Chapter 13
Adult Congenital Heart Disease

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Abstract The increasing number of patients with either uncorrected or repaired congenital cardiac lesions is on the rise, likely reflective of improvements in care and detection. With more patients surviving well into adulthood, they often present for non-cardiac surgery. Echocardiography remains central to the detection, and aids in both surgical and device-based correction. It can also provide important prognostic information and quantify the effects (from the lesion) on ventricular function, pulmonary and systemic flow, etc. It is important for the beginner and intermediate practitioner to familiarize himself/herself with basic lesions encountered in adulthood, the echocardiographic techniques used to further identify and evaluate each condition, and the associated conditions that need to be sought and ruled out.

Keywords TEE for congenital heart disease · Ventricular septal defect · Atrial septal defect · Patent foramen ovale · Bubble study · Dilated coronary sinus · Persistent left SVC · PDA · Device closure · Tetralogy of fallot

Introduction

With the advancement of technology, transesophageal echocardiography (TEE) has quickly become invaluable in pediatric congenital heart surgery, both as a diagnostic tool and to assess repair [1]. Patients with congenital heart disease (CHD) are now

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surviving longer, and will ultimately need further anesthesia for non-cardiac and cardiac procedures [2]. Echocardiography has remained the cornerstone of diagnosing and managing these conditions. The basic perioperative transesophageal echocardiography (PTE) examination consensus statement recommends that a physician trained in basic perioperative TEE uses the exam to identify simple congenital heart disease lesions as a potential cause for right to left or left to right shunts [3]. Complex lesions are beyond the scope of the basic echocardiographer and if suspected, consultation with an advanced PTE echocardiographer or another diagnostic technique is warranted. Perioperatively, TEE provides real-time monitoring of ventricular filling, myocardial performance, and identification of intracardiac shunting, in addition to optimization of hemodynamic management strategies. A brief outline of the major congenital cardiac lesions and their echocardiographic correlates is provided here. The bicuspid aortic valve, which is the most common congenital heart lesion seen in adulthood, is discussed separately in Chap. 7.

Atrial Septal Defect (ASD)

ASDs comprise 7–8 % of all congenital heart disease and thus, are relatively common, either in combination with other lesions, or by themselves. Its location and size, which are related to its embryonic origin, often determine the magnitude of its hemodynamic effects. Due to the proximity of the left atrium to the probe, TEE results in excellent imaging of the interatrial septum (IAS), and is superior to TTE in this respect.

A patent foramen ovale (PFO), present in up to 27 % of population, can cause an intracardiac shunt if right atrial pressure exceeds the left atrial pressure [4]. Although a PFO represents a possible communication between the atria, it is technically not considered an ASD as there is no actual defect or tissue missing. Identification of a PFO may be accomplished through the use of color flow Doppler (CFD). While the flow through a PFO may be small, lowering the aliasing velocity (Nyquist limit) on CFD may help to identify the lower flow (Fig. 13.1; Video 13.1). As well, an agitated saline study with identification of "microbubbles" moving across the foramen during a Valsalva maneuver can help identify a PFO. The use of a Valsalva maneuver is important as the release of the Valsalva maneuver temporarily increases right atrial pressure in comparison to left atrial pressure, thereby exacerbating the shunt and visualizing the interatrial septal crossing of the agitated saline [4] (Fig. 13.2; Video 13.2).

The defects or gaps truly classified as ASDs involve some degree of absent tissue, allowing the potential for various degrees of intracardiac shunting. The ASDs are subdivided based upon their location, which relates to the defect during embryologic development. The subdivided defects are described below (Fig. 13.3):

1. Ostium secundum ASD:
   It is the most common ASD (approximately 70 %), and generally occurs in the area contained in the limbus of the fossa ovalis [5]. During embryologic

