Chapter 3
Congenital Heart Defects Which Include Cardiac Valve Abnormalities

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Abbreviations

AO	Aorta
AS	Atrial septum
IBL	Inferior bridging leaflet
LA	Left atrium
MV	Mitral valve
OS	Outlet septum
PT	Pulmonary trunk
RA	Right atrium
RPA	Right pulmonary artery
RV	Right ventricle
RVOTO	Right ventricular outflow tract obstruction
SBE	Superior bridging leaflet
TV	Tricuspid valve
VSD	Ventricular septal defect

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P. A. Iaizzo et al. (eds.), *Heart Valves*, https://doi.org/10.1007/978-3-031-25541-0_3

3.1 Inherited Valve Diseases

Isolated congenital valve disease	Valve disease with other congenital heart defect
Aortic valve stenosis/regurgitation	Atrioventricular septal defect
Pulmonary valve stenosis/regurgitation	Tetralogy of Fallot
Mitral valve (MV) stenosis/ regurgitation	Truncus arteriosus
Ebstein's anomaly	Shone's complex
	cc-TGA with Ebstein's anomaly
	Complex left ventricular outflow tract obstruction
	Pulmonary stenosis with ventricular septal defect (VSD)
	Subaortic VSD causing aortic insufficiency

3.1.1 Aortic Valve Stenosis

Aortic valve stenosis (AS) [1] in children is a congenital heart malformation causing a fixed left ventricular outflow tract obstruction which is found at birth or shortly thereafter. Presentation varies in severity and can be found from minor signs of murmur evaluation to more severe presentation with congestive heart failure with a severe lactic acidosis and cardiovascular collapse requiring emergency treatment. The natural history of mild to moderate stenosis is a gradual increase in severity and patients will often develop aortic regurgitation. Other types of aortic stenosis include discreet fibromuscular and tunnel subaortic stenosis together with supra valve stenosis. The optimal treatment for moderate to severe congenital AS has been debated in the last few decades, either balloon aortic valvuloplasty (BAV) versus surgical aortic valvotomy (SAV). In determining the best treatment, factors that are considered include procedural success, in hospital morality, development of aortic regurgitation, and reintervention rates. There is a strong case for BAV in the neonate or young infant with severe aortic stenosis and resultant severe left ventricular dysfunction. Brown and colleagues stated that overall SAV provides a better gradient reduction, less postoperative aortic regurgitation, and a lower reintervention rate at 10 years compared to BAV without a difference in survival or need for aortic valve replacement. However, a meta-analysis, recently described by Saung and colleagues, showed that although the reintervention rate was higher for BAV compared to SAV, the survival rates, need for aortic valve replacement, and late aortic valve regurgitation are similar.

3.1.2 Aortic Valve Regurgitation

Isolated congenital aortic valve regurgitation is extremely rare. Most often, it is associated with congenital AS or occurs following aortic valve procedures. It may arise as a complication of surgical procedures on the left ventricular outflow tract or as a complication of other types of surgery (i.e. VSD closure, subaortic stenosis resection, or other types of interventional cardiology procedure). Beside aortic valve replacement, there are different types of repairs to address these cases.

3.1.3 Pulmonary Valve Stenosis

Congenital pulmonary valve stenosis can range in severity from minimal disease to critical disease requiring immediate intervention. Most commonly, if intervention is indicated, this valvular defect is treated with transcatheter balloon valvuloplasty. In the most severe cases, neonates may require prostaglandin infusion before and after intervention in order to augment pulmonary blood flow until the right ventricular compliance improves after relief of the valve obstruction. In rare extreme cases, a pulmonary artery shunt is needed. Percutaneous valvuloplasty remains the treatment of choice with rare complication rates. It can result in excellent intermediate and long-term results, with only 10% restenosis requiring reintervention. More commonly following balloon intervention, patients can develop pulmonary insufficiency, however, this is typically well tolerated for many years. In some cases, if there is severe pulmonary valve insufficiency, patients may require pulmonary valve replacement. A close cousin of severe (critical) pulmonary valve stenosis is pulmonary atresia with intact ventricular septum in which there is an imperforate pulmonary valve. Initial treatment is by percutaneous trans-venous radio-frequency perforation of the atretic valve accompanied by balloon pulmonary valvuloplasty.

