subpulmonary VSD appears at the 12 o'clock to 1 o'clock positions. Both are in relationship with the right coronary sinus of Valsalva. Most of the time, VSD are identified on the aortic valve plan by a flow void due to a small size of the defect creating blood flow acceleration. When a flow void is found in the plane of aortic valve, a complementary image should be taken. In order to visualize the relationship between the VSD and the aortic valve, an acquisition perpendicular to the aortic valve, parallel to the jet, and crossing the center of the aortic valve should be obtained (Fig. 28). The resulting image is almost a three-chamber view and this projection permits to look for a prolapse of aortic valve and aortic regurgitation (Fig. 29). To evaluate adequately the ventricular

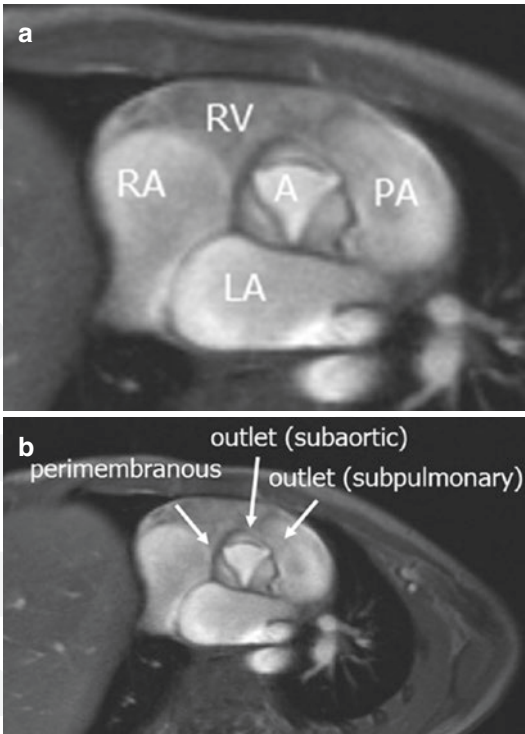


Fig. 26 (a), (b) Location of the VSD based on the aortic valve acquisition

outflow tract, a three-chamber view imaging for left side should be obtained and a sagittal plan for the right side.

3.4 Indications for Defect Closure and Treatment Options

A Qp:Qs value of >1.5 to 2.0 is considered by many cardiologists and cardiac surgeons as a therapeutic indication. Ventricular enlargement, pulmonary hypertension, or aortic regurgitation are also accepted indications for defect closure, either surgically or by transcatheter closure using occluder devices (Figs. 30 and 31) (Mongeon et al. 2010; Lun et al. 2001; YC et al. 2006). Small, isolated muscular defects often close spontaneously (Gersony 2001). However, multiple muscular defects with “Swiss cheese” appearance do not close spontaneously and require surgery (Saremi 2014). The surgical treatment of multiple muscular VSDs is technically difficult due to

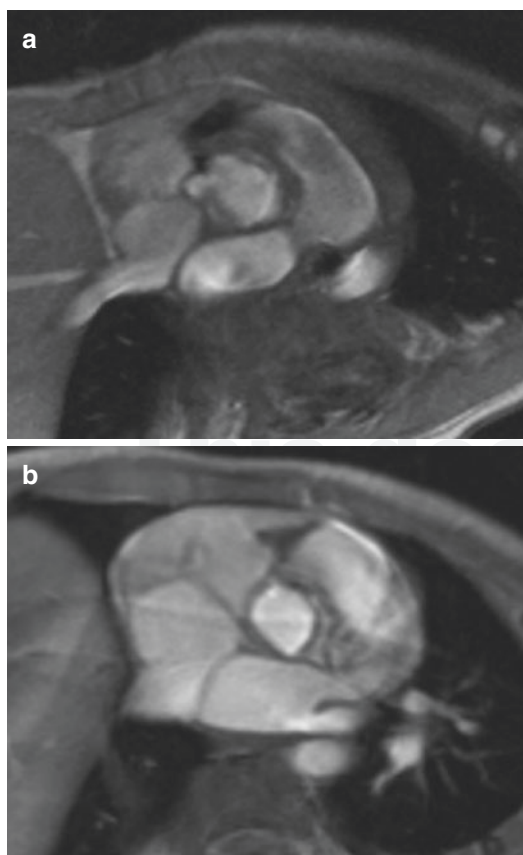


Fig. 27 Classification of VSD based on the aortic valve acquisition. Cine FLASH MRI sequence images show in (a) a jet 10 o'clock position indicating a perimembranous VSD and in (b) at 12 o'clock position, a small subaortic outlet VSD

often inadequate exposure of the defects and due to significant operative mortality and complications (Corno et al. 2013; Alsoufi et al. 2006; Stellin et al. 2000). Several techniques have been described, including single-stage repair with a transatrial or a transventricular approach (Wollenek et al. 1996; Kirklin et al. 1980; Kitagawa et al. 1998), initial pulmonary artery banding and closure of the residual VSDs in a second step (Alsoufi et al. 2006; Serraf et al. 1992; Seddio et al. 1999), interventional catheter device closure (Holzer et al. 2004), or a hybrid approach with operative patch and interventional device closure techniques (Bacha et al. 2005; Ootaki et al. 2003b; Brizard et al. 2004). Preoperative pulmonary artery banding is reserved for pediatric