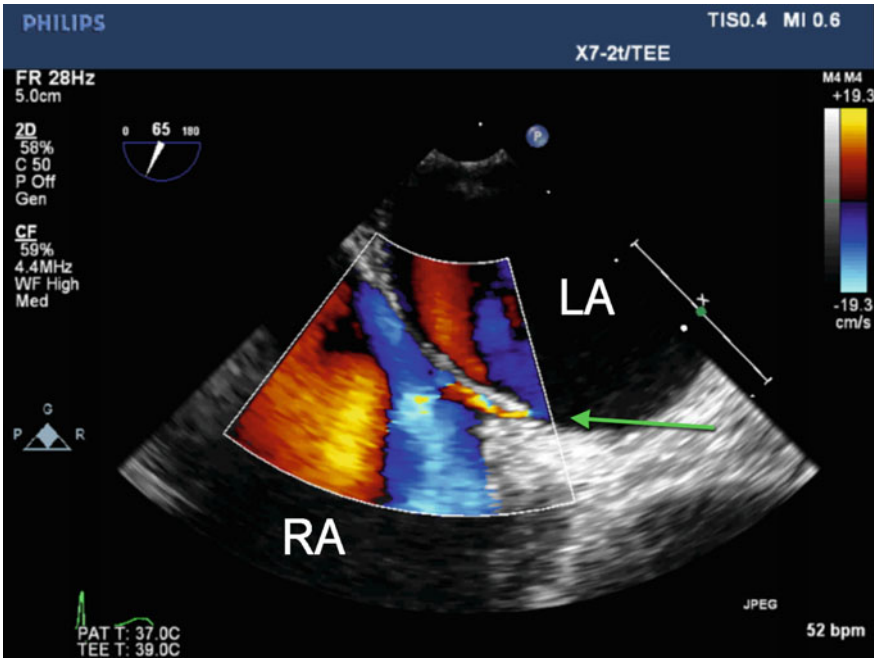

Fig. 13.1 Midesophageal view of interatrial septum with color flow Doppler. The *green arrow* indicates a *left* to *right* shunt across a patent foramen ovale (PFO). *LA* left atrium; *RA* right atrium

development, the septum primum grows toward the atrioventricular canal. An ostium develops centrally (termed the ostium secundum) which allows oxygenated blood in utero to cross the interatrial septum. Subsequently, a septum secundum develops to cover this ostium yet still allows flow through as the foramen ovale. After birth, with the increase in left atrial pressure from increased pulmonary blood flow, the foramen ovale is functionally pushed closed. Fusion of the two septums finalizes the process, leaving a fossa ovalis. A defective closure of the ostium secundum leads to the ostium secundum ASD (Fig. 13.4; Video 13.3). It may be circular in shape or may be a series of fenestrations associated with an aneurysmal interatrial septum.

2. Ostium Primum ASD:
These defects are the second most common type of ASD (approximately 20 %) and occur in the inferior and anterior portion of the IAS, near the AV valves. This defect generally represents the smallest degree of an atrioventricular canal defect. Ostium primum ASDs have been associated with Down's syndrome (Trisomy 21). During embryologic development, the septum primum develops in the direction of the atrioventricular valves, leaving the ostium primum to be developed by the endocardial cushion. Failure of this closure leaves the ostium primum atrial septal defect (Fig. 13.5; Video 13.4). As the endocardial cushion

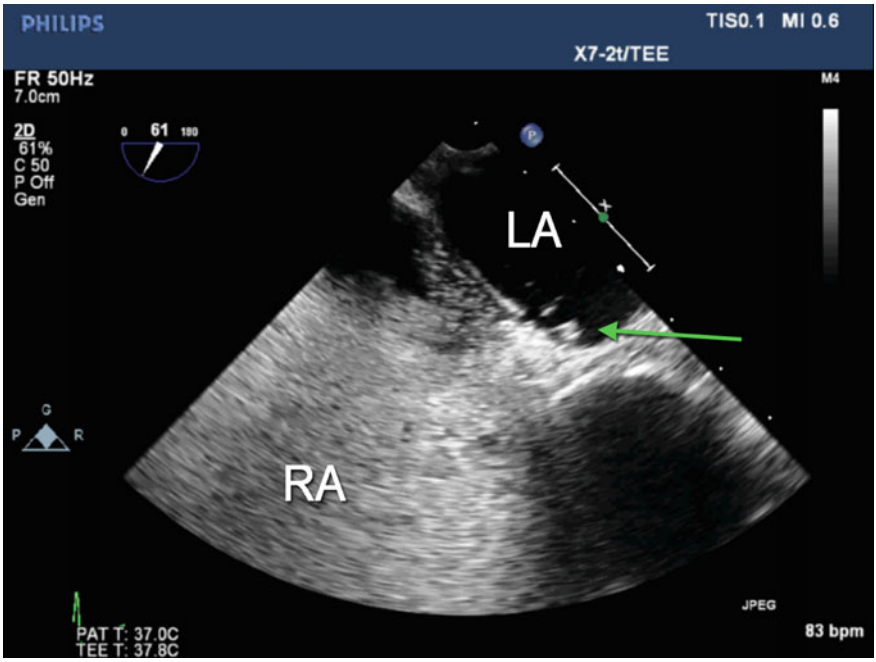

Fig. 13.2 Midesophageal view of interatrial septum during agitated saline injection. *Green arrow* indicates transseptal flow of agitated saline from right atrium (RA) to left atrium (LA)

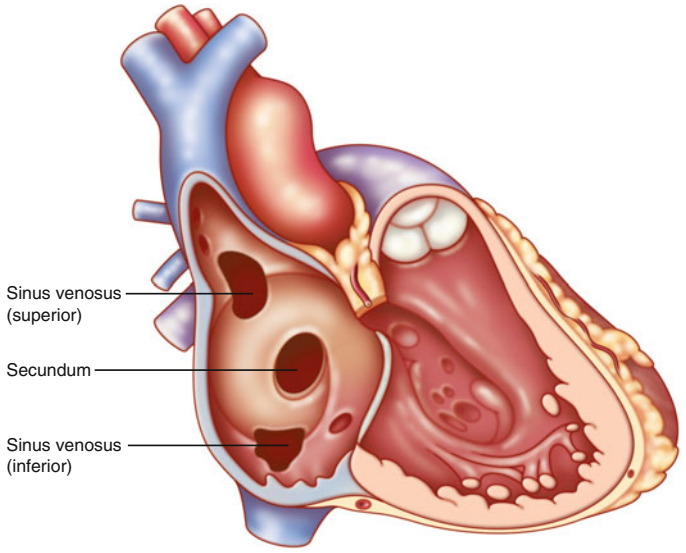

Fig. 13.3 Diagram of interatrial septum from the perspective of the right atrium and right ventricle