3.1.4 Pulmonary Valve Regurgitation

Pulmonary valve regurgitation is commonly the acquired outcome of transcatheter intervention for pulmonary stenosis or surgical intervention for tetralogy of Fallot or severe right ventricular outflow tract obstruction (RVOTO). Congenital moderate to severe regurgitation is extremely rare. The only congenital heart defect associated with pulmonary regurgitation as a prominent feature is tetralogy of Fallot with absent pulmonary valve syndrome. This syndrome often requires neonatal intervention due to severe pulmonary valve insufficiency with severe dilation of the

pulmonary artery tree which can compromise the airway anatomy with severe trachea-bronchomalacia. Intervention includes surgical repair of tetralogy of Fallot with pulmonary valve placement with valved conduit and pulmonary arterioplasty.

3.1.5 MV Stenosis

Congenital mitral stenosis can present as an isolated defect or in association with other left heart obstructive lesions [2]. In the latter, patients often need to pursue single ventricle palliation due to hypoplasia of multiple left heart structures. In isolated cases, obstruction around the valve can happen at different levels, either supravalvular, valvular, or subvalvular, or in combination such as in parachute, arcade lesion, or hammock MV. Surgical procedures aim to remove the obstruction and restore leaflet mobility and function. There have been different techniques described to achieve this. The results have improved over the years, although there are several factors that appear to be important for survival and long-term outcomes, including age at presentation, development of pulmonary artery hypertension, and severity of the lesion. In few cases, valve replacement in a supra-annular position can be used. The association of some form of mitral stenosis with left ventricular outflow obstruction and with or without coarctation of the aorta (AO) is sometimes called Shone's complex.

3.1.6 MV Regurgitation

MV regurgitation can result from abnormal development of the MV including MV prolapse, collagen vascular disorders/connective tissue disorders, mucopolysaccharidosis, and papillary muscle dysfunction. In the last 20 years, different surgical techniques have been developed to repair these valves, and in severe cases MV replacement is needed. The technique of percutaneous delivery of a 'Mitra-clip' used in some older adults with severe regurgitation has not been applied to children and younger adults.

3.2 Ebstein's Malformation of the Tricuspid Valve (TV)

Ebstein's anomaly is a complex abnormality of the TV and right ventricular myocardium in which the hinged attachments of the septal and inferior leaflets of the TV are displaced away from the atrioventricular junction towards the apex of the right ventricle (RV). The valve leaflets become adherent to right ventricular myocardium giving rise to 'atrialization' of a portion of the RV, right atrial enlargement, and tricuspid regurgitation. Dearani reports the anatomical features of this defect

including failure of the leaflet delamination, apical descent of the functional valvular orifice, right ventricular dilation and atrialization, anterior leaflet abnormal fenestrations and tethering, and right atrioventricular junction dilation. The degree of apical displacement and severity of tricuspid regurgitation remains the most important clinical determinant of the outcome of Ebstein's malformation. Asymptomatic patients can be managed medically for many years, but TV repair using the Cone Operation [3] described by Da Silva, in the correct hands, can produce outstanding results and should be considered if patients start to develop severe tricuspid regurgitation, worsening exercise capacity, cyanosis, and right ventricular dysfunction. Any arrythmias are managed in advance of surgery by radiofrequency ablation techniques. The requirement for TV replacement with a bioprosthetic valve is becoming less frequent.

