REVIEW ARTICLE

A Review of Selected Adult Congenital Heart Diseases Encountered in Daily Practice

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Abstract: The advancement in corrective surgical procedures and anaesthesia technology has resulted in the increased survival of patients with Congenital Heart Diseases (CHD). Most of the surviving CHD patients have successfully reached adulthood and those surviving adults now outnumber the infants born with the CHD. Unfortunately, the surviving adults with CHD do not get proper care due to either inconsistent follow-up or not getting care from a specialist in the field of CHD. It is imperative for general practicing clinicians to be aware of the congenital diseases as well as the current clinical recommendations. This manuscript reviews some of the common congenital diseases seen in adults such as cardiac shunts, left heart obstructive lesions, and aortopathies.

Keywords: Atrial septal defect, ventricular septal defect, patent ductus arteriosus, bicuspid aortic valve, coarctation of aorta, aortopathies.

1. INTRODUCTION

Birth defects related to Congenital Heart Disease (CHD) are the most prevalent form of congenital diseases, with a reported prevalence between 0.8 - 1.0% of live births worldwide (8 cases per 1000 live births) [1]. Since the introduction of cardiopulmonary bypass procedure in 1950s and subsequent advancements in diagnostic, surgical and interventional techniques have translated into a significant improvement in morbidity and mortality of CHD patients [2, 3]. This is reflected in CHD demographics as the number of adults living with CHD now outnumbers the pediatric CHD population. This focused review on selected common acyanotic lesions (shunts, left heart obstructive lesions and related aortopathies) and post-operative tetralogy of Fallot which is the most common cyanotic heart disease that presents to adult cardiologists following repair. The review aims to address common problems faced by the general cardiologist caring for the adult patient with CHD and provides updated key guidelines based on recommendations for one of the fastest-growing populations in cardiology. The 2008 ACC/AHA and 2018 ACC/AHA adult congenital guidelines were reviewed and where relevant the 2018 guidelines with grade of recommendation and strength of evidence are referenced in parenthesis for each lesion.

2. CARDIAC SHUNT LESIONS

2.1. Atrial Septal Defects

Atrial Septal Defect (ASD) is the second most common congenital heart defect, accounting for 10-15% of all CHD cases. As the name implies, congenital ASD is a defect in the development of the atrial septum resulting in a communication between the two upper chambers of the heart (atria) [4]. Another form of ASD is the acquired variant (Iatrogenic ASD), most commonly due to trans-septal percutaneous punctures, which occurs during procedures like mitral balloon valvuloplasty or mitral clip. Congenital ASD often presents as an isolated finding, or in approximately 30% of cases, associated with other malformations [5]. The following are the most common types of ASD.

- Secundum ASD see (Fig. **1A-C**, **video 1**) is the most common subtype, occurring in 75% of ASD cases. It is found at the level of the fossa ovalis. It can be associated with valvular Pulmonic Stenosis (PS) and Bicuspid Aortic Valve (BAV).
- Primum ASD see (Fig. **2**) is the second most common subtype of ASD, occurring in 15 - 20% of cases, located anteriorly and inferior to the fossa ovalis. The primum ASD is a type of Atrio-Ventricular Septal Defect (AVSD) and is commonly accompanied by a cleft mitral valve involving the anterior mitral leaflet.
- Sinus venosus ASD (Fig. **3**) occurs in 5-10% of cases, with two subtypes, superior and inferior.
  - The superior type is more common and is located near the superior vena cava entry.
  - The inferior type is less common and is located near the inferior vena cava entry.

Anomalous right pulmonary vein drainage is a common association with sinus venosus type ASD.

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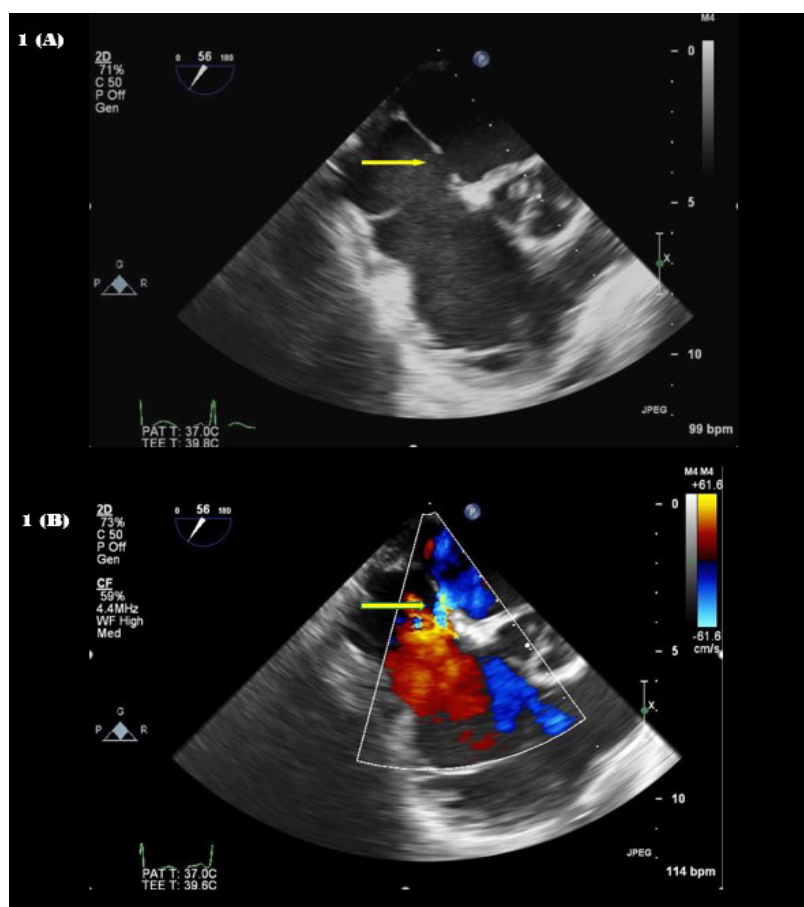
Fig. (1A and B). Secundum ASD demonstrated on trans-esophageal echocardiogram in both 2D (**1A**) and color modes (**1B**). Visualization of the entire intra-atrial septum can be challenging by transthoracic examination, and TEE study may be required. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
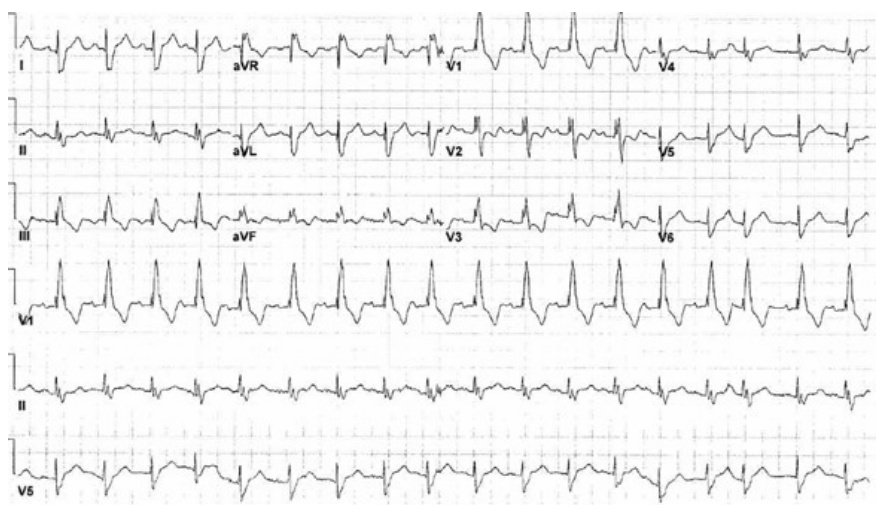
Fig. (1C). 12 lead EKG of same patient showing sinus rhythm, complete right bundle branch block and rightward axis. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
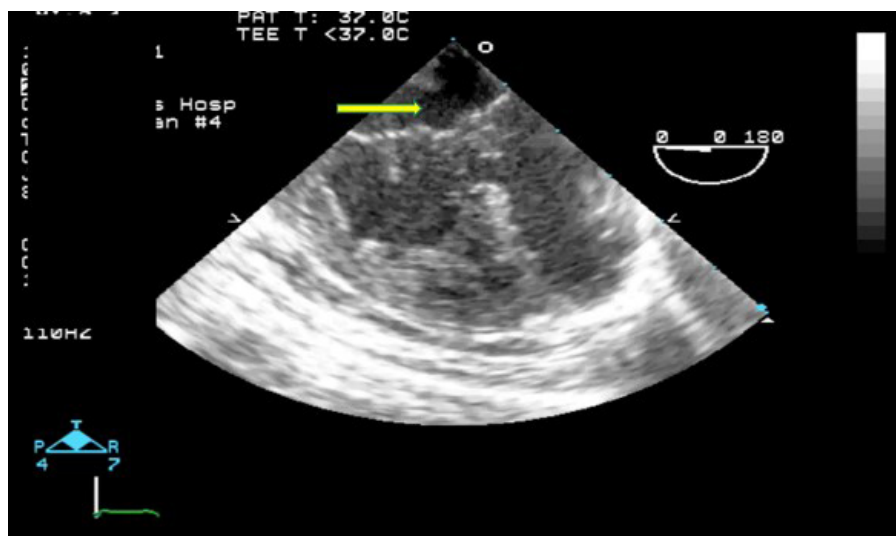
Fig. (2). Complete AV canal defect with Septum primum ASD (arrow),VSD and malalignment of atrioventricular leaflets seen on transesophageal echocardiogram (TEE). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
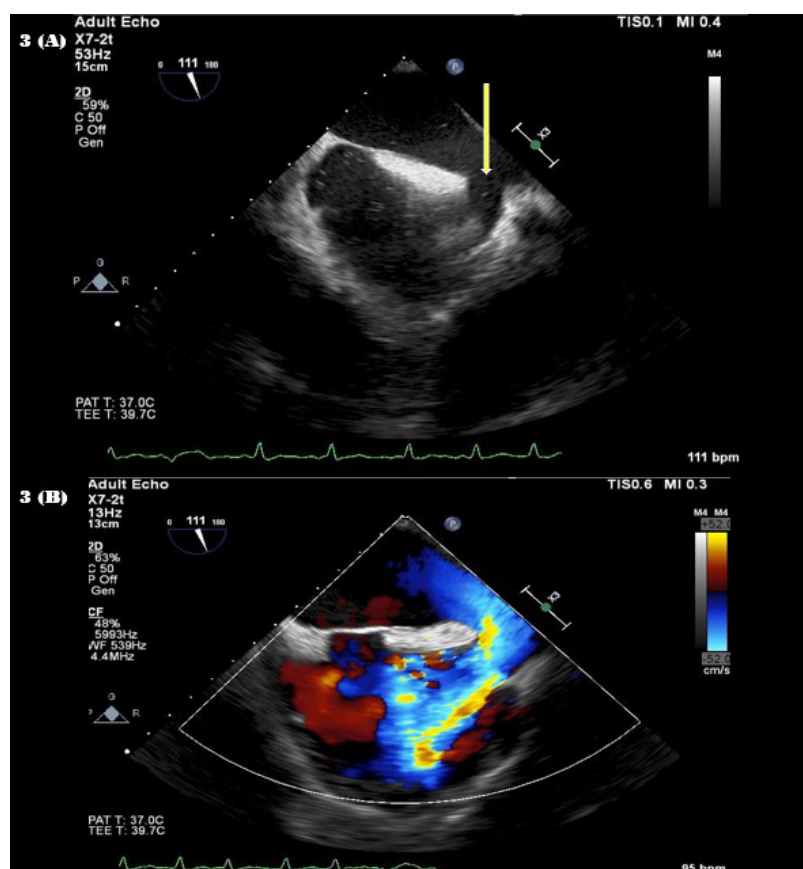
Fig. (3). Sinus venosus ASD (superior type; arrow) is shown. There is a missing tissue at right atrial to superior vena cava junction as shown in bicaval view of tranesophageal echocardiogram (**3A**). Bicaval view with color doppler is shown. Color flow can be seen from left atrium (upper chamber) towards right atrium (inferior chamber) (**3B**). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