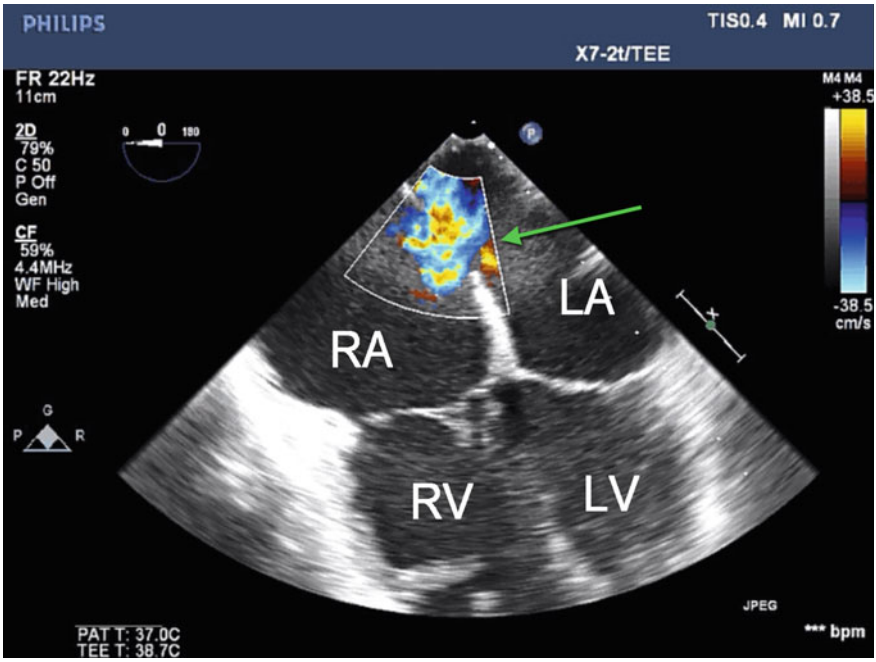

Fig. 13.4 Midesophageal four chamber view demonstrating an ostium secundum ASD (*green arrow*) with left to right flow noted on color flow Doppler. *RA* right atrium; *LA* left atrium; *RV* right ventricle; *LV* left ventricle

is also involved in the development of the atrioventricular valves, ostium primum defects can be associated with a cleft in the anterior mitral leaflet.

3. Sinus Venosus ASD:
   This defect represents an atrial communication adjacent to the attachment of either the superior or inferior vena cavae, and results in the respective vena cava overriding the defect. Sinus venosus defects account for about 8 % of all ASDs [5]. Embryologically, the vena cavae are derived from the sinus venosus. Abnormal resorption of the sinus venosus leads to a defect between the cavae and the left atrium (Fig. 13.6; Video 13.5). The defect is often associated with partial anomalous pulmonary venous return (i.e., anomalous right upper pulmonary vein draining into the right atrium).
4. Coronary sinus ASD (unroofed coronary sinus):
   These rare defects (<1 %) result from a partial or complete defect in the separation between the LA and the coronary sinus, resulting in "unroofing", and causing communication between the right and the left atria.

