Congenital Heart Disease: A Medical Overview

1

Massimo Chessa and Francesca Romana Plucchinotta

Congenital heart disease with an approximate incidence of 1 % and a prevalence at birth of 5–11 per 1,000 live births [1] is the most common inborn defect. By definition, "congenital" heart disease means usually a disease that has been present since birth. However, a wide variety of defects can be either not present or not evident from birth. Those present but not usually detected in early life include lesions such as a moderate size atrial septal defect. Others that are only anatomically present in later years with a latent predisposition prior to this, such as subaortic stenosis or many of the cardiomyopathies, are not strictly "congenital" but are often included in this group.

The most important physiopathologic aspects of the congenital cardiac malformations are the presence of shunt between arterial and venous blood and presence or absence of cyanosis.

A shunt occurs when there is abnormal communications between two chambers or two vessels. It may be described as right-to-left, left-to-right, or bidirectional. The direction of the shunt is controlled by the pressure differential between the two cardiac chambers. Most commonly the blood goes from the high-pressure left side of the heart to the low-pressure right side of the heart resulting in a left-to-right shunt. The consequences of this shunt are (1) excessive blood flow into the lungs causing shortness of breath and increased pulmonary vascularity on a chest x-ray, (2) increased volume overload of one ventricle resulting in hypertrophy of myocardium and chamber dilatation, and (3) turbulence of abnormal blood flow producing a heart murmur.

Untreated left-to-right shunts will eventually result in increased pulmonary arterial pressure that may alter the normal maturation of the pulmonary vascular bed [2]. The pulmonary arteriole transitions from having a reactive muscular wall to

M. Chessa (✉) • F.R. Plucchinotta
Pediatric and Adult Congenital Heart Disease Centre,
IRCCS Policlinico San Donato University Hospital, Via Morandi 30, Milan, Italy
e-mail: massichessa@yahoo.it

E. Callus, E. Quadri (eds.), *Clinical Psychology and Congenital Heart Disease: Lifelong Psychological Aspects and Interventions*,
DOI 10.1007/978-88-470-5699-2_1

potentially irreversible structural changes consisting of medial hypertrophy and intimal proliferation, which manifests as increased pulmonary vascular resistance (PVR). Eventually, PVR exceeds systemic vascular resistance, leading to right-to-left shunting across the VSD, and cyanosis (Eisenmenger syndrome). The development of permanent injury to the pulmonary vessels is a function of the duration of the exposure to excessive blood flow and the anatomy.

In presence of a right-to-left shunt, oxygen-poor blood from the right side of the heart mixes with oxygen-rich blood in the left side of the heart, and the blood that is pumped out to the body is low in oxygen causing cyanosis. Cyanosis is the appearance of a blue or purple discoloration of skin and mucous membranes. It results from presence of 5.0 g/dL or greater of deoxyhemoglobin in the blood [3].

Although congenital cardiac malformations may be classified in various ways, we propose a simplified classification in four subgroups of malformations: septal defects, defects of the outflow tracts and great vessels, univentricular hearts, and other malformations.

1.1 Septal Defects

1.1.1 Atrial Septal Defects (ASD)

ASD are defects that allow interatrial communication and result from openings in the atrial septum. Defects of the atrial septum are the third most common type of congenital heart defect and the type most likely to be diagnosed in late childhood or adults (Fig. 1.1).