- Coronary sinus ASD is uncommon, occurring in less than 1% of cases. There are two types, partial and complete unroofing of the coronary sinus. It can be associated with persistent left superior vena cava and partial or total anomalous pulmonary venous connection.

Unrepaired ASD can result in left to right shunting and eventually lead to Right Ventricular (RV) volume overload and excess pulmonary blood flow. Patients can develop symptoms of right-sided heart failure such as fatigue, palpitations, exercise intolerance and frequent pulmonary infections [6]. Atrial fibrillation, atrial flutter and sick sinus syndrome are common atrial arrhythmias, resulting from long-standing right atrial and ventricular volume and pressure overload. The initial complaint of most unrepaired ASDs is breathlessness and palpitations. Sometimes ASD is discovered incidentally on chest x-ray (RV and Right Atrial (RA) enlargement, prominent pulmonary vascularity), murmurs auscultated in the pregnant patient, or possibly after a paradoxical embolic event.

When performing diagnostic workup of a patient with the possibility of ASD, it is important to determine the presence, size, location of the ASD, and any other associated lesions. The clinical examination may unveil the presence of a fixed split of the second heart sound, precordial lift, or a systolic pulmonary flow murmur. In the case of larger shunts, there may be a mid-diastolic flow-related rumble across the tricuspid valve.

ECG findings may show:

- Right-axis deviation, incomplete/complete right bundle-branch block, and right atrial enlargement (secundum ASD)(Fig. **1C**).
- Superior left axis deviation (primum ASD).
- Abnormal P-wave axis (sinus venosus ASD - superior type).
- Familial ASD may present as a complete heart block and can be associated with skeletal abnormalities.

Holt-Oram syndrome and down syndrome are known to have some association with ASD. It is prudent for the clinician caring for the patient with ASD to conduct thorough family history and screen for possible manifestations in family members.

Transthoracic echocardiography is extremely helpful in diagnosing and providing a complete characterization of ASD including type, location, dimensions, and number of ASDs. Visualization of the entire Inter-Atrial Septum (IAS) can be challenging in some patients and therefore it is recommended to obtain multiple imaging views including the parasternal, apical, and subcostal windows in addition to color Doppler examination. Septal dropout on Trans-Thoracic Echocardiography (TTE) can result in the false-positive diagnosis of ASD. Contrast (agitated normal saline) echocardiography can be utilized to determine the presence of a true shunt and can confirm a right-to-left shunt. If a negative contrast sign is present in the right atrium, this can identify the

presence of a left-to-right shunt. In some instances, a Trans-Esophageal Echocardiogram (TEE) is needed, particularly for the superior type sinus venosus ASD [7]. If TTE imaging reveals unexplained RV volume overload, a TEE or other advanced cardiac imaging modality can be used to fully evaluate the atrial septum and pulmonary veins [8]. 3D echocardiography imaging is a powerful tool in specifying the measurements of septal rims around the defect, especially if intervention is planned.

Uncomplicated ASDs with adequate non invasive imaging do not typically require diagnostic cardiac catheterization. Invasive workup is typically reserved for when there is concern about underlying coronary artery disease, and to assess Pulmonary Vascular Resistance (PVR) and reactivity in patients with Pulmonary Arterial Hypertension (PAH) [9].

Therapy is not required in asymptomatic patients with small ASDs with a diameter of less than 5 mm and with no evidence of RV enlargement or PAH. Serial echocardiograms can be performed every 2 - 3 years [10] to monitor for changes in RV function, size, and pulmonary pressure. In the case of a small ASD with paradoxical embolism, closure of the ASD is indicated. Large defects with associated RV volume overload typically meet the criteria for closure to prevent long-term complications [10].

Primum, sinus venosus and coronary sinus ASDs are not amenable to percutaneous catheter intervention, and therefore surgical closure is recommended. Uncomplicated secundum ASDs with appropriate margins are closed with a percutaneous catheter intervention in the majority of the cases [11]. High-quality non-invasive imaging and more specifically 3-dimensional TEE seems integral and important to verify a rim of 5 mm and diameter < 3.8 cm for appropriate device closure. Catheter-based interventions have exceptionally good outcomes, with < 1% of patients experiencing serious complications [12]. Surgical intervention is also an option when device closure is not feasible. In the pediatric and adolescent patient population without significant co-morbidities, surgical outcomes are excellent with <1% mortality [13]. Mortality rates increase with patients who are elderly or have significant co-morbidities.

The postoperative visit should screen for symptoms of chest pain, arrhythmia, or embolic events. TTE examination is performed 24 hours, 1 month, 6 months, and 1-year post-procedure to track device position, formation of residual shunting, thrombus formation, and pericardial effusion. Following surgical repair, cardiac tamponade may occur at any time postoperatively for several weeks. Chest or abdominal pain, emesis, fever, and fatigue may represent early signs of cardiac tamponade, emphasizing the need for close follow up. Infective endocarditis prophylaxis is only indicated for the first 6 months post-procedure [10].

Some important updates compared to 2008 guidelines are that closure of the ASD for right heart enlargement in the absence of symptoms is currently not a Class 1 recommendation anymore and has received a Class IIA recommendation in 2018 guidelines. However in ASDs with net left to

right shunt > 1.5: 1 with reduced functional capacity, absence of cyanosis at rest or with exercise, pulmonary artery systolic pressure < 50% of systemic pressures and pulmonary vascular resistance < 1/3 of systemic resistance closure is recommended (Class 1B; 2018 guidelines). As stated above, key aspects to focus for decision making in the closure of ASD emphasized in the 2018 guidelines include pulmonary vascular resistance < 1/3 of systemic vascular resistance, pulmonary artery systolic pressure < 50% systemic, right heart enlargement and a physiologic significant shunt (≥ 1.5: 1). Abnormal invasive right heart hemodynamics requires careful multidisciplinary discussion with adult congenital experts with regards to the safety of the intervention and in patients with established Eisenmenger physiology referral to pulmonary hypertension specialist for vasodilator therapy is important, given favorable outcomes of vasodilator therapy in this subgroup. Importantly this has received the only Class 1A recommendation in the 2018 guidelines and is applicable to Eisenmenger physiology with any intracardiac septal defect for use of bosentan in conjunction with other vasodilators. Also new in the 2018 guidelines are formal recommendations on the assessment of pulmonary venous anatomy by cardiac MRI, cardiac CT or TEE given the importance of preprocedural knowledge of anomalous anatomy which may alter the percutaneous versus surgical approach.

Atrial arrhythmias are prevalent in patients with ASD. There is decrease in the incidence of arrhythmias with timely closure of hemodynamically significant ASDs and it also decreases the arrhythmia burden in patients who have already developed arrhythmias. These patients remain at high risk for atrial arrhythmias during their lifetime as compared to the normal population. It remains unclear if there is a subgroup of these patients that may be benefited from prophylactic electrophysiological procedures. However, early diagnosis and documentation of arrhythmia are pivotal for long term prognosis as some degenerate into complex arrhythmias. Holter monitoring or implantable loop recorder can be immensely helpful to aid in early diagnosis of potential malignant arrhythmias [14].

2.2. Ventricular Septal Defect

Ventricular Septal Defects (VSDs) are the most common congenital heart defects as an isolated finding or in combination with other defects, with an incidence of 3 - 3.5 infants per 1000 live births [15]. The incidence decreases in older infants and adults, as a significant number of small VSDs spontaneously close [16]. The direction and severity of shunting are influenced by the size of the defect, PVR, left and right ventricular function, and the presence of RV outflow tract obstruction.

There are 4 VSD subtypes that have many synonyms in the literature [17].

- Type 1 VSDs (supracristal) account for 6% of defects in non-Asian patients but occur in up to 33% in Asian populations [18]. Type 1 VSDs lie in the out-

flow portion of the RV and spontaneous closure is uncommon.
- Type 2 VSDs (perimembranous) are the most common subtype accounting for 80% of cases. The defect is a deficiency at the level of membranous septum adjacent to the septal leaflet of the tricuspid valve. (**video 2**)
- Type 3 VSDs (inlet) (Fig. **4**) are found at the inlet of ventricular septum immediately inferior to the AV valve apparatus. There is an association between inlet VSD and Down syndrome.
- Type 4 VSDs (muscular) account for up to 20% of VSDs in infants and are surrounded by septal myocardium. Muscular VSDs can be located centrally, apically, or at the margin of the septum and RV free wall. They can be single or multiple in numbers and spontaneous closure is common, therefore incidence is much lower in adults [16, 17].
- VSDs are a common component of complex congenital heart abnormalities, including conotruncal defects such as tetralogy of Fallot and transposition of the great arteries. There can also be an association between VSDs and left-sided obstructive congenital abnormalities such as subvalvular AS and coarctation of the aorta. Type 1 VSDs can be associated with aortic cusp prolapse (usually the right and sometimes the non-coronary cusp) through the defect that can result in progressive aortic regurgitation.