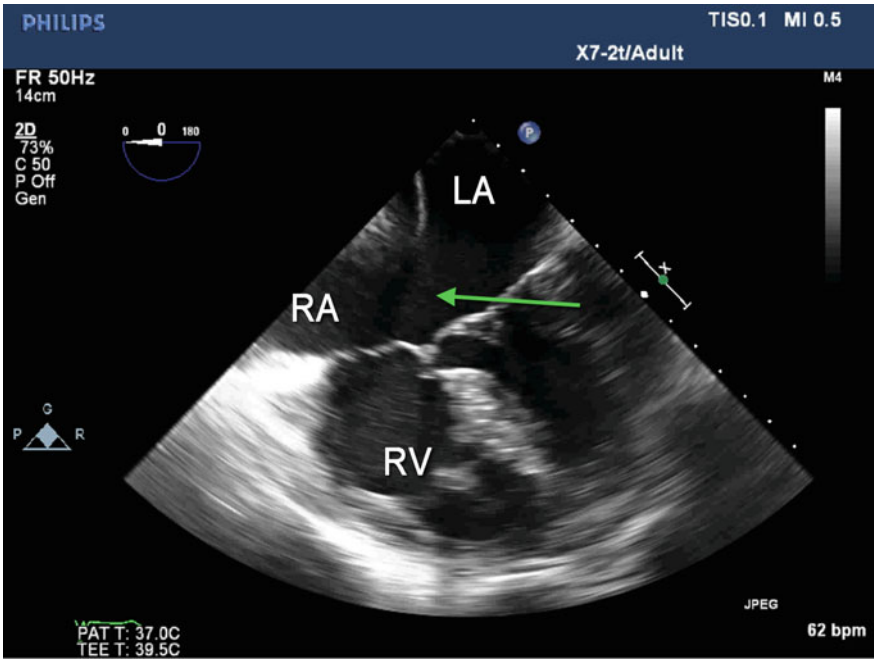

Fig. 13.5 Midesophageal four chamber view demonstrating an ostium primum ASD (*green arrow*). *RA* right atrium; *LA* left atrium; *RV* right ventricle

Echocardiographic Examination for ASDs

Atrial Septal Defects

2D	• Echogenic defect of tissue in interatrial septum – Secundum—ME Four Chamber or Bicaval Views – Primum—ME Four Chamber View – Unroofed Coronary Sinus—Difficult to visualize on 2D • Associated Findings – Cleft Anterior Mitral Leaflet (Primum) – Anomalous Pulmonary Venous Return (Sinus Venosus) – Atrial Enlargement – Ventricular dilation
CFD	• Interatrial flow – note directionality – May or may not be turbulent (dependent on ASD size)
Spectral	• Calculate Pulmonary to Systemic Flow Ratio (Qp:Qs)

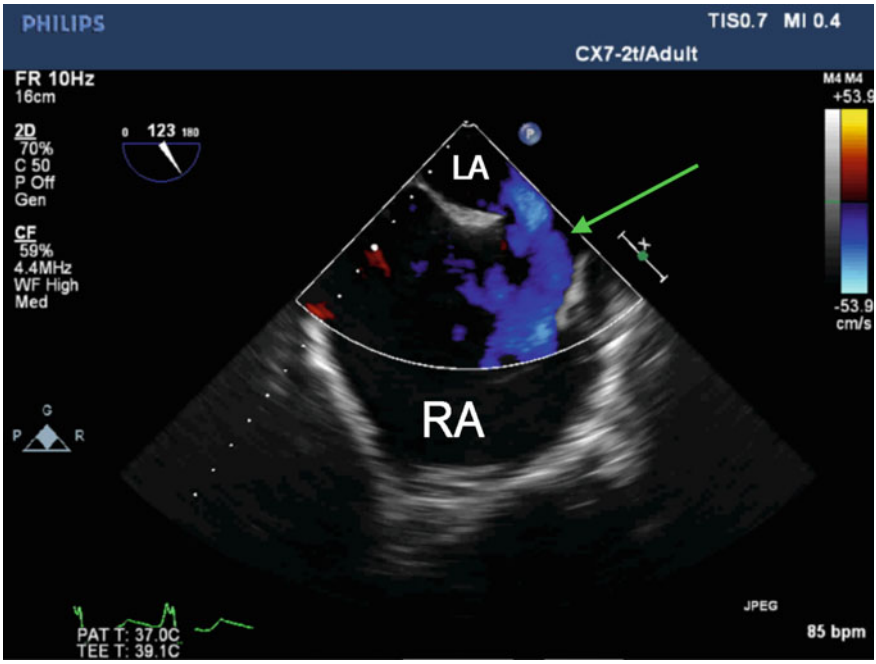

Fig. 13.6 Midesophageal bicaval view in a patient with a superior sinus venosus ASD and a grossly dilated right atrium (RA). The *green arrow* indicates left to right flow from the left atrium (LA) to the RA near the superior vena cava and RA junction

The midesophageal (ME) four chamber view can interrogate the majority of the interatrial septum, though probe withdrawal and insertion may be required for superiorly and inferiorly located ostium secundum defects as well as sinus venosus defects. Advancing the probe to the AV groove allows detection of ostium primum defects, evaluation of the coronary sinus, and Doppler evaluation of atrioventricular valves. Rotation of the probe to right and left is recommended to thoroughly interrogate the area. The ME Aortic Valve SAX or ME right ventricular inflow-outflow views can also be used to evaluate the septum, tricuspid valve (TV), and pulmonic valve (PV). In addition, these views can be used to quantify the tricuspid regurgitation, estimate pulmonary artery systolic pressure (PASP) as well as RV function, and search for abnormalities of venous return. The ME bicaval view provides a good cross-sectional display of the septum (superior to inferior), aligns the Doppler beam perpendicular to the septum, and is also an excellent view for agitated saline studies. This view is not particularly suited for detection of ostium primum defects, but can be modified by clockwise or counterclockwise rotation and omniplane manipulation to detect and evaluate all other ASDs. The transgastric mid-papillary SAX views can be used to detect flattening of the interventricular septum and help diagnose RV pressure or volume overload (see

Chap. 8). While beyond the scope of this textbook, TEE, especially 3D TEE, can be invaluable during device closure of ASDs by identifying the site and size of the defect, evaluating adequacy of the tissue ring around the defect (generally 5 mm) to hold the device, and follow the deployment of the device in real time.