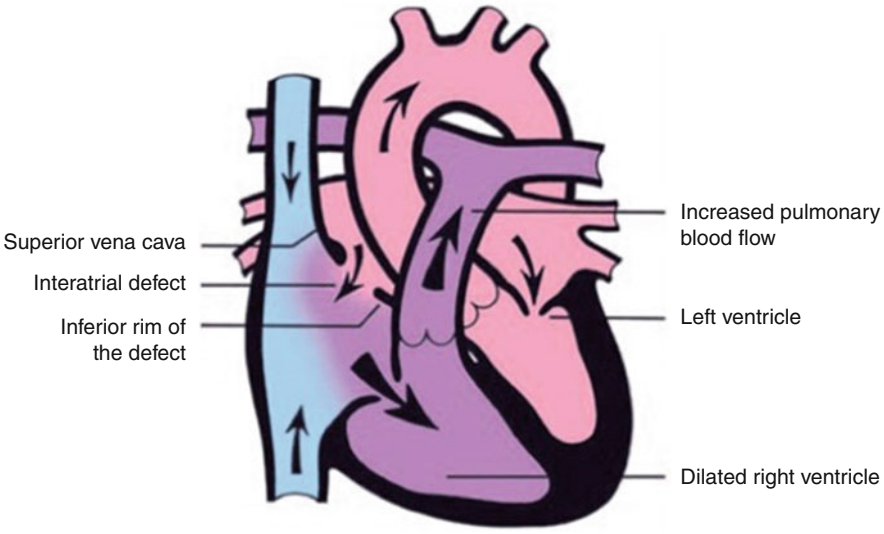

Fig. 1.1 Secundum atrial septal defect (Courtesy of the HeartLine Association)

Patent foramen ovale (PFO) is an interatrial communication that exists normally in the fetus where it allows blood to bypass the pulmonary circulation. Normally, this opening closes at birth when the lungs become functional. A PFO is seen in almost all newborns and with a decreasing frequency in older individuals [4, 5]. Complete anatomic closure of the foramen ovale occurs in 70–75 % of adults [6]. It assumes clinical importance in certain congenital heart defects and in older patients with paradoxical emboli and stroke.

Atrial septal defects are further classified into secundum and primum defects. Secundum atrial septal defect (ASD II) is a single or multiple defects within a part of the septum named septum primum. With the exception of PFO, secundum ASD is the most common cause of an atrial-level shunt. The shunt causes volume loading of the RA and RV, resulting in chamber enlargement [7]. Increased pulmonary blood flow over decades can damage the pulmonary vascular endothelium leading to an increase in pulmonary vascular resistance called pulmonary vascular obstructive disease. Most interatrial communications do not cause symptoms in childhood allowing some to go undetected until adulthood [8].

Closure of an ASD II can be accomplished by surgery or interventional catheterization [9]. Atrial arrhythmias are the most frequent late complication. Patients repaired early in life have a small risk for supraventricular tachycardia, and the risk increases with advancing age at repair. Pulmonary hypertension, another important late complication, is rare in patients operated before 25 years of age, and the risk increases with advancing age at repair [10].

1.1.2 Atrioventricular Endocardial Cushion Defect (AV Canal)

AV canal defect groups a spectrum of cardiac malformations derived from defects in the formation of the endocardial cushions.

The simplest malformation, ASD primum (ASD I) or incomplete AV canal, consists of an atrial septal defect located low in the atrial septum, adjacent to the mitral valve annulus, which is often associated with a cleft in the anterior leaflet of the mitral valve leading to mitral insufficiency (Fig. 1.2).

In other cases, the ostium primum type of defect is continuous with a larger defect in the adjacent ventricular septum. In these instances, the defect crosses both the mitral and tricuspid valvar annulae, causing deficiencies of the septal leaflets of both valves. This form of endocardial cushion defect is called complete AV canal.

Two major hemodynamic abnormalities are found. The first is the volume overload on the right atrium and right ventricle, and pulmonary overcirculation, as in patients with a left-to-right shunt at the atrial level. The second abnormality is mitral insufficiency, which leads to increased left ventricular volume because the left ventricle handles not only the normal cardiac output but also the regurgitated volume.

Infants with the complete form of AV canal frequently develop congestive cardiac failure in the first few weeks or months of life, whereas patients with ASD I may be asymptomatic, as in the ASD II.