3.3 Atrioventricular Septal Defects ('AV Canal' Defects)

The atrioventricular septum is that part of intracardiac septal structures separating left ventricle from right atrium. The characteristics of an atrioventricular septal defect (AVSD) are complete deficiency of the atrioventricular septum resulting in a common atrioventricular junction, common atrioventricular valve, primum atrial septal defect whose inferior border is the common valve leaflets and interventricular defect whose superior border is the common valve leaflets [4] (Fig. 3.1). AVSD comprises a spectrum of defects ranging from partial to intermediate to complete AVSD. The 'complete' form is characterized by a primum atrial septal defect, inlet VSD, common atrioventricular valve with abnormal leaflet support structures leading to variable degrees of regurgitation. The complete form of AVSD requires surgical repair within

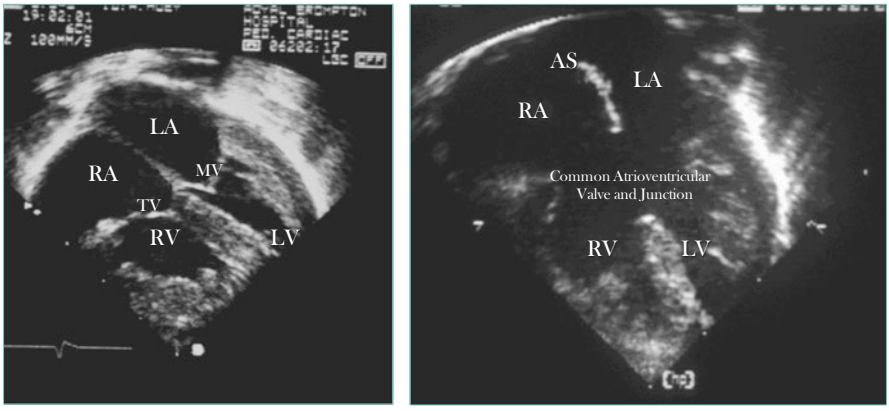

Fig. 3.1 (**a**) Normal 4 chamber echocardiographic section of the atrioventricular junction with normal tricuspid and MVs. (**b**) By way of contrast this echocardiographic 4 chamber section demonstrates the common atrioventricular junction during diastole in a complete atrioventricular septal defect (AVSD)

the first 6 months of life, either with single or two-patch technique to close the ASD and VSD and repair the commissural 'cleft' between the superior and inferior bridging leaflets of the part of the common valve within the left ventricle. Repair of a so-called 'partial' AVSD is to close the primum ASD and partly to repair the valve leaflets in the same way. The right- and left-sided atrioventricular valves have no similarity to the normal tricuspid and MVs. The results are normally excellent, however, left-sided atrioventricular valve regurgitation is a major cause of morbidity and need for reintervention including re-repair or valve replacement. The outcome of AV canal repair depends on several factors related to the anatomy of the valve leaflets, relative size of the left and right ventricular components of the common valve and ventricular proportions. Association with Down syndrome portends a better prognosis for complete AVSD repair, with non-Down's patients more likely to require more repeat intervention on the left atrioventricular valve.

3.3.1 Tetralogy of Fallot

Tetralogy of Fallot is characterized by constellation of features resulting from anterior and cephalad deviation of the outlet ('infundibular') septum giving rise to a large VSD with overriding of the AO, infundibular pulmonary stenosis and consequently right ventricular hypertrophy (Figs. 3.2, 3.3, and 3.4) [5]. The RVOTO and pulmonary stenosis can occur at multiple levels (infundibular, valve, supra valve, and pulmonary artery bifurcation). The timing of repair is dependent on the severity of the right ventricular outflow obstruction, although in recent years there is a strong tendency for complete repair earlier in life and certainly within 9–12 months of age. Historically, the management of the RVOTO and the rudimentary pulmonary valve was a transannular patch with autologous pericardium, although in recent years there is a tendency for surgeons to reconstruct valve patency in a different way including monocusp valve taken from allograft or re-create valve leaflets from other synthetic materials.

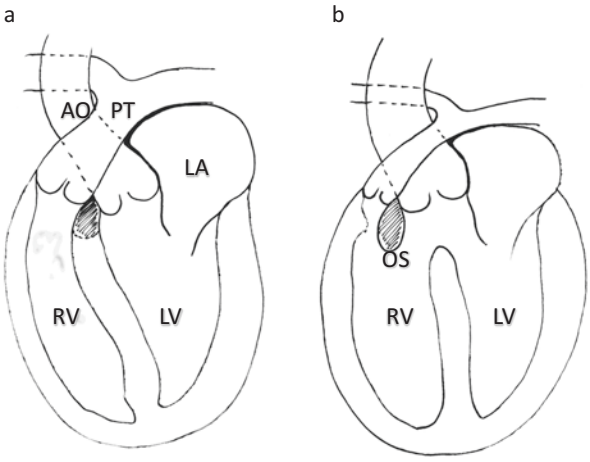

Fig. 3.2 Line drawing of the essential anatomic features of the normal heart (**a**) compared with tetralogy of Fallot (**b**) illustrating a large VSD, overriding of AO and infundibular pulmonary stenosis