Ventricular Septal Defect (VSD)

VSDs account for approximately 10 % of all adults with congenital heart disease, and can occur isolated or in association with other disorders. The ventricular septum can be divided into four components, each with its distinct morphology: Membranous, Inlet, Trabecular (muscular), Outlet [6] (Fig. 13.7). VSDs follow similar nomenclature, but can span more than one segment. Spontaneous closure is more likely for VSDs of the membranous or muscular type.

The most common of the four subtypes is the perimembranous VSD, occurring in 75–80 % of all VSDs. This defect is found in the membranous portion of the septum beneath the tricuspid valve and allows a connection to the left ventricular outflow tract (LVOT) immediately beneath the aortic valve [6]. It is best seen in the ME right ventricular inflow-outflow or ME aortic valve SAX views (Fig. 13.8; Video 13.6). Associated aneurysm of the membranous septum or accessory tricuspid tissue may be visualized. Perimembranous defects that occur high in the

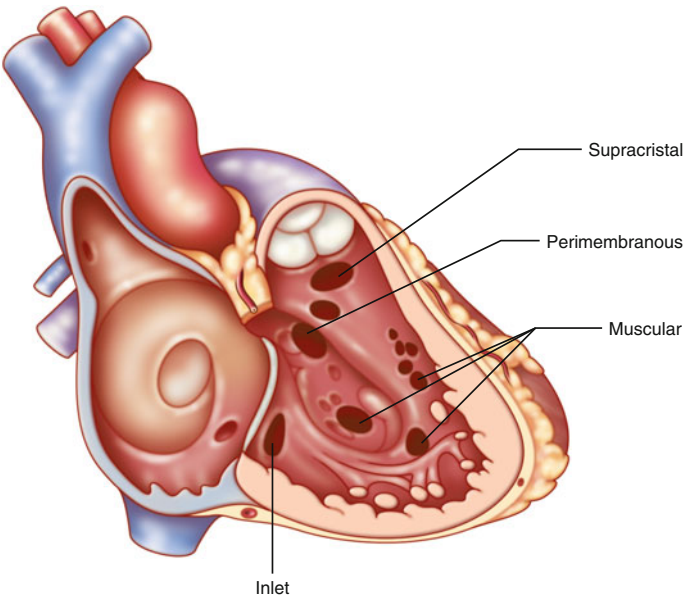

Fig. 13.7 Diagram of the interventricular septum from the perspective of the right atrium and right ventricle

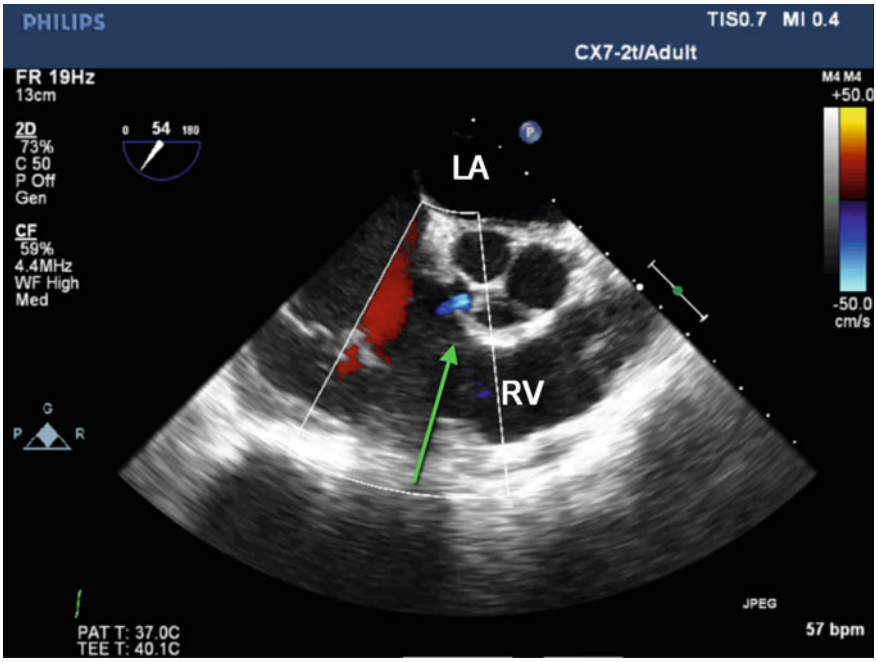

Fig. 13.8 Midesophageal right ventricular inflow-outflow view in a patient with a perimembranous ventricular septal defect (VSD) indicated by the green arrow. *LA* left atrium; *RV* right ventricle

LVOT can result in aortic regurgitation due to cusp herniation through the defect (most commonly the right coronary cusp).