There are a variety of clinical presentations for adult patient with unrepaired VSD. Possible presentations include:

- A small VSD with a small left-to-right shunt, with normal pulmonary pressures and LV volume that was observed throughout childhood.
- An asymptomatic patient with a presumed innocent murmur.
- VSD with a left-to-right shunt, elevated pulmonary pressures, and LV volume overload.
- Infectious endocarditis causing fever and bacteraemia.
- Aortic valve prolapse causing a diastolic murmur of Aortic Regurgitation (AR).
- Large VSD with the transition from large left- to-right shunt with severely elevated pulmonary pressures and reversal of shunt to right-to-left in association with cyanosis and exercise intolerance.

Small VSDs are 25% or less than the diameter of the aortic annulus having small left-to-right shunt with normal pulmonary pressures (restrictive) and without evidence of LV volume overload. Spontaneous closure of small VSDs most often occurs during infancy but can happen at any time [19]. Adults with small, restrictive VSD can be followed clinically with symptom check, physical exam, and periodic echocardiography as clinically indicated. Moderate sized VSDs are between 25% and 75% of the aortic diameter, with small to moderate left-to-right shunt, mild PAH, and mild to moderate LV volume overload. Defects larger than 75% of the
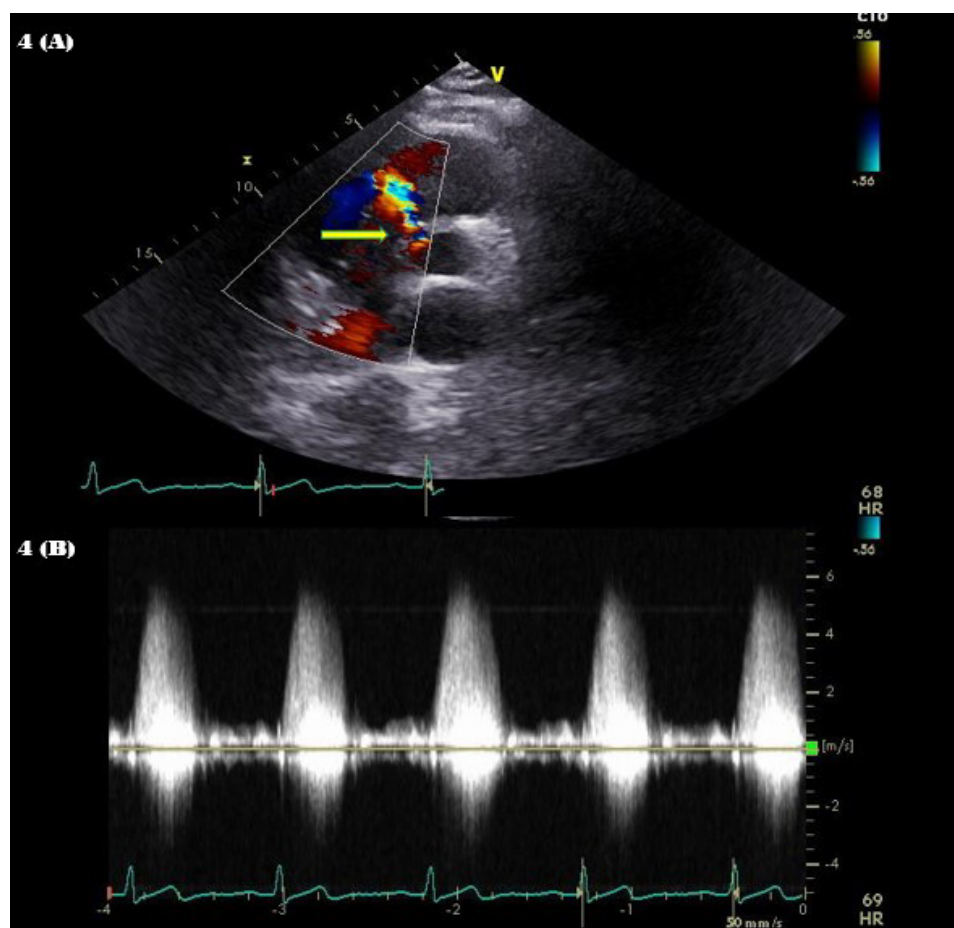
Fig. (4). Perimembranous VSD (arrow) with color doppler is shown (parasternal short axis view in transthoracic echocardiogram). There is color flow from high membranous septum into the right ventricle (**4A**). Figure (**4B**) is showing continuous wave doppler across VSD with peak velocity as shown. This can be used to calculate the right ventricular systolic pressure (using modified Bernoulli equation by calculating trans defect pressure gradient),assuming that the patient's systolic blood pressure at time of exam is similar to left ventricular systolic pressure. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

aortic diameter usually have a moderate to large left-to-right shunt, elevated pulmonary pressures with LV volume overload and are classified as large.

The clinical examination may reveal a holosystolic murmur over the third to fourth left intercostal space or a precordial thrill. ECG may show biventricular hypertrophy or isolated RV hypertrophy in patients with large VSD and moderate to severe PAH. A chest x-ray may be normal or demonstrate left atrial and LV enlargement. Transthoracic echocardiography is the first-line imaging modality in VSD work-up, providing the location, number, size, estimation of pulmonary pressures and estimation of LV volume overload. In the case of Type 1 (supracristal) VSDs, careful examination of the aortic valve must be performed to screen for AR due to the prolapse of the Right Coronary Cusp (RCC) or Non-Coronary Cusp (NCC). TEE may be valuable in adult patients with poor windows by the TTE examination. Cardiac Computed Tomography (CT) or Magnetic Resonance (MR) techniques are also available to define the severity of the de-

fect should the TTE prove to be non-diagnostic, or in the presence of VSD with other complex congenital heart defects. When pulmonary artery pressures are elevated by echocardiographic assessment, cardiac catheterization is performed to determine the PVR. Vasodilator testing should be utilized to evaluate the reversibility of PAH. When the Qp/Qs > 1.5, VSD closure is generally recommended [10].

Surgical repair by a surgeon with expertise in CHD is the treatment of choice and can be performed with low mortality (1-2%) and good long-term results. Patch closure can be performed with synthetic material such as Dacron or Gore-Tex, or in rare instances by primary closure. Intraoperative TEE is an important tool that may reveal the presence of an associated VSD after the closure of the dominant VSD. Transcatheter device closure of VSD can be performed for residual defects after a prior attempt of surgical repair, trauma, patients that are considered high risk for surgery, restrictive VSDs with a significant left-to-right shunt, or VSDs that are difficult to close surgically. In Type

2 (perimembranous) VSDs, percutaneous closure is challenging due to the proximity of the conduction system, aortic, and tricuspid valves.

The surgical complication rate is nearly 11%, most commonly in the form of arrhythmias and conduction abnormalities [20]. Patients can develop transient bi-fascicular or tri-fascicular block that can lead to a complete heart block. These patients should be screened with an ECG annually in addition to ambulatory monitoring and exercise stress testing.