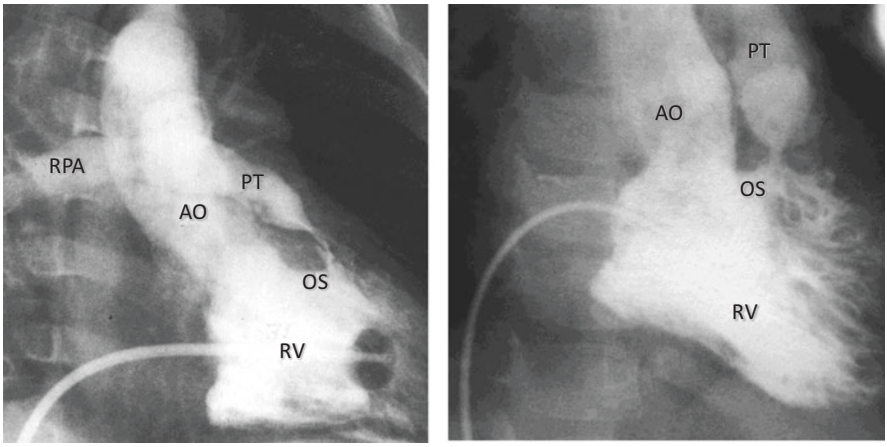

Fig. 3.3 Right anterior oblique projections of right ventricular angiography in two patients each demonstrating anterior deviation of the outlet (infundibular) septum with severe infundibular pulmonary stenosis. On the right-sided image, there are extensive septo-parietal trabeculations contributing to the stenosis

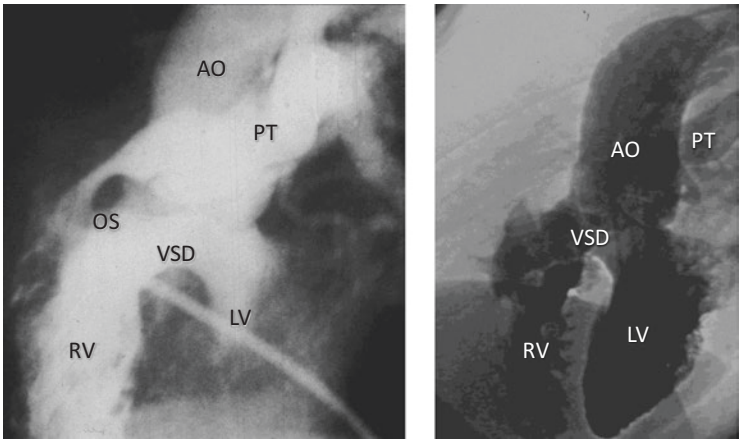

Fig. 3.4 Right and left ventriculograms in long axis projection from two patients each demonstrating a large VSD, overriding of the AO and infundibular pulmonary stenosis together with the pulmonary trunk and branch pulmonary arteries

3.3.2 Truncus Arteriosus

Truncus arteriosus ('common arterial trunk') is a 'cono-truncal' abnormality in which there is a failure of septation of the AO and main pulmonary artery [6]. A single great artery, the common trunk, overrides a large VSD and arises from the ventricles giving rise to the AO, pulmonary artery, and coronary arteries in its ascending part. The truncal valve usually has three or four leaflets, but can have anything from one to six with resultant stenosis or regurgitation. In 25% of cases,

there is valve insufficiency, and this has been identified as a risk for poor outcome and for subsequent need for truncal valve surgery if not addressed at the primary surgery. However, even after the initial repair, there is a high incidence of reoperation for the truncal valve. It has also been reported that creation of a tricuspid truncal valve confers the best outcome amongst the types of repairs, with the best freedom from truncal valve reoperation. The association with interrupted aortic arch is also a risk factor for poor outcome.