Inlet VSDs, also part of the endocardial cushion defect spectrum, are located in the posterior portion of the interventricular septum immediately below the atrioventricular valves (mitral and tricuspid valves) [6]. Echocardiographically, these two valves tend to be located at the same level, however the normal insertion of the tricuspid valve is typically a few millimeters distal. These defects are large and generally do not close spontaneously. Multiple configurations of the atrioventricular valves can occur, the details of which are outside the scope of this text. Endocardial cushion defects represent defects in the separation of the right and the left heart chambers, and can have complete absence of the septa, one common atrioventricular valve, and an ostium primum ASD, among other abnormalities.

Muscular defects, approximately 5–20 % of all VSDs, occur centrally or apically in the trabecular portion, and can have multiple openings ("swiss cheese" appearance). Apical VSDs may occur after myocardial infarctions [7]. Color flow Doppler is invaluable to detect multiple defects in the muscular septum.

Outlet VSDs are also known by several terms: supracristal, infundibular, doubly committed, or subarterial VSDs. Irrespective of the nomenclature used, they occur in the region just below the aortic and pulmonic valves and can have associated

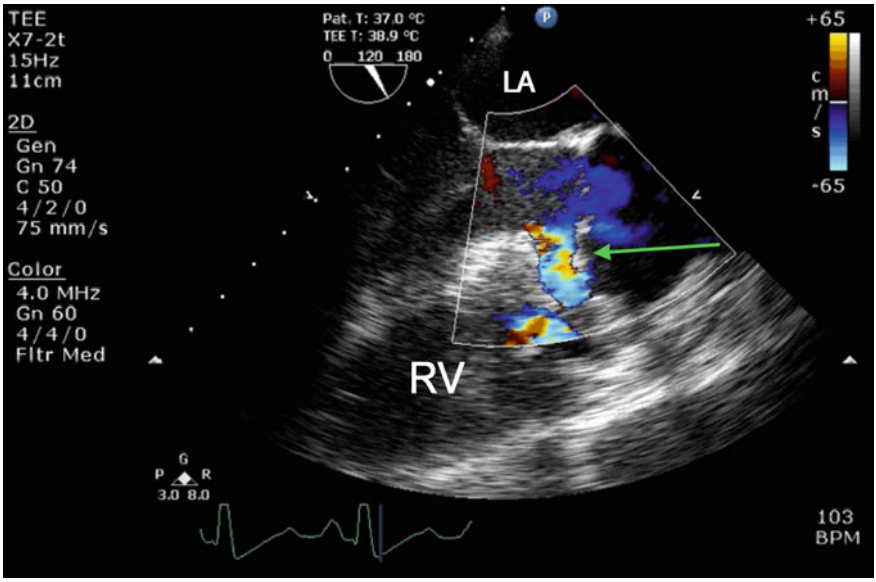

Fig. 13.9 Midesophageal aortic valve long-axis view in a patient with an outlet VSD (*green arrow*). *LA* left atrium; *RV* right ventricle

aortic insufficiency (related to the herniation of the right coronary cusp). Interrogation of the outflow tracts side by side, done as a modification of the ME aortic valve LAX view or the ME right ventricular inflow-outflow view, is used to detect these defects (Fig. 13.9; Video 13.7).

Echocardiographic Examination of VSDs

Ventricular Septal Defects

2D	• Echogenic defect of tissue in interventricular septum – ME Four Chamber View (Muscular and Inlet) – ME AV SAX View (Perimembranous and Outlet) • Associated Findings – Atrial Enlargement – Ventricular dilation
CFD	• Presence of Interventricular flow – May or may not be turbulent (dependent on VSD size)
Spectral	• Calculate Pulmonary to Systemic Flow Ratio (Qp:Qs) • Estimate Pulmonary Arterial Systolic Pressure (TR Jet)

The complexity of the interventricular septum requires multiple views as well as rotation and use of the omniplane angle at nonstandard imaging planes. The ME four chamber, ME aortic valve short axis and long axis, ME RV inflow-outflow and deep transgastric long axis views are recommended for a focused interrogation. Apart from number, size, location, and nature of defects, other pertinent findings to look for include: additional congenital heart disease, aortic valve abnormalities, signs of RV pressure and volume overload, and its functional consequence. Doppler interrogation can quantify the nature and magnitude of the intracardiac shunt, estimate valvular regurgitation, and estimate PA systolic pressure (see Chap. 3).

High velocity through the defect as evidenced by Doppler interrogation is indicative of a restrictive shunt whereas low, nonturbulent flow denotes a nonrestrictive defect. Generally, a nonrestrictive defect indicates a more severe lesion [8, 9]. The ratio of pulmonary to systemic blood flow (Qp/Qs) should be measured since it has diagnostic and therapeutic implications. A high Qp/Qs indicates that there is a significant left to right shunt which may eventually lead to pulmonary overcirculation and Eisenmenger's syndrome. A low Qp/Qs is indicative of a right to left shunt.