If there is no residual VSD or associated lesions patients do not have to follow up with an ACHD center and can return to the care of their primary general cardiologist. Patients with residual heart failure, shunting, elevated pulmonary pressures, and significant valvular heart disease are advised to follow up once a year at a specialized ACHD center. Adults who have undergone transcatheter device closure of a VSD can follow up every 1 to 2 years. If there is a small residual VSD without evidence of other lesions, follow up is advised every 3 to 5 years [10].
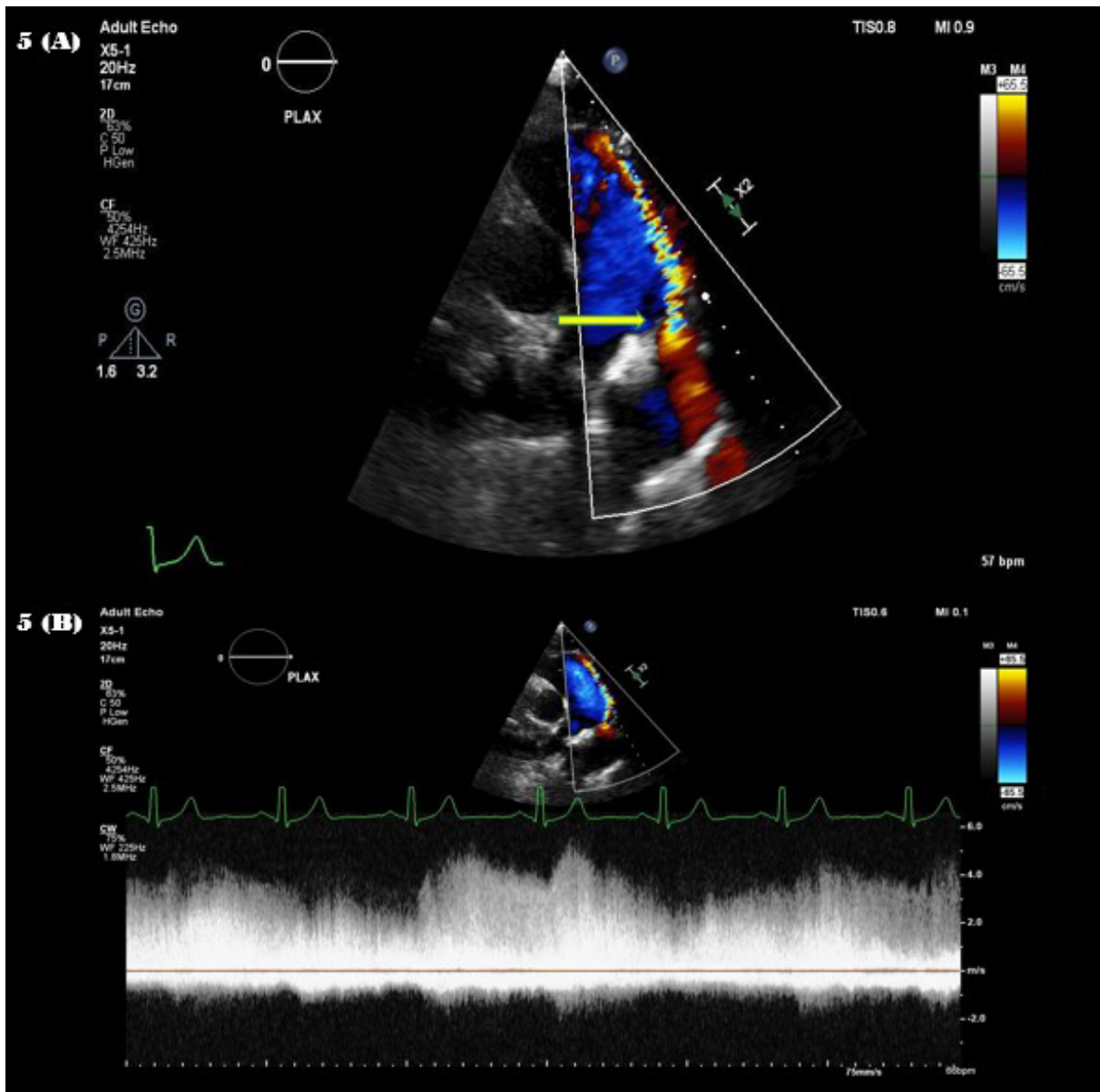
Fig. (5A and B). Modified parasternal short axis view showing aorta and pulmonary artery bifurcation. This is an excellent view for visualizing the PDA originating from the upper descending aorta to the left pulmonary artery by color flow (**5A**). Continuous wave Doppler examination showing the left to right shunt pattern throughout the cardiac cycle (continuous flow), more prominent in systole, which characteristic of PDA (**5B**). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
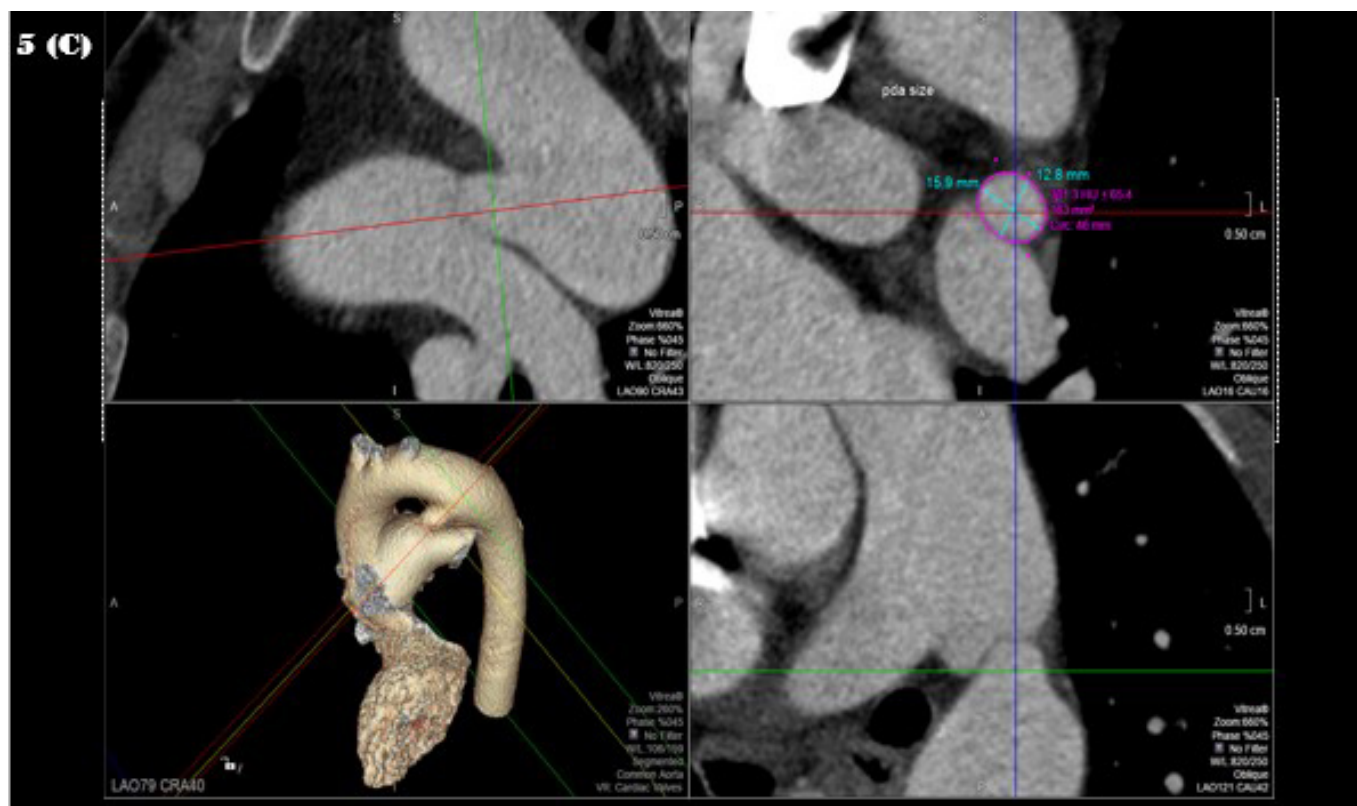
Fig. (5C). This is multiplanar reconstructions of the PDA by gated cardiac CT scan, which can provide precise measurement of the defect for percutaneous closure planning. Also shown is the 3-dimensional rendering (color image) delineating the PDA. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

Key aspects to focus for decision making in the closure of VSD is similar in many ways to ASD and include pulmonary vascular resistance < 1/3 of systemic vascular resistance, pulmonary artery systolic pressure < 50% systemic, and a physiologic significant shunt ($\geq$ 1.5: 1) and specific to VSD is the presence of aortic valve prolapse and or history of infective endocarditis. Abnormal invasive right heart hemodynamics requires careful multidisciplinary discussion with adult congenital experts with regards to the safety of the intervention and in patients with established Eisenmenger physiology, referral to pulmonary hypertension specialist for vasodilator therapy is important given favorable outcomes of vasodilator therapy in this subgroup.

Most patients with small restrictive VSD do well with periodic observation, although there is a small risk of infective endocarditis. Supracristal VSDs are at risk of developing aortic cusp prolapse leading to progressive aortic regurgitation which may impact the decision for earlier intervention despite having a restrictive VSD [10].

2.3. Patent Ductus Arteriosus

A patent ductus arteriosus is the result of a persistent connection between the proximal left pulmonary artery and descending aorta (see Fig. **5a**). It can be an isolated finding or associated with other CHD lesions such as ASDs or VSDs.

Left- to- right shunting often leads to fatigue, dyspnea, and LV volume overload. Clinical examination in the setting of a moderate to large PDA may reveal continuous machinery like murmur, most prominently auscultated at the left infraclavicular region. Clubbing and differential cyanosis are signs of a large non-restrictive PDA with Eisenmenger physiology. Patients are at elevated risk for endarteritis and congestive heart failure. A key aspect as part of the examination is an accurate assessment of oxygenation in hands and feet with pulse oximetry (Class 1C; 2018 guidelines). As some patients with left- to- right shunts can have exercise-induced desaturation, it is important to perform assessment at rest and with ambulation.

ECG examination in patients with left –to- right shunting may show left atrial enlargement and Left Ventricular Hypertrophy (LVH). If that patient has elevated pulmonary artery pressure, then RVH may also be seen. Echocardiography with color Doppler imaging can help to confirm the diagnosis of PDA (see Fig. **5a**, **video 3**). Echocardiographic continuous wave Doppler examination will show continuous left- to- right shunt pattern which is commonly seen in PDA (see Fig. **5b**). If the echocardiogram is non-diagnostic, advanced imaging with cardiac MRI or gated cardiac CT can be particularly useful to delineate the patent ductus (see Fig. **5c**). Cardiac catheterization should be performed if elevated

pulmonary artery pressures are found by echocardiography to estimate the PVR. A chest x-ray may show calcification of the PDA, which is an important finding as they are at increased risk of rupture during surgical repair [21]. Patients with a small PDA without evidence of left ventricular volume overload can be followed every 3 to 5 years [10].

Closure of a PDA is indicated if there is left heart chamber enlargement (atrium or ventricle) attributable to net left-to-right shunt, PA systolic less than 50% systemic, and PVR < 1/3 systemic vascular resistance (Class 1C). Percutaneous catheter closure is performed either with occlusion devices or coils and is the treatment of choice for adult patients with PDA. In selected cases, PDA closure can be done when the net shunt is left to right and PA pressure and PVR are above desired cut-offs determined by cardiac catheterization, after careful multidisciplinary assessment with adult congenital and pulmonary hypertension specialists. Advanced imaging can be immensely helpful in preprocedural planning prior to PDA closure (see Fig. **5d**). PDA closure is contraindicated for patients with net right-to-left shunt with Eisenmenger physiology (patients exhibit differential cyanosis in lower extremities), and these patients require more frequent follow-up and referral to pulmonary hypertension specialists for treatment options. Surgery is indicated only in rare instances when device closure is not feasible due to aneurysm

formation, or if the duct size is too large. A TTE should be performed prior to discharge to verify PDA closure. Patients should be on antibiotics for endocarditis prophylaxis for six months, postoperative therapy can be discontinued after that. Patients with successful PDA closure can then be followed at 5-year intervals. PDA is probably the only congenital lesion where the term "cure" after congenital heart disease applies following successful ligation without any long-term consequences.