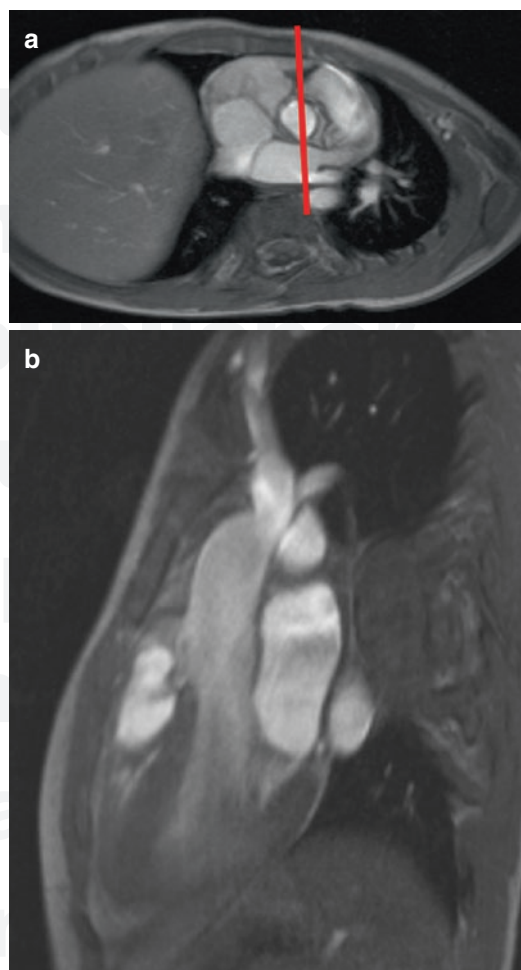


Fig. 28 Acquisition sequence to depict prolapse of the aortic valve. (a) In order to visualize the relationship between the VSD and the aortic valve, an acquisition perpendicular to the aortic valve, parallel to the jet, and crossing the center of the aortic valve, should be obtained. The red line delineates the optimal orientation. (b) The resulting image, almost a three-chamber view, shows the prolapse of the aortic valve reducing the size of the VSD

patients with other associated cardiac anomalies like double outlet right ventricle or with congestive heart failure uncontrolled by medical management before definitive correction.

A small VSD is defined as <1 cm in diameter with a left-to-right shunt of <50%, without pulmonary stenosis and pulmonary artery hypertension. Isolated, small VSDs only need prophylactic treatment for endocarditis (Saremi 2014; Neumayer et al. 1998).

Fig. 29 Two examples of VSD associated with prolapse of the right coronary cusp at cine FLASH MRI sequence images. (a) A saccular and (b) a more diffuse deformation are visualized as shown in Fig. 20 on the left and right respectively