Persistent Left Superior Vena Cava (SVC)

Approximately 0.5 % of the population has a persistent left SVC, which drains into the coronary sinus 90 % of the time. It is uncommonly associated with an absent right SVC. In embryologic development there are two superior vena cavae. Normally the left-sided SVC regresses with blood from the internal jugular (IJ) and left subclavian returning to the heart via the innominate vein. In the setting of a persistent left SVC, the left IJ and subclavian typically return blood flow to the heart via the left SVC into the coronary sinus. In its presence, central venous cannulation and pacemakers can take an abnormal orientation. In cardiac surgery, retrograde cardioplegia can be ineffective and venous cannulation strategies may need to be adjusted.

The coronary sinus (CS) can be imaged by advancing the probe from a ME four chamber view, in a modified bicaval view or in the posterior AV groove in the ME two chamber view. Dilation of the coronary sinus (>10 mm in diameter) should arouse suspicion for a persistent left SVC (Fig. 13.10; Video 13.8). Other causes such as elevated right atrial pressures from heart failure, atresia, or stenosis of the ostium or a coronary artery fistula to the CS need to be ruled out. The suspicion can then be confirmed with injection of agitated saline into the left upper extremity and resultant coronary sinus opacification. A large coronary sinus and its drainage into the right atrium have been mistaken for an ASD in some patients, and this makes thorough imaging essential [10].

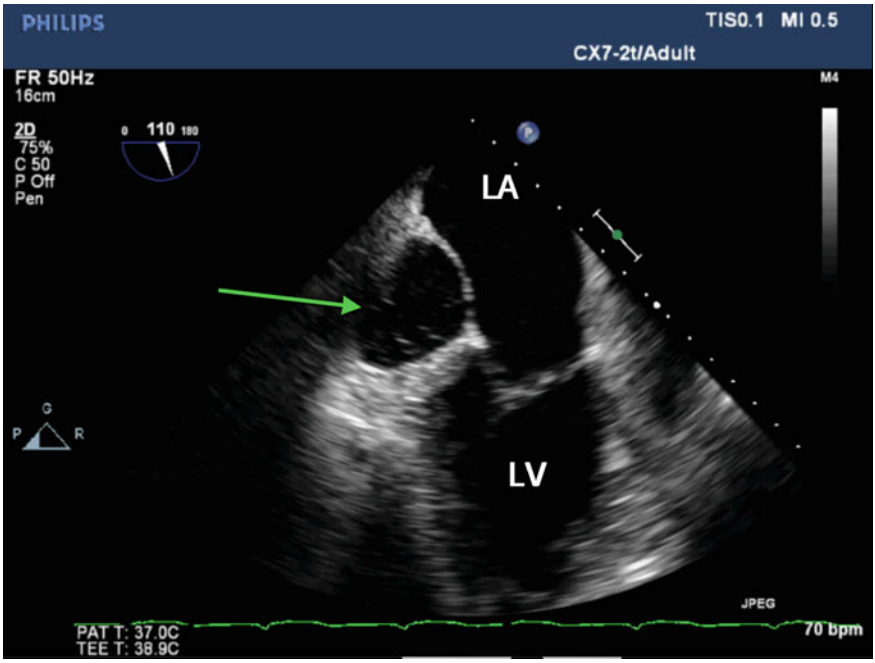

Fig. 13.10 Midesophageal two chamber view with the dilated coronary sinus in cross section to the posterior aspect of the top of the left ventricle. Agitated saline injected into the left arm has resulted in "microbubbles" within the dilated coronary sinus confirming a persistent left superior vena cava

Patent Ductus Arteriosus (PDA)/Aorticopulmonary Window

The ductus arteriosus is a vascular communication between the proximal descending aorta and the main or the left pulmonary artery in its roof. It closes spontaneously after birth but can persist into adulthood in rare cases, causing a left to right shunt. It is usually a coincidental finding picked up due to a murmur that leads to an echocardiography exam [11]. Echocardiography is helpful to not only diagnose the lesion, but also to evaluate the shunt magnitude and volume load, estimate pulmonary artery pressures, and identify associated cardiac pathology. Some patients may present with endarteritis (endocarditis of the ductus), which is responsible for almost half of the deaths in adult patients with a PDA [12]. The pulmonary side of the ductus is more commonly the site of infection. The patent ductus can be closed either surgically or often with a transcatheter device with excellent results. Upper esophageal views have been used to visualize a PDA, with nonstandard orientation of the omniplane angles. Since the connection between the aortic isthmus and the main pulmonary artery hides behind the left main bronchus,

it is often difficult to visualize with TEE. The demonstration of flow abnormality in the pulmonary artery using color flow Doppler is excellent supportive evidence but not diagnostic by itself of a PDA. Proper parallel alignment of the Doppler beam with the flow is even more difficult. It is important to note the FiO_2 during the shunt calculation since hyperoxia leads to reduction in PA pressures and an increase in the shunt [12]. The aorticopulmonary window represents a more proximal communication between the ascending aorta and the pulmonary artery, and can be easier to pick up on TEE. Hemodynamic consequences tend to be similar to a PDA.