3. LEFT HEART OBSTRUCTIVE LESIONS

3.1. Bicuspid Aortic Valve and Stenosis/Regurgitation

Bicuspid AORTIC VALVE (BAV) has an incidence of 1% - 2%, making it one of the most common congenital heart malformations [15]. In BAV there is abnormal cusp formation and fusion between two cusps of unequal size. There are multiple variants that have been described, a fusion of the RCC and LCC is the more common phenotype in >70% of cases [22] (Fig. **6**, **video 4**). Histologically in many patients with BAV, there are abnormalities related to smooth muscles, extracellular matrix, and collagen, similar to Marfan syndrome [23]. Patients may develop significant aortic stenosis or regurgitation during mid-adulthood, with peak surgical interventions performed between 60 - 80 years of age [15].
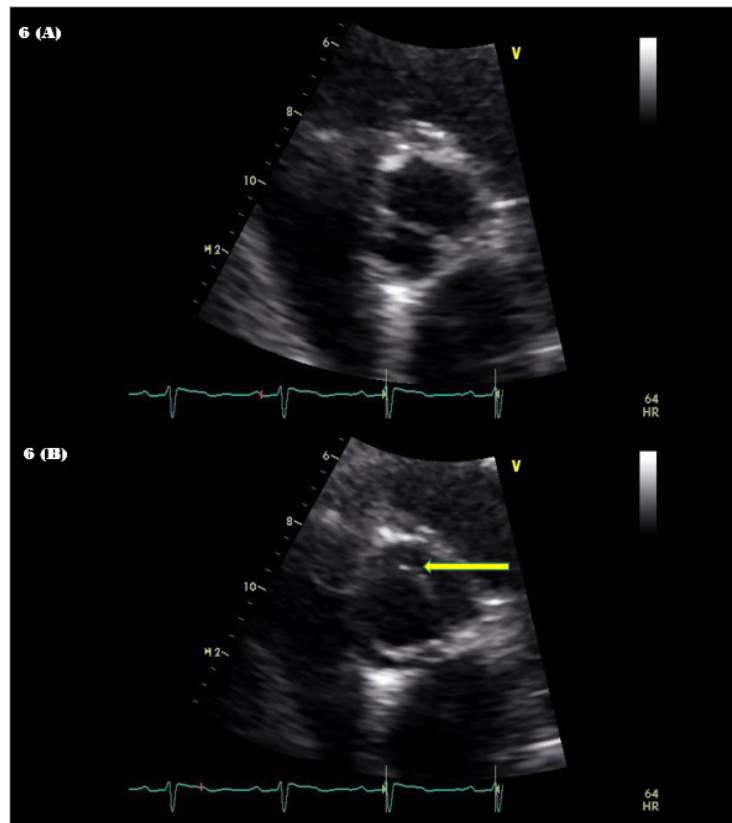
Fig. (6). Parasternal short axis examination at the level of aortic valve by TTE of a patient with bicuspid aortic valve (diastole frame (**6A**) and systolic frame (**6B**)). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

Subaortic stenosis, parachute mitral valve, VSD, PDA, and coarctation of the aorta are all associated lesions with BAV. BAV patients tend to have left dominant coronary artery anatomy [24]. BAV may be found as a part of the Shone complex, which has multiple left heart obstructions (supra-mitral ring, parachute mitral valve, subaortic stenosis, BAV, and coarctation of the aorta). Aortopathy associated with BAV is well described and includes aortic coarctation, progressive aortic root dilation, aortic aneurysm, aortic dissection, and aortic rupture (section 3.0) [25, 26]. Thus, patients with BAV should be screened for coarctation by clinical exam and imaging studies as needed (Class 1B; 2018 guidelines).

Adult patients with unrepaired BAV are at risk of developing Aortic Stenosis (AS) or Aortic Regurgitation (AR) [27]. Standard assessment of AS and AR by echocardiography is paramount to the management of patients with BAV. Systolic doming of the valve leaflets may be seen by echocardiogram. Aortic sclerosis can become evident by the second decade, with progression to calcification by the fourth decade of adulthood in most cases. The course of valvular lesions can be more rapid if there is superimposed acute endocarditis. Hyperlipidemia appears to be a risk factor for the progression of AS in BAV patients [28].

The most common complication of BAV is progressive AS; less than 33% of patients have a functionally normal valve by the fifth decade [29]. Many patients will require valve surgery or percutaneous valvuloplasty. The progression of AS is accelerated in patients with concomitant AR [27]. Several mechanisms may cause the progression of AR in BAV including leaflet fibrosis, leaflet prolapse, leaflet edge retraction, or aortic root dilatation.

Although a specific gene for BAV has not been identified, in 20 -30% of cases there are family members who also have BAV and/or aortopathy with variable inheritance patterns [30]. It is important to conduct a family history, including a history of CHD or aortopathy. Women with Turner syndrome should be evaluated for BAV, coarctation, and aortic enlargement (Class 1B; 2018 guidelines). It is appropriate to image first-degree relatives of BAV for AV disease or aortopathy (Class 11A; 2018 guidelines).

BAV patients frequently have aortic dilation in the ascending aorta, and less frequently in the aortic root at the level of the Sinus of Valsalva. BAV valve morphology appears to influence the incidence of dilation, more frequently in patients with RCC and NCC fusion. Patients with evidence of significant aortic dilatation (> 4.5 cm), a rapid rate of increase in dilation, or a family history of dissection should have aortic imaging at least annually. Stable dilation without significant change in serial imaging and negative family history may be imaged at a longer interval between studies. When TTE does not provide adequate imaging windows, TEE, CT, or MR angiography may be indicated. Four-dimensional flow MR evaluation can help to identify patients at risk for aortic dilation in patients with BAV. Patients with the right-sided eccentric flow in the ascending aorta are more likely to have greater systolic flow angles, higher helical flow, systolic wall shear stress and tend to have larger ascending aorta diameters compared to patients who have BAV without eccentric flow patterns [31]. In patients with Turner syndrome prophylactic root/ascending aortic replacement is recommended when the aortic diameter is 2.5cm/sq m or greater (Class 11A; 2018 guidelines).

Clinical studies have shown that medical therapy is not effective to slow the rate of progression of aortic dilation in patients with BAV. Beta-blocker therapy remains a common practice despite a lack of evidence.

Traditionally bicuspid aortic valve has always been treated with surgical aortic valve replacement. With the dramatic take-over of tricuspid aortic valve stenosis treatment by Transcatheter Aortic Valve Replacement (TAVR), there has been a significant interest in TAVR being an option for patients with BAV. The safety, efficacy, and clinical outcomes of TAVR were studied in patients with bicuspid aortic valve stenosis [32]. At two years of follow-up, the cumulative event rates for all-cause mortality were similar to the tricuspid group [32]. Although TAVR in patients with bicuspid AV was associated with adverse procedural outcomes such as stroke as compared to patients with tricuspid AV, there was no significant difference in procedural success outcomes between two groups with the use of new-generation devices, which is definitely an improvement compared to older studies [32]. This is an area of active research and BAV registries are providing insights into long term outcomes in these patients. BAV TAVR should be performed only after careful consideration of the likelihood of success by centers of expertise using a heart team approach and advanced imaging tools like CT and 3D printing to predict success. Also, there should be an evaluation of stroke risk, given known long-term success for surgical AVR in this population. Furthermore, the presence of aortopathy (aortic dilation) will influence decision making for surgical AVR.

3.2. Subaortic Stenosis

Subaortic Stenosis (SubAS) is a narrowing below the aortic valve either as a discrete fibrous ridge or tunnel-like narrowing and has a reported prevalence of 6.5% in the ACHD population [32, 33]. It can present as an isolated finding or as part of Shone syndrome. Other associated lesions include VSD, AVSD, or conotruncal anomalies. If left unrepaired, SubAS can lead to obstruction or in >50% of cases AR [33]. These patients are also at elevated risk for endocarditis.

On clinical examination, a crescendo-decrescendo murmur without evidence of an ejection click is auscultated over the left parasternal area and the apex. If presenting in adulthood it is commonly confused with hypertrophic obstructive cardiomyopathy. Concomitant aortic regurgitation related diastolic murmur may also be heard related to the turbulent jet damage to the aortic leaflets. ECG may be normal or show LVH depending on the severity of the lesion. A transthoracic echocardiogram is the diagnostic modality of choice. 3D imaging provides an additional tool to image complex LV outflow tract anatomy. TEE may be used pre- and intra-operatively (Fig. **7a** and **7b, video 5**). Cardiac MRI and less often gated cardiac CT may be useful as an adjunctive modality to evaluate the subaortic area when not well delineated in echo studies and to differentiate from hypertrophic cardiomyopathy (Fig. **7c** and **7d**).
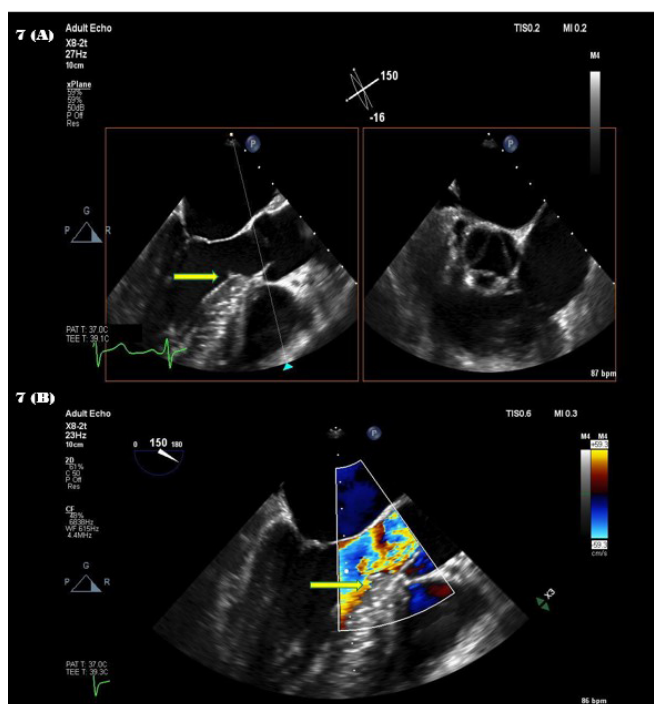
Fig. (7A and B). Discrete subaortic ridge/ subaortic membrane (top image; arrow) noted below the aortic valve annulus (**7A**). Significant flow turbulence (bottom image ; arrow) is visualized by color doppler (**7B**).Note that aortic valve is not stenotic as seen in figure **7A** in short axis view thus not accounting for any of flow turbulence. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
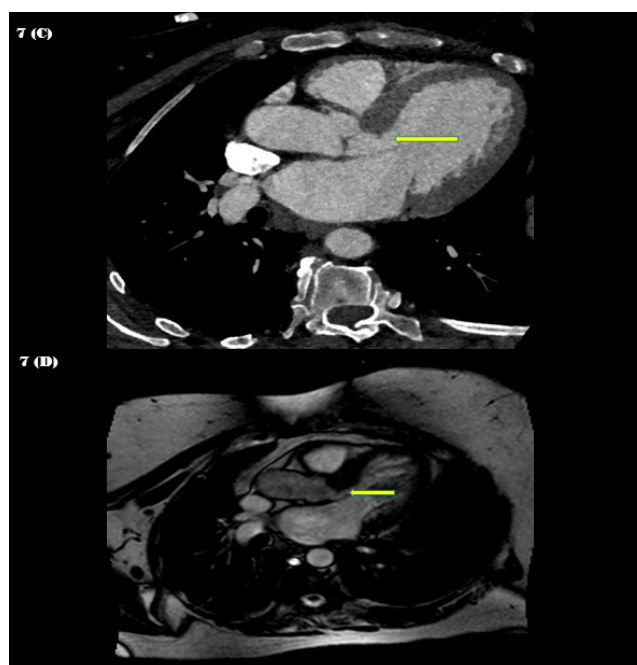
Fig. (7C and D). (**7C**) Showing prominent protuberant subortic ridge in the left ventricular outflow tract with flow turbulence in close proximity to aortic valve on a CT three chamber reconstruction. (**7D**) shows cardiac MRI (steady state free precession cine image) still frame from a patient with elevated LV outflow velocities showing protuberant subaortic membrane. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