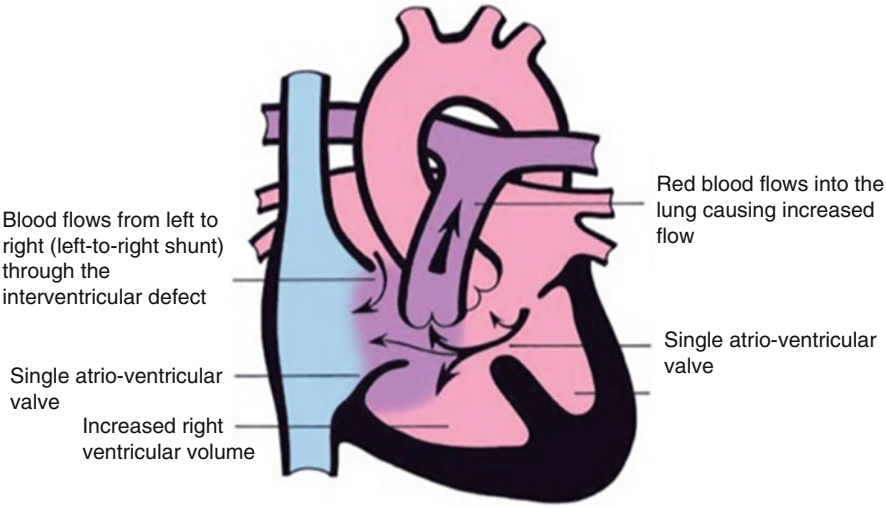

Fig. 1.2 Atrioventricular endocardial cushion defect (AV canal) (Courtesy of the HeartLine Association)

In patients with ASD I and a cleft mitral valve who are asymptomatic or who have few symptoms, corrective surgery can be delayed and can be performed at a low risk. The defect is closed; and the cleft of the mitral valve is sutured, which may greatly reduce the degree of mitral insufficiency. In patients with complete AV canal, corrective operation can be indicated in very young symptomatic infants who often respond poorly to medical management. Infants are routinely sent for operation at 2–3 months of age. The risk of pulmonary vascular disease developing within the first 6–9 months of life is high, especially in Down syndrome. The operative results are good in almost all, although some infants have such deficient anatomy of the mitral valve that prosthetic replacement of the mitral valve is required. Surgically induced AV block is likely but mostly uncommon.

Arrhythmias appear to have an earlier age of onset in patients with ASD I or repair AV canal defect than in other atrial shunts, likely due to concomitant left AV valve regurgitation. In addition, atrial arrhythmias are a common cause of deterioration [11, 12].

1.1.3 Ventricular Septal Defects (VSD)

A VSD is defined as a defect between the right and left ventricles. This defect may occur in isolation or, less commonly, may be part of a complex cardiac malformation.

The etiology of VSDs is felt to be multifactorial. Certainly, VSDs are quite prevalent in association with genetic abnormalities, especially in trisomies 13, 18, and 21 as well as other less common syndromes (Fig. 1.3).

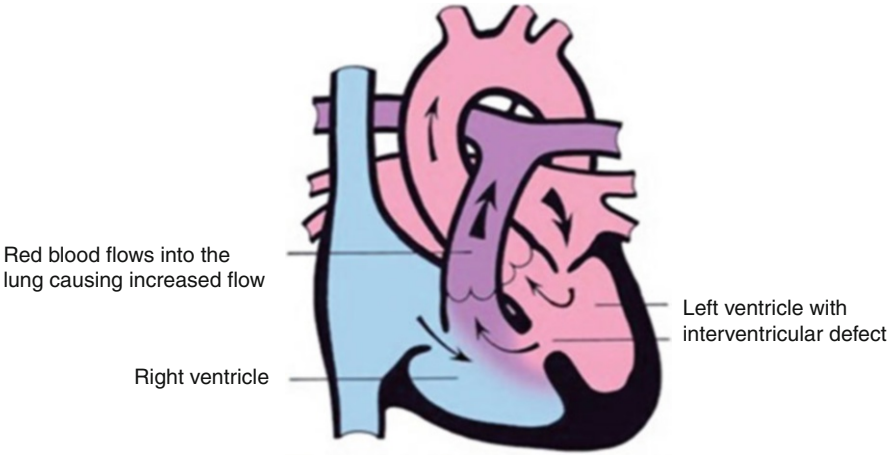

Fig. 1.3 Ventricular septal defect (Courtesy of the HeartLine Association)

Left-to-right shunting across a VSD provides greater blood flow to the pulmonary circulation than to the systemic circulation [13] leading to left ventricular volume overload and increased pulmonary blood flow. Increased pulmonary blood flow over time can damage the pulmonary vascular endothelium leading to an increase in pulmonary vascular resistance called pulmonary vascular obstructive disease. The magnitude of the left-to-right shunt depends mostly on VSD size: the bigger the defect, the larger the shunt.

Aortic regurgitation (AR) is an acquired and frequently progressive lesion in some patients with VSD [14, 15] usually due to prolapse of one or more valvular leaflets into the defect during systole.