3.4 Shone's Complex

This term describes a combination of lesions that lead to multiple levels of left heart obstruction, including any type of mitral stenosis, variable degrees of left ventricular hypoplasia, subaortic or valvular aortic stenosis, hypoplastic aortic arch, and coarctation of the AO. Some of these infants will be palliated with single ventricle pathway, particularly if the MV and left ventricular are too small. If repair is needed, the aforementioned techniques for congenital mitral stenosis are used in association with relief of aortic stenosis and/or coarctation and arch repair. Relief of left ventricular outflow obstruction varies from simple aortic valvotomy to more complex forms of repair including Ross-Konno procedure (described in detail in another chapter).

3.5 Congenitally Corrected Transposition of the Great Arteries (CC-TGA) with Ebstein's Anomaly

CC-TGA is characterized by a discordant atrio-ventricular connection (RA to LV and LA to RV) with discordant ventricular-arterial connection. The left-sided TV found in the 'systemic' morphologically RV is often dysplastic in similar fashion to Ebstein's malformation (described by some as 'Ebsteinoid') with varying degrees of regurgitation, although true Ebstein's malformation with significant leaflet displacement occurs in only 5% of hearts with discordant AV connection (Fig. 3.5). The ventriculo-arterial connection may be double outlet RV with subpulmonary VSD in 20% of cases of discordant AV connection but basic surgical management remains the same.

In the anatomical repair of this malformation which includes an atrial switch operation (Senning or Mustard procedure) combined with an arterial switch operation or Rastelli type of intraventricular repair, the TV 'moves' to the sub-pulmonary morphologically RV and rarely requires surgical attention. Following the physiological repair of the malformation, or in previously unoperated cases where the RV remains in the systemic position, progressive tricuspid regurgitation and right ventricular dilation and dysfunction are often the indication for intervention, although

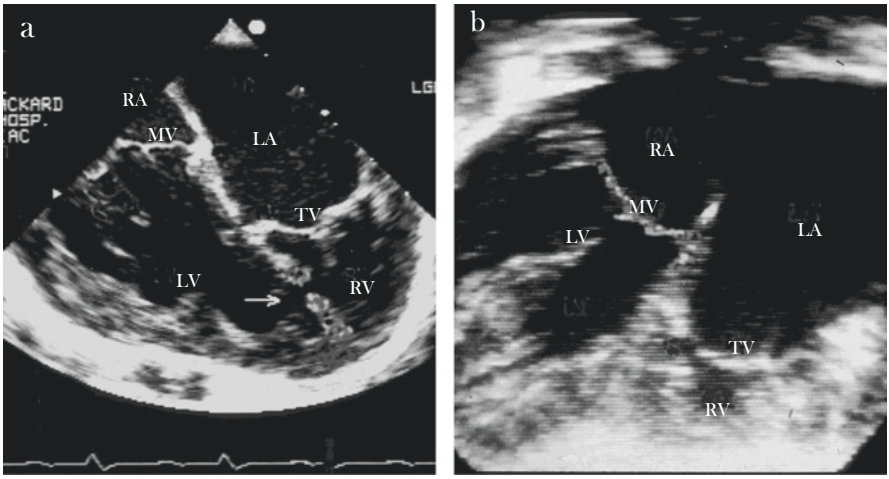

Fig. 3.5 Four chamber echocardiographic sections from two hearts with discordant atrioventricular connection and Ebstein's malformation of the TV. (**a**) The cardiac apex directed to the right and (**b**) the apex to the left. The arrow highlights a small muscular VSD. In each case, the TV is displaced from the atrioventricular junction into the left sided morphologically RV

in older patients, cardiac transplantation needs to be considered as the preferred option. CC-TGA with moderate pulmonary stenosis with or without VSD carries a good prognosis and surgical intervention may be better avoided is selected cases, delaying any treatment until there is a need for cardiac transplantation in the fourth or fifth decade.