If the peak instantaneous gradient is 50 mmHg or a mean gradient is 30mmHg, surgical intervention is advised if patients are symptomatic (Class 1C; 2018 guidelines). This is different from the 2008 guidelines, which recommended intervention regardless of symptoms. If there is a lesser degree of obstruction (peak gradient < 50 mm hg) but with superimposed depressed LV ejection fraction, heart failure or ischemic symptoms, moderate to severe aortic valve regurgitation, planned pregnancy or participation in sports, surgical intervention may be considered (Class 1C ;2018 guidelines). If the maximum gradient is > 50 mm hg and mild AR is present in asymptomatic patients, prophylactic intervention to prevent AR progression may be considered (Class 11B; 2018 guidelines). Stress testing can be considered to assess symptoms or other concerning findings in the absence of symptoms (Class 11b; 2018 guidelines). If high gradients (maximum gradient > 50 mm hg) are noted due to subaortic obstruction post atrioventricular septal repair, surgical repair may be considered even in asymptomatic patients (Class 11b ; 2018 guidelines). Asymptomatic patients with subAS and peak gradients > 50 mm hg may benefit from prophylactic intervention to prevent long term complications including progressive AR. Surgery for subAS has been reported to have higher complications especially complete heart block (10-15%) and a higher chance of recurrence if initial surgery was in childhood [32].

3.3. Supravalvular Aortic Stenosis

Supravalvular Aortic Stenosis (SupraAS) can be discrete or diffuse and is a subtype of LVOT obstruction accounting for less than 7% of all cases [34]. SupraAS is defined as a fixed obstruction distal to the coronary artery ostia resulting in high coronary systolic pressures with the impaired diastolic flow. SupraAS is associated with mutation of the elastin gene on chromosome 7q11.23 resulting in loss-of-function [35]. In some situations, there can be partial or complete coronary artery ostial obstruction, ectasia, and aneurysm. SupraAS can be associated with Williams syndrome, an autosomal dominant disorder with hypercalcemia and there can be hypoplasia of the entire aorta, stenosis of major branches of the aorta, renal artery stenosis and segmental pulmonary artery stenosis. Aortic imaging with TTE, TEE, CT or MRI is recommended in patients with William syndrome suspected of having SupraAS (Class 1C; 2018 guidelines). Furthermore, in patients with Williams syndrome and SupraAS with angina symptoms, coronary imaging is recommended (Class 1C; 2018 guidelines).

In adult patients with unrepaired SupraAS, the presentation may be due to symptoms of outflow obstruction (such as dyspnea, syncope, or angina), hypertension or myocardial ischemia. In non-familial SupraAS sudden cardiac death is less common compared with familial SupraAS with associated Williams syndrome. On clinical examination, there may be differential blood pressure measurements noted with a right upper extremity systolic blood pressure greater than

the left. This can be manifested in patients with the preferential flow (Coanda effect) into the right brachiocephalic artery.

ECG can show LVH with non-specific ST and T wave abnormalities. Trans-thoracic and trans-esophageal echocardiography can be used to define aortic arch anatomy, quantify the gradient across the area of narrowing and identify the presence of LVH. Cardiac MRI or CT provides more detailed and precise evaluation of the aorta and its branches as well as the pulmonary vasculature to find other areas of narrowing. Non-invasive workup of ischemic symptoms is preferred with cardiac CT, but if necessary invasive workup can be performed with extreme caution as many patients do have ostial coronary artery stenosis [36].

Surgery is the primary treatment, with an operative mortality of <5% [34]. It is recommended in patients with SupraAS with decreased LV function and symptoms attributable to supravalvular obstruction (Class 1C; 2018 guidelines) and coronary revascularization is recommended in SupraAS patients with ostial coronary stenosis (Class 1C; 2018 guidelines). Patients should be followed annually postoperatively, and imaging modalities such as echocardiogram or cardiac MRI should be used to monitor the repair site. There is a strong inheritance pattern in both familial and non-familial SupraAS, and family members should be screened [10].

4. AORTOPATHY RELATED TO CONGENITAL HEART DISEASE

The aorta plays many important roles, primarily as a conduit, but it also functions to control the systemic vascular resistance and heart rate by means of pressure-responsive receptors across the ascending aorta and aortic arch. The diaphragm divides the thoracic from the abdominal aorta. Histologically the aorta has three layers. The tunica intima is thin and lined by endothelium. The tunica media is thick with concentric layers of collagen and elastin fibres in addition to smooth muscle cells. The outer layer known as the tunica adventitia is composed of collagen, vasa vasorum, and lymphatics [37].

In a healthy adult patient, the aortic diameter does not usually exceed 4.0 cm [38]. The aorta gradually tapers downstream. Aortic size can vary with age, gender, BSA, height, weight, and blood pressure. There is a trend of slow progressive dilation of the aorta over the period of mid to late adulthood, which is thought to be due to the aging processes, related to the higher collagen to elastin ratio, increased stiffness and pulse pressure. The rate of aortic expansion is about 0.9 mm in men and 0.7 mm in women for each decade of life [39, 40].

4.1. Coarctation of the Aorta

Coarctation of the Aorta (CoA) has an incidence of 5-8% of all congenital heart defects, occurring as isolated forms in approximately 3 out of 10,000 live births [38]. CoA

may be caused by narrowing with discrete stenosis or hypoplasia of the aortic segment, typically at the insertion area of ductus arteriosus in proximity to the origin of the left subclavian artery. It is associated with BAV in up to 85% of cases and as discussed under BAV section, active screening for associated coarctation is needed in patients with BAV [34]. It may be part of the Shone syndrome complex (supra-mitral ring, parachute mitral valve, SubAS, and BAV), Turner syndrome, congenital rubella, Williams-Beuren syndromes, VSD, the circle of Willis cerebral artery aneurysm, Takayasu aortitis and neurofibromatosis.

Histologically CoA exhibits cystic medial necrosis, early fibre fragmentation and fibrosis in the ascending and descending segments of the aorta [23]. This results in increased stiffness of the aorta and carotid arteries. CoA increases LV afterload, which increases wall stress, that over time contributes to LVH and LV dysfunction. CoA causes an increase in the risk of dissection or rupture in the ascending aorta or area of narrowing itself.

The presentation is dependent on the severity of the lesion. Patients may present in childhood with severe cases while more mild forms may not manifest till adulthood. In young adult patients, the development of hypertension with discrepant upper and lower extremity pulses or underdevelopment of lower extremities should warrant further investigation for possible CoA. Symptoms may include headache, exertional leg fatigue, and claudication. Unrepaired CoA in adults leads to aortic rupture *via* progressive aortic dilation. Patients with poorly controlled hypertension are more likely to develop aortic dilation, especially for the ascending aorta and post-coarctation segment of aorta [41]. A blood pressure gradient of > 20 mmHg between the upper and lower extremities marks significant CoA. Collateral vessel formation is common and is often responsible for masking symptoms. The collaterals can cause a continuous murmur on examination, and rib notching on a chest x-ray. Other x-ray findings that may be present include an ectatic ascending aorta, and the "Fig. **3** sign" (pre-stenotic dilatation of the aortic arch, indentation at the area of coarctation, and post-stenotic dilatation of the descending aorta).

The presence of hypertension should be treated medically with goal-directed medical therapy which can be beta-blockers, angiotensin-converting enzyme inhibitors, or angiotensin receptor blockers (Class 1C; 2018 guidelines). Recurrence of hypertension is common and in some cases manifests clinically years later, after the initial coarctation repair. TTE imaging can help to screen for CoA. The suprasternal view is an important window to assess the aortic arch and proximal descending aorta. Even if the aortic arch and upper thoracic aorta cannot be well visualized, color doppler guided turbulence and combination of pulse and continuous wave doppler can serve as a valuable initial assessment to suspect coarctation. Significant CoA will show a high-velocity systolic peak (> 1.5 m/s) with gradual deceleration throughout diastole on the doppler examination. Peak and

mean gradients can be recorded (see Fig. **8**). A low velocity signal with antegrade continuous diastolic flow on the doppler examination of the descending aorta at the level of the diaphragm can aid in forming the diagnosis. Abdominal aorta examination may show an abnormal doppler flow pattern with decrease pulsatility without evidence of early diastolic flow reversal.

While echocardiography remains useful as a diagnostic tool, CT or MR imaging are excellent to delineate the aorta and site of coarctation as the aortic isthmus and proximal descending aorta can be difficult to image in adults by TTE and it should be performed once in every patient with CoA repaired or unrepaired. MRI or CT angiography may be reasonable as a screening tool for the presence of intracranial aneurysm (berry aneurysm) in patients with CoA (Class 11b; 2018 guidelines).

In some centers, cardiac catheterization is still considered a gold standard for evaluation before and after surgical or interventional treatment. A hemodynamically significant coarctation will have >20 mmHg peak-to-peak gradient if collaterals are not well developed. The presence of a significant gradient and hypertension is currently an indication for surgical repair or stenting of the coarctation if the anatomy is appropriate (Class 1B; 2018 guidelines). This is an important change as previous guidelines required only a significant gradient but not hypertension. If anatomy is not appropriate for stenting or surgery is not an option, balloon angioplasty can be considered (Class 11b; 2018 guidelines). The use of covered or non-covered stents remains an area of debate. Surgical risk in uncomplicated CoA is 0.1%, but this risk increases after the patient reaches the third or fourth decade of adulthood [34]. Surgical techniques include resection of the narrowing with end-to-end anastomosis of the aorta, resection and extended end-to-end anastomosis, patch aortoplasty, subclavian flap aortoplasty, tube graft and bypass tube grafts. There is an operative risk of spinal cord injury. Invasive repair should be performed in a specialized center with high volume and expert care in the treatment of CHD.