Tetralogy of Fallot (ToF)

Tetralogy of Fallot

2D	• Pulmonic Stenosis – Narrowed RVOT in ME RV inflow-outflow view • Right Ventricular Hypertrophy – Measure in ME Four Chamber view • Overriding Aorta – Observed in ME LAX view • Ventricular Septal Defect – Typical Perimembranous VSD in ME AV SAX View
CFD	• Presence of Interventricular flow – May or may not be turbulent (dependent on VSD size) • Turbulence in RVOT / Pulmonary Artery
Spectral	• Calculate Pulmonary to Systemic Flow Ratio (Qp:Qs) • Estimate Pulmonary Arterial Systolic Pressure (TR Jet)

Classic ToF patients manifest a VSD, pulmonic stenosis, an overriding aorta, and RV hypertrophy. Addition of an ASD makes it a pentalogy (present in about a third of cases). Multiple other congenital cardiac lesions can accompany a ToF, such as a right aortic arch, systemic venous abnormalities, and LVOT obstruction among others. Most patients require surgery early due to the cyanosis and right to left shunt, and thus patients that survive into adulthood generally have little RV obstruction. The goals of echocardiography should include (apart from confirming the diagnosis): quantification of the RV obstruction, VSD shunt magnitude and direction, and detection of associated anomalies.

The large, perimembranous VSD is best seen in the ME aortic valve LAX or SAX views. The defect is located between the right and the non-coronary cusps of the aortic valve. This generally permits shunt interrogation by Doppler (either color flow or spectral). The entire septum should be carefully interrogated to rule out additional defects. The ME aortic valve LAX view can also demonstrate the aortic override, which can be variable in presentation. In the post-repair adult, the repair of

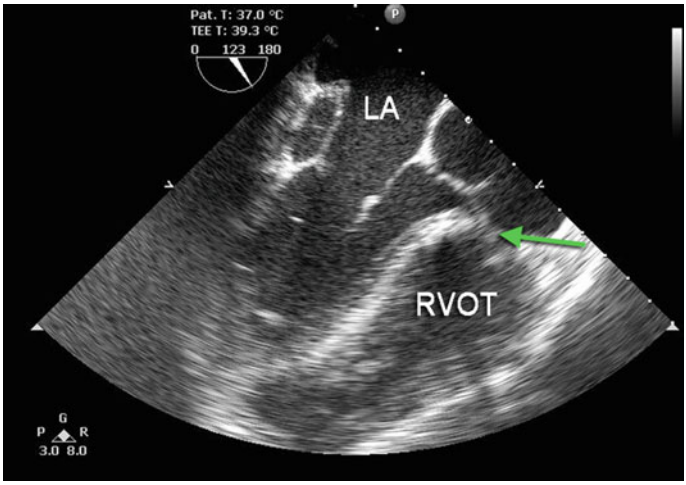

Fig. 13.11 Midesophageal long-axis view in a patient with tetralogy of fallot s/p VSD repair. Note the overriding aorta (*green arrow*) that remains and the evidence of right ventricular hypertrophy. Incidentally there is a dilated coronary sinus in this patient that was a persistent left-sided superior vena cava. *LA* left atrium; *RVOT* right ventricular outflow tract

the VSD using a patch still permits identification of the override (Fig. 13.11; Video 13.9). Involvement of the aortic valve, either as part of the primary lesion or during repair needs to be carefully ruled out.

Manipulation of the omniplane angle from either the ME aortic valve LAX or ME right ventricular inflow-outflow view can be used to interrogate the RV outflow tract. Obstruction is suggested (using color flow Doppler) by the presence of aliasing and turbulence in the RVOT. RV wall thickness can be measured in the ME right ventricular inflow-outflow to quantify the hypertrophy while transgastric views can be used to examine the RVOT. These views can help detect abnormalities of the pulmonic valve, if present. Interrogation of the RV outflow and the pulmonary artery with two-dimensional echocardiography, color flow Doppler, and spectral Doppler is helpful after repair since stenosis can persist and indeed, even worsen as the patient continues to grow into adulthood. After repair, left and right ventricular outflow obstruction and valvular regurgitation remain as the focus. Assessment of RV systolic pressure, size, and function is vital to the exam. Tricuspid and pulmonic regurgitation is not uncommon, and their quantification can be useful for future comparison.

Conclusion

With more congenital heart disease patients now surviving longer, it is likely that these patients will ultimately need further anesthesia for non-cardiac and cardiac procedures. Echocardiography has remained the cornerstone of diagnosing and managing these conditions. The basic perioperative echocardiographer should be familiar with and able to recognize simple congenital heart lesions. Complex lesions should involve consultation with an advanced echocardiographer. Perioperatively, TEE provides useful information about the real-time monitoring of ventricular filling, myocardial performance, and identification of intracardiac shunting, in addition to optimization of hemodynamic management strategies.

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