3.5.1 Complex Left Ventricular Outflow Tract Obstruction

Besides the AS which as described previously, left ventricular outflow tract obstruction may occur in subaortic and supravalvular components as well. These areas of obstruction are often amenable to straight forward surgical intervention. However, there are also complex cases with subaortic obstruction with small aortic valve annulus which may require more complicated repair such as the Ross or Ross-Konno procedures, particularly because of the size of the aortic annulus.

3.5.2 Pulmonary Stenosis with VSD

Pulmonary stenosis with VSD is a form of congenital heart disease which differs from tetralogy of Fallot because of the absence of anterior deviation of the infundibular septum and frequently a smaller VSD. However, the treatment remains very

similar to that described above in management of tetralogy. Not infrequently because of the small VSD and gradually increasing severity of muscular subpulmonary stenosis, surgical repair is often later in childhood.

3.6 Subaortic VSD and Aortic Insufficiency

Sometimes perimembranous or muscular outlet VSDs in the subaortic or supracristal position, the so-called doubly committed sub-arterial defect, can require surgical intervention due to the development of progressive aortic insufficiency. These are often small to moderate defects that would not require surgery for VSD physiology; however, they can produce a 'venturi effect' on the aortic valve leaflets and lead to progressive aortic insufficiency. Closure of the VSD sometimes is sufficient alone, however, in some cases repair of the aortic valve leaflets or even leaflet resuspension is required.

3.7 Valve Disease Late After Repair of Congenital Heart Defects

Following repair of several types of congenital heart defects, valvular disease may develop during follow-up which may necessitate further intervention for repair or replacement. The most common pathology encountered in practice is pulmonary valve regurgitation following tetralogy of Fallot repair which may require treatment in different age groups. Without discussing the indications for treatment which will be addressed in another chapter, historical surgical treatment with valve replacement, with either bioprosthetic or homograft valve or in specific cases with mechanical prosthesis, was the gold standard. As more transcatheter valve options are developed, this has become the preferred choice in cases which are amenable to transcatheter valve placement. Additionally, in tetralogy of Fallot, because of the anatomic nature as part of the conotruncal abnormalities, aortic root enlargement and aortic valve regurgitation may develop particularly in adult congenital age groups and may require surgical intervention with root replacement ± valve repair or replacement. Similarly, in truncus arteriosus, another conotruncal defect, reintervention in the truncal valve for either stenosis or insufficiency may be required particularly in those cases where the valve has previously been addressed during the primary repair.

The other common post-operative valve disease we encounter is mitral regurgitation following AVSD repair. As aforementioned, some of these valves are amenable to further repair or may need replacement with bioprosthesis or mechanical prosthesis depending on the age of the patient. In recent years, techniques have been developed for surgical deployment of Melody Valves, used in percutaneous pulmonary valve replacement, in the mitral position.

Less common malformation in which there is valvular disease include complete transposition of the great arteries ('D-TGA') late following repair with the arterial switch operation. Either the neo-aortic valve (former pulmonary valve) or neo-pulmonary valve can progressively develop regurgitation necessitating repair or replacement.

3.8 Valve Disease Related to Inherited Conditions

There are several genetic syndromes that have specific associated valvular disease. Most commonly seen is MV prolapse, whereas MV regurgitation is seen in connective tissue disorders, including Marfan's syndrome, Ehlers-Danlos syndrome, and Loeys-Deitz syndrome. In addition to MV disease, in particular in the Marfan population, they can develop aortic root dilation with subsequent development of aortic valve regurgitation. This is not truly a valvular disease, but is a consequence of the root dilation, therefore in most cases surgical treatment with valve sparing root replacement, as popularized by Magdi Yacoub and Tyrone David, will suffice and rare cases require valve repair or replacement.

Inherited storage disorders, in particular the mucopolysaccharidosis diseases, have progressive multi-valvular disease related to deposition of mucopolysaccharides in the valvular tissue. The most common that requires surgical intervention is development of mitral or aortic stenosis. Due to the nature of the underlying disease, most commonly these require replacement with mechanical valve, as the mucopolysaccharides could be deposited in a bioprosthetic valve.

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