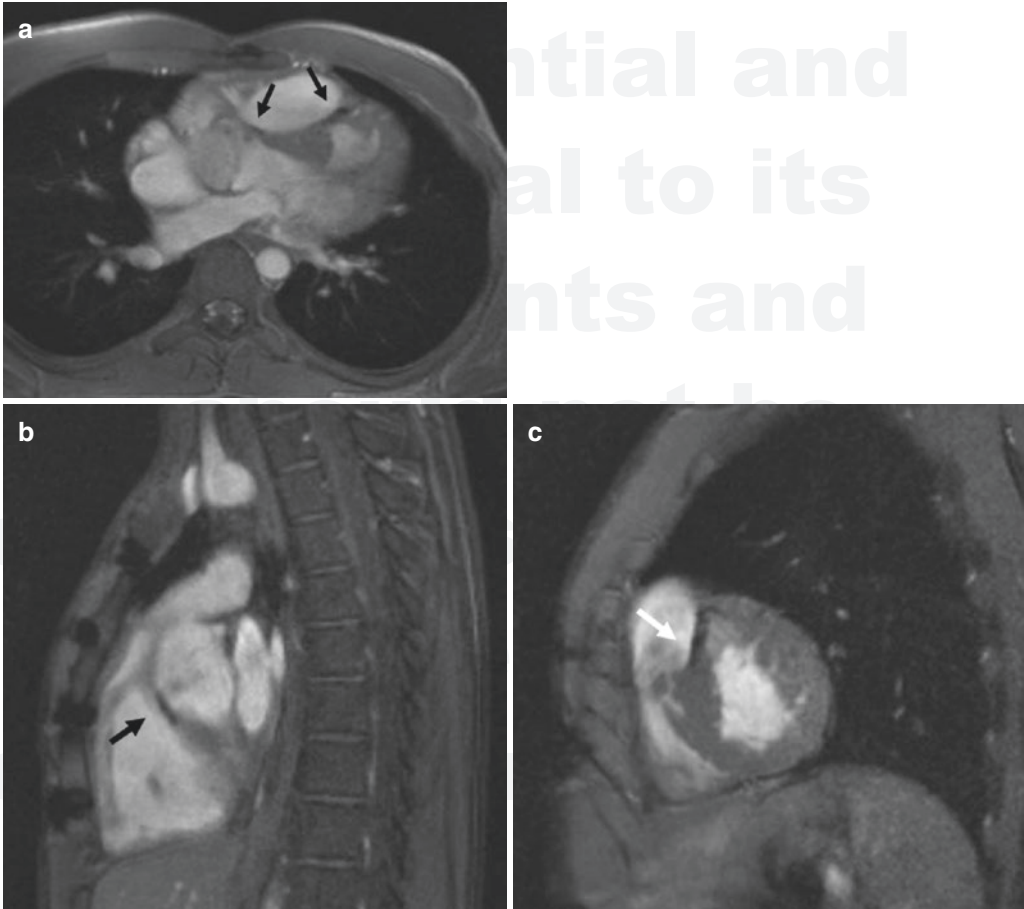


Fig. 30 Surgical closure of a perimembranous and muscular VSD at cine FLASH MRI sequence. (a) Axial, (b) sagittal oblique, and (c) short-axis view images show the

hypointense surgical patch at two locations: perimembranous (*black arrow*) and muscular interventricular septum (*white arrow*)

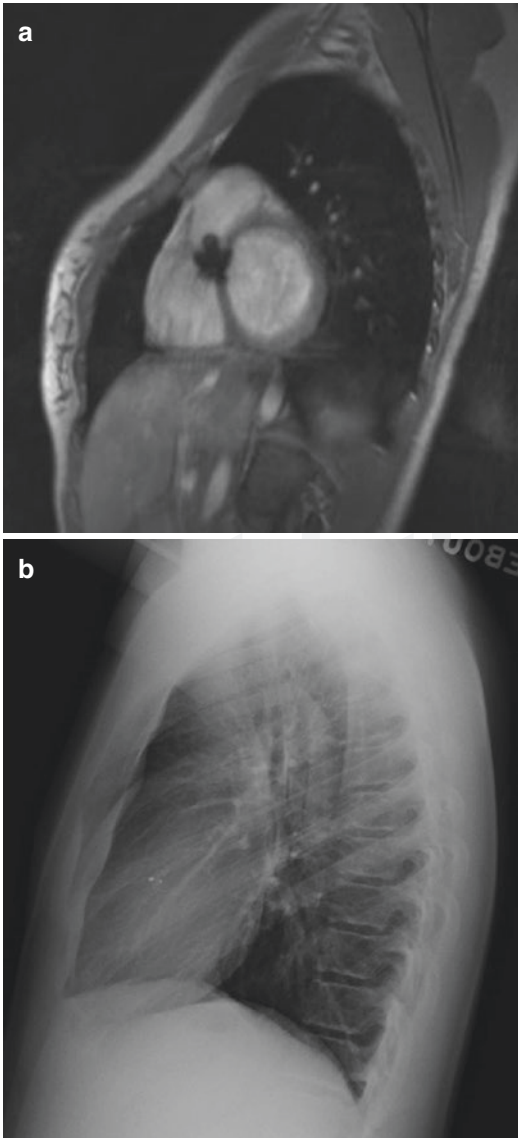


Fig. 31 (a) Short-axis view (left) and (b) lateral chest X-ray (right) in a patient with a perimembranous VSD treated with a septal occluder device

3.5 Outcomes

In general, children with VSDs have good outcomes and excellent long-term survival. Therefore, these patients are frequently seen in adult cardiology dealing with GUCH patients (Grow-Ups with Congenital Heart defects).

Conclusion

Atrial septal defects and ventricular septal defects are among the most common congenital cardiac anomalies. They can occur as isolated defects or in association with other congenital heart lesions. They are classified according to their location or embryologic origin. Important features include location, number, and size. Small defects are often hemodynamically insignificant, whereas larger defect can cause heart failure and alteration of the pulmonary vasculature. Echocardiography is often sufficient for diagnosis and treatment decision. Cross-section imaging (either MRI or CT) can be necessary for surgical or interventional planning, for functional analysis, and for the detection of posttreatment complications. In general, patients with ASDs and VSDs have good outcomes and long-term survival.

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