Post operatively the recurrence of systemic hypertension in well documented and should be treated medically and exercise testing may be useful in identifying hypertensive response which may predict future hypertension in those who are normotensive. Recurrence of the CoA or aneurysms of the repair site are known complications and follow up imaging is recommended. Cardiac MR is the preferred imaging modality at least once in 5 years with follow up in the clinic advised at least every year [10].

4.2. Aortic Dilation

Atherosclerosis is the most common cause of aortic disease, followed by aortic aneurysm [38]. Traditionally aneurysm is further classified as thoracic or Abdominal Aortic Aneurysm (AAA), although it is possible to have both lesions simultaneously [42]. The 2010 ACC/AHA/AATS guidelines for diagnosis and management of the thoracic
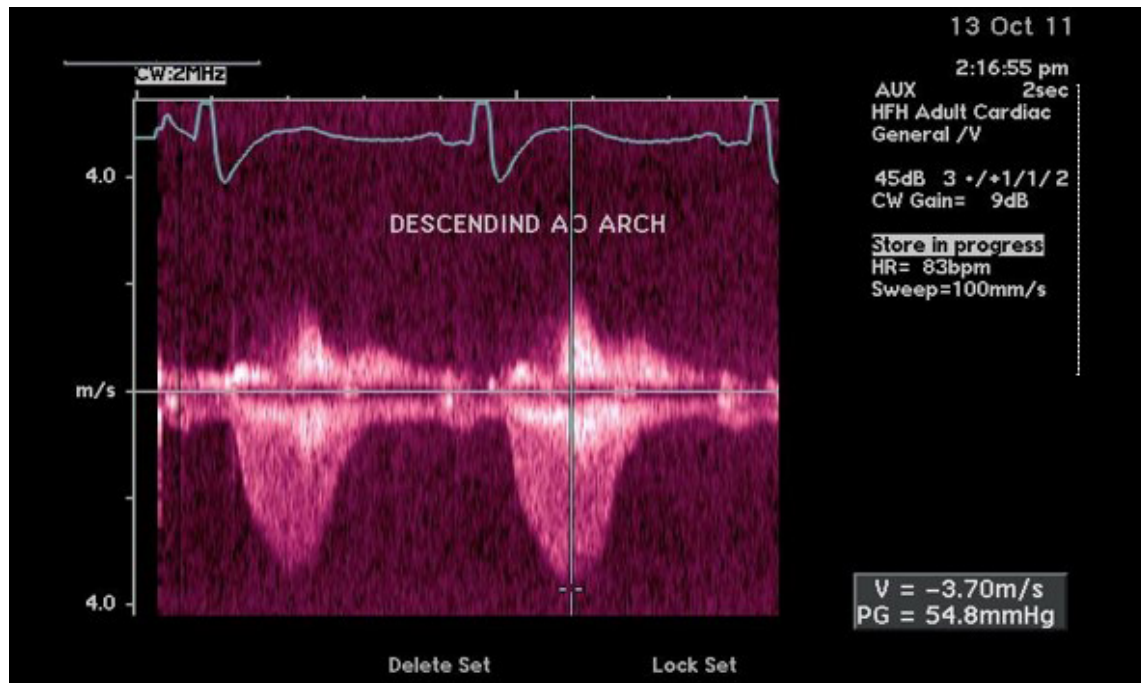
Fig. (8). Coarctation of the aorta noted by PW Doppler in the descending aorta showing a continuous sawtooth pattern. The peak velocity is 3.7 m/s with a maximum instantaneous gradient of 55 mmHg, indicating severe obstruction. Please also note persistent flow into diastole below the baseline indicating significant obstruction. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

aortic disease [43] detail the approach to aortic dilation in various conditions and serves as a key reference for recommendations for surveillance and timing of intervention. This section will primarily focus on aortic root and thoracic aorta anomalies.

### 4.2.1. Aortic Root and Thoracic Aortic Aneurysm

There are certain patterns between the location and etiology of Thoracic Aortic Aneurysm (TAA) that have been recognized. The most common cause is a degenerative aneurysm of the ascending aorta [42]. BAV patients frequently have aortic dilation in the ascending aorta and less frequently in the aortic root at the level of the sinus of Valsalva (Fig. **8**). BAV valve morphology appears to influence the incidence of dilation, more frequent in patients with RCC and NCC fusion [43, 44]. Marfan syndrome is caused by FBN1 mutation and in 10% of cases Marfan phenotype is associated with TGFBR1/2 and tends to cause aortic enlargement at the level of the sinuses of Valsalva and the expansion of the sinuses includes the wall of sinuses proximal to the coronary orifices [42, 43].

The type of aortic dilation in Marfan is very amenable to initial TTE evaluation and if needed TEE may aid in a comprehensive assessment of aortic root and aortic regurgitation (Figs. **9a** and **9b**). If a complete morphological assessment of aorta is warranted, it is recommended to image the entire aorta, and accurate dimensional assessment is best made with CT or MRI at initial diagnosis and at 6 months. Care

should be made to accurately measure the diameter perpendicular to the long axis. Patients are often asymptomatic, and imaging alone often makes the diagnosis [38-43]. Once TTE and CT or MRI imaging correlates to each other, TTE could be used for follow-up in Marfan patients.

Loeys –Dietz and related syndromes such as familial thoracic aortic syndrome is another cause of aortic dilation caused by TGFBR1 and TGFbR2 mutations and may have Marfan phenotype. These patients should undergo complete aortic imaging at initial diagnosis and 6 months thereafter to establish if there is a progressive enlargement (Class 1C) [43]. Cardiac CT or MRI can be useful in the complete delineation of the aorta (Figs. **10a** and **10b**). Given the young age of many of these patients and to avoid radiation, MRI may be preferred for follow-up. Guidelines indicate that they should have yearly magnetic resonance imaging from the cerebrovascular circulation to the pelvis (Class 1B) [43].

In adults with Marfan syndrome, yearly TTE measurement is recommended as long as aortic root size is < 45 mm. If > 45 mm then the twice-yearly measurement should be done to evaluate for stability versus a continued increase in size. TAA of the ascending aorta increases by 0.1 cm/year and in familial cases the rate is faster up to 0.21 cm/year [10]. Descending aorta growth in TAA is even faster at 0.3 cm/year [42, 44]. Patients with TAA with ascending aorta diameter > 6.0 cm or descending aorta diameter > 7.0 cm are at high risk of dissection or rupture [43, 45].
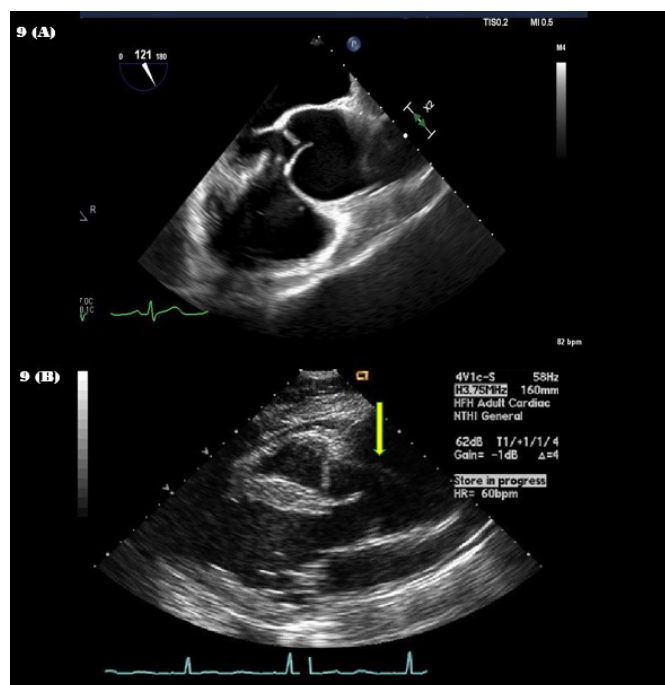
Fig. (9). Aortic root aneurysm in a patient with Marfan syndrome as shown in long axis (TEE) view also showing effacement of the sinotubular ridge and malcoaptation of aortic valve leaflets (**9A**). TTE image (parasternal long axis view) of a patient with Marfan syndrome with aortic root aneurysm and dissection flap (yellow arrow) (**9B**). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
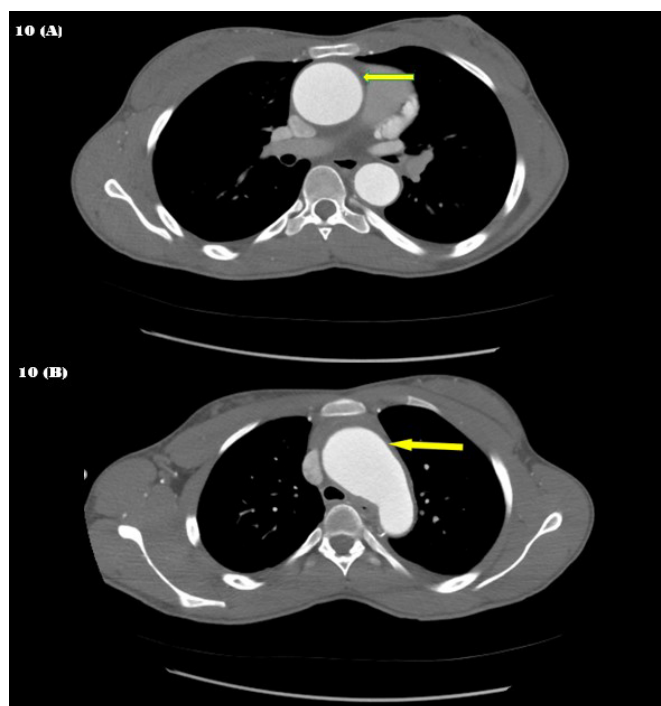
Fig. (10A and B). Gated cardiac CT scan (axial) images of aorta , showing superb delineation of the aortic aneurysm involving the ascending aorta and extending to the arch in a 21-year old patient with Loey-Dietz syndrome (arrows). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