Spontaneous closure of small VSD may occur. Closing a large VSD by open-heart surgery usually is done in infancy to prevent complications later. The transcatheter closure is a possible alternative in selected cases either in children or in adulthood.

Complete heart block secondary to injury to the conduction system during repair of a VSD may require a pacemaker in the postoperative period. The knowledge of the location of the conduction system in relationship to the defect now makes this a rare complication [16].

1.2 Defects of the Outflow Tracts and Great Vessels

1.2.1 Tetralogy of Fallot (ToF) and Pulmonary Atresia with Ventricular Septal Defect

Tetralogy of Fallot (ToF) constitutes 4–9 % of congenital heart disease and is the most common cyanotic congenital heart disease [14]. It involves four anatomic abnormalities of the heart such as (1) malaligned VSD, (2) anterior shift of the aorta

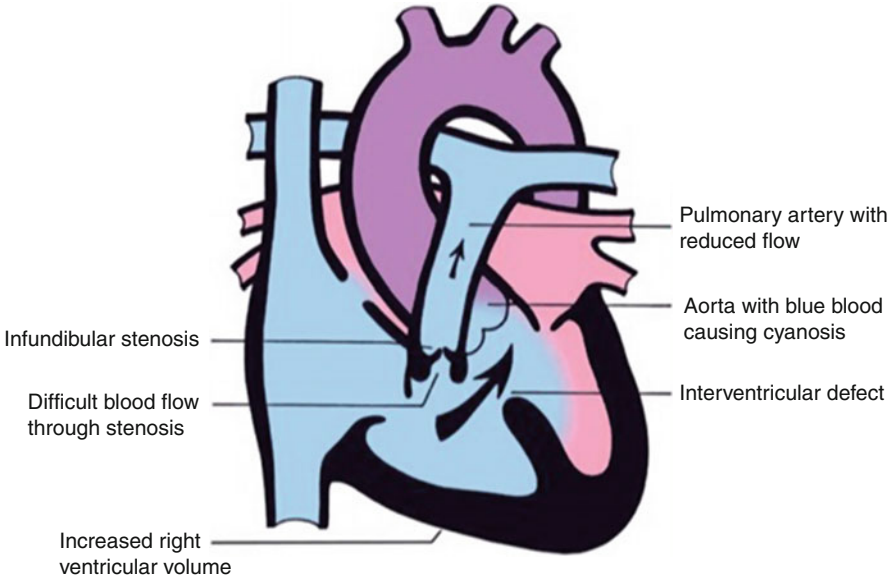

Fig. 1.4 Tetralogy of Fallot (Courtesy of the HeartLine Association)

over the VSD (overriding aorta), (3) obstruction of the right ventricular outflow tract (RVOT), and (4) right ventricular hypertrophy. Approximately 25 % have a right-sided aortic arch, and about 4 % have a coronary artery anomaly (Fig. 1.4).

The major right ventricular outflow obstruction in ToF is infundibular stenosis. The degree of cyanosis depends on the degree of right ventricular outflow obstruction. This is quite variable, from a slight obstruction to severe obstruction with pulmonary atresia. With mild pulmonary stenosis, also known as "pink tetralogy of Fallot," ToF behaves as a VSD with pulmonary overflow. As infundibular stenosis increases, progressive cyanosis due to less pulmonary blood flow develops [17]. ToF with pulmonary atresia, also known as pulmonary atresia with VSD, is a ToF severe variant [18] in which there is complete obstruction (atresia) of the right ventricular outflow tract. In this form of the disease, blood shunts completely from the right ventricle to the left where it is pumped only through the aorta (Fig. 1.5). The lungs are perfused via extensive collaterals from the systemic arteries and sometimes also via the ductus arteriosus.

Surgical treatment of the defect includes patch closure of the VSD and relief of pulmonary outflow obstruction with patch augmentation of the outflow at the expense of creation of free pulmonary regurgitation (PR). In selected cases with particular anatomic features, bypass of obstruction can be done using a right ventricle-to-pulmonary artery conduit (RV-PA conduit).

Total surgical correction can now be performed in young infants from 3 to 6 months of age or earlier [7]. Prognosis is good with total correction. After repair, the majority of patients have normal oxygen saturation and no residual shunt. The most common late complication is chronic PR. Residual RVOT obstruction and branch pulmonary artery stenosis [19] are less frequent but important late complications. Late right ventricle dilation and dysfunction are common [20].

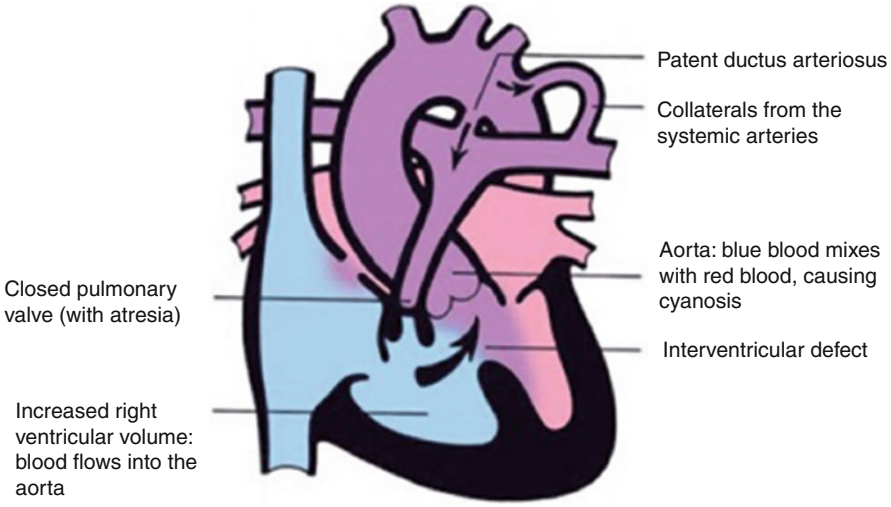

Fig. 1.5 Pulmonary atresia with ventricular septal defect (Courtesy of the HeartLine Association)

PV replacement is the treatment for chronic PR in patients with severe PR and RV dysfunction, or with symptomatic or sustained atrial or ventricular arrhythmias. Surgical valve replacement has been for a long time, the only option available for the native RV outflow tract. Nowadays, percutaneous pulmonary valve implantation (PPVI) is a valid alternative when the pulmonary valve must be changed, either in a native RVOT (in selected patients) or for a failed RV-PA conduit [21, 22]. Balloon dilation or stenting should be considered for branch pulmonary artery stenosis if flow in the artery is reduced and especially when accompanied by PR [23].

Progressive aortic regurgitation, usually associated with aortic root dilation, has been reported to occur in 15–18 % of patients [24, 25]. Arrhythmias are another important late complication. The most frequent are atrial flutter or fibrillation. Prevalence of ventricular arrhythmias increases with age and is associated with LV dysfunction [26]. Atrial arrhythmias can be addressed by a Maze procedure at the time of PV replacement [23]. Patients with documented sustained VT or aborted sudden cardiac death should receive an ICD for secondary prevention [27].

Survival after repair of TOF is less than expected for the general population at all times [28]. Sudden death due to ventricular arrhythmia is the most common cause of death after surgical repair of TOF [28]. The risk for sudden death is 3–6 % over the 25–30-year follow-up period [19].

Associated anomalies are common, such as DiGeorge syndrome.

1.2.2 Transposition of the Great Arteries (TGA)

TGA accounts for 4–5 % of all congenital heart defects [29]. Transposition derives from the Latin verb *transponere* meaning "to place across." That is, the great arteries are placed across the ventricular septum: the aorta arising from the right