Surgical intervention is recommended based on the diameter and etiology of aortopathy. In Marfan syndrome or patients with Marfanoid pattern without meeting criteria for Marfan syndrome, surgical intervention is indicated when the aortic diameter is ≥5.0 cm [43, 46]. If there is a family history of dissection, rapid increase in size (> 0.3 cm/year), severe aortic insufficiency, or planned pregnancy, then a diameter of 4.5 cm can be used [43, 47]. Surgical repair cut-offs are different in Loeys-Dietz syndrome with earlier intervention at 4.2 cm (by TEE internal diameter) or 4.4 to ≥4.6 cm by CT and or MRI [43]. In BAV, surgical intervention is recommended with aortic diameter > 5.5 cm, or at an aortic diameter of 5.1cm to 5.5 cm, if there is a family history of aortic dissection or rapid progression of dilation [43, 48]. In patients with BAV undergoing Aortic Valve Replacement (AVR) for severe stenosis or insufficiency, if the aortic diameter is > 4.5 cm then replacement of the aortic arch is reasonable [43, 47].
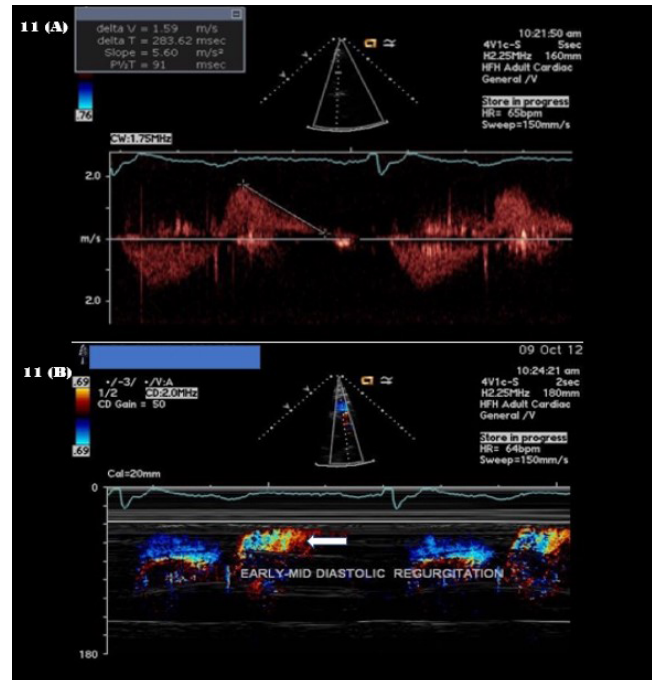
4.3. Tetralogy of Fallot

Tetralogy of Fallot is one of the most common congenital cyanotic heart disease conditions that adult cardiologists will encounter following complete repair. It is a combination of four congenital abnormalities which include a Ventricular Septal Defect (VSD), pulmonary valve stenosis, overriding aorta, and a thickened right ventricular wall (right ventricular hypertrophy) [49].

With the advent of improved surgical techniques, the long-term survival after Tetralogy of Fallot (TOF) repair is improved. Adult patients with a history of TOF repair are more likely to have heart failure symptoms, increased risk of arrhythmias, and sudden death during early adulthood [49, 50].

The focus in surgical repair of TOF is to fix the RVOT obstruction by infundibulotomy, resection of obstructive muscle bundle and to enlarge RVOT with the help of patch. This complex surgical procedure can result in scar tissue, aneurysmal area in RVOT, residual RVOT stenosis, and obligatory PI. PI is the most common valvular finding in adults with a history of TOF repair. Chronic PI leads to progressive right ventricular dilation and dysfunction, LV dysfunction and electromechanical dyssynchrony [10]. Echocardiography serves as the best initial modality to evaluate for left and right heart function and PI. Care needs to be taken for interpreting PI severity, which may be underestimated given wide open PI, which will appear as laminar short duration diastolic flow between the pulmonary artery and right ventricle (Fig. **11**). The assessment of the right ventricular size and function can be challenging with echocardiogram, despite the use of echo contrast true volumetric assessment is suboptimal. Cardiac MRI is essential for RV assessment in post-repair TOF patients and is recommended to assess for RV size (volume) and function and to assess for PI severity, regurgitant fraction by phase-contrast techniques can be calculated to aid in timing of PV replacement [10] (Fig. **12**). MRI measured RV volumes with values exceeding 160ml/sqm usually suggest the need for PV replacement even in

Fig. (11A and B). Continuous wave doppler across right ventricular outflow tract showing brief pulmonic regurgitation due to rapid equilibration of PA-RV pressure from a wide open PI post-op tetralogy of Fallot patient, presenting with right ventricular dilation (**11A**). Color M – mode image below showing early to mid –termination of PI jet (**11B**). (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).
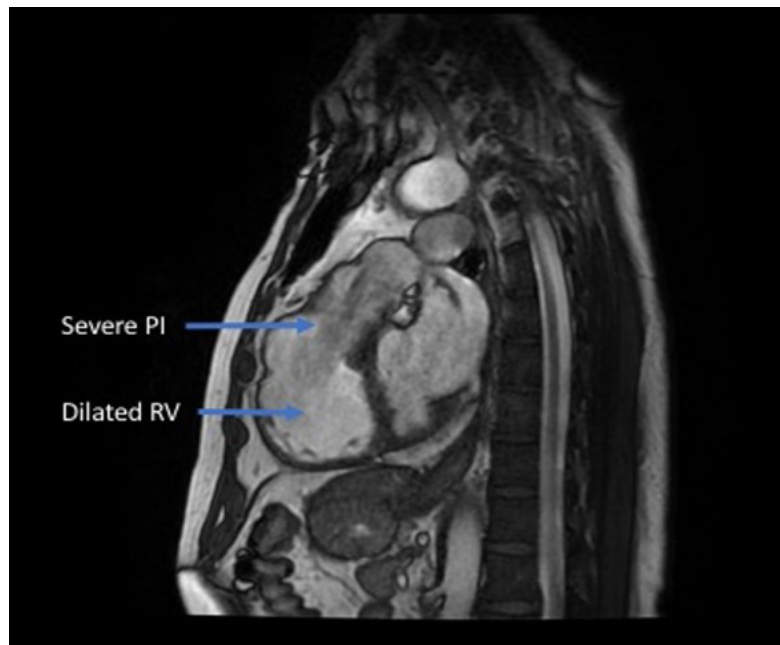
Fig. (12). Steady state free precession MRI image of right ventricular outflow tract showing RV dilation and severe wide-open PI. (*A higher resolution / colour version of this figure is available in the electronic copy of the article*).

asymptomatic patients [10]. RV volumes exceeding 170 ml/sqm have been associated with a lack of regression of RV dilation following PV replacement.

Typically, surgical replacement of PV is recommended in symptomatic patients (with dyspnea, chest pain, and/or exercise intolerance otherwise unexplained) with repaired TOF and severe PI (Class 1, LOE B; 2018 guidelines). Early data is emerging regarding the evaluation of transcatheter options for PV replacement as an alternative for surgery, but data is still limited in this area.

Pulmonary valve replacement can also be considered in patients if 2 of the following are met (Class 2a, LOE B; 2018 guidelines):

[a] Mild or moderate RV or LV systolic dysfunction.
[b] Severe RV dilation determined by MRI (RV end-diastolic volume index ≥ 160 mL/m^2, or RV end-systolic volume index ≥ 80 mL/m^2, or RV end-diastolic volume $\geq$2x LV end-diastolic volume).
[c] RV systolic pressure due to RVOT obstruction $\geq$2/3 systemic pressure.
[d] Progressive reduction in objective exercise tolerance.

Following repair, adult patients may present for routine follow-up and physical examination. These patients often have Pulmonary Insufficiency (PI) murmur which may not be well appreciated in case of severe PI, due to rapid equilibration of PA-RV pressures. The left radial pulse may be diminished if the patient had undergone initial palliation such as a modified Blalock-Taussig shunt (a graft between the subclavian artery and the pulmonary artery). Aortic regurgitation can be a long-term complication along with aortic dilation. Progressive right ventricular dilation and dysfunction can develop related to PI and needs evaluation. Other symptoms include palpitations that should prompt further investigation, given the predilection for atrial and more importantly ventricular arrhythmias [10]. The first step is to exclude hemodynamic and anatomical triggers and to start appropriate pharmacotherapy. Holter monitoring or implantable loop recorder can be immensely helpful to aid in early diagnosis of potential malignant arrhythmias [51]. One study has demonstrated that the main source of morbidity in these patients is related to atrial arrhythmias: atrial fibrillation, atrial flutter, supraventricular tachycardia, and sinus node dysfunction. The same study also demonstrated that a significant number of patients needed pacemakers for heart blocks or symptomatic bradycardia [51]. Symptoms such as pre-syncope or syncope may be a signal for serious ventricular arrhythmias and should prompt electrophysiologic consultation [10].

CONCLUSION

This review is generated for busy clinicians to reference the following most common adult congenital heart diseases, such as left to right shunts, left heart obstructive lesions, and aortopathies. In this review, we have used current literature and have highlighted key aspects of the 2018 adult congenital heart disease guidelines pertaining to the above-mentioned adult congenital heart diseases. Ongoing surveillance and periodic diagnostic evaluation using imaging modalities may be needed in selected instances as outlined in the 2018 adult congenital heart disease guidelines [10]. This review article will prove greatly beneficial for clinicians to use as a quick reference.

VIDEO LEGENDS

Video 1: transesophageal echo short-axis view of atrial septum showing large secundum ASD with color flow left –to- right.

Video 2: transthoracic echo parasternal long-axis view showing color flow across perimembranous septal defect consistent with perimembranous VSD.

Video 3: transthoracic echo short-axis view of pulmonary artery showing large PDA with a color flow from upper descending thoracic aorta streaming into left and main pulmonary artery.

Video 4: cardiac gated CT of the aortic valve showing the bicuspid aortic valve in systole and diastole.

Video 5: transthoracic echo apical long-axis view showing subaortic ridge near left ventricular outflow tract.

CONSENT FOR PUBLICATION

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CONFLICT OF INTEREST

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SUPPLEMENTARY MATERIAL

Supplementary material isavailable on the publisher's web site along with the published article.

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