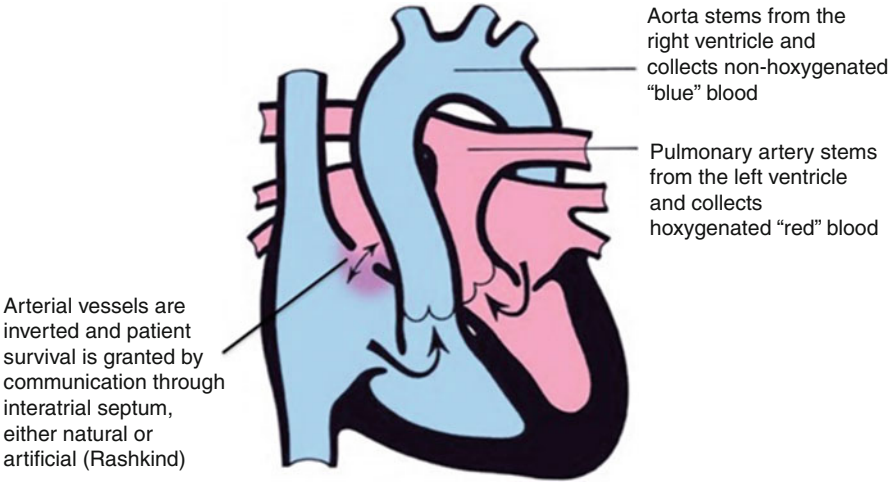

Fig. 1.6 Transposition of the great arteries (Courtesy of the HeartLine Association)

ventricle and the pulmonary arteries from the left ventricle. This is incompatible with life unless a communication exists between systemic and pulmonary circulation, as the two circulations are parallel and independent. During the newborn period, the patent ductus arteriosus (PDA) and PFO maintain this communication. As the PDA starts to close and the PFO by itself is inadequate in size, the patient develops intense cyanosis (Fig. 1.6).

At repair, the circulations are placed in series either by switching the inflow sources (atrial switch operation) or by switching the outflows (arterial switch operation and placement of a RV-PA conduit, also known as Rastelli operation). After an atrial switch operation, the right ventricle remains the systemic ventricle, while after an atrial switch operation, the left ventricle becomes the systemic ventricle. Patients born before the early 1980s most likely underwent an atrial switch operation; patients born after the late 1980s were most likely repaired using the atrial switch operation. Survival without surgery is unlikely. The arterial switch procedure offers the best prognosis with a mortality of about 5 %.

Sinus node dysfunction is the most frequent complication after an atrial switch operation. Loss of sinus node function is progressive, and by 20 years, only 40 % of patients remain in sinus rhythm [30]. Sinus node dysfunction with tachyarrhythmia or bradyarrhythmia is an indication for pacemaker therapy.

Complications related to Rastelli operation are mainly conduit obstruction and subaortic stenosis from inadequate enlargement of the VSD [31]. Several adult patients may face a right ventricular failure during their life, and a cardiac transplantation may become necessary as a treatment option.

Pulmonary artery stenosis is the most frequent complication following the arterial switch operation. Mechanisms include inadequate growth of the suture line, scarring and retraction of the material used to fill the coronary artery button sites, and tension at the anastomotic site if there is inadequate mobilization of the distal

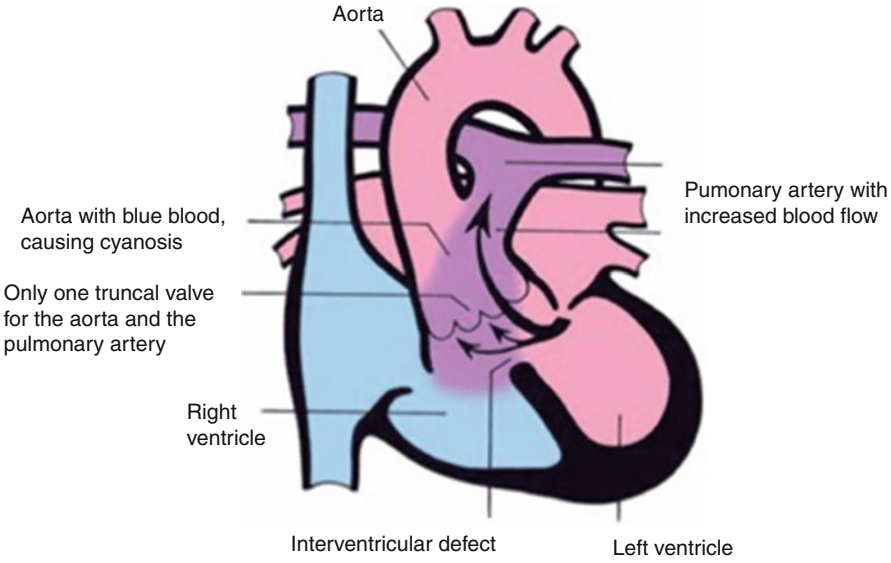

Fig. 1.7 Truncus arteriosus (Courtesy of the HeartLine Association)

pulmonary arteries [32]. Stent placement is usually effective for branch pulmonary artery stenosis.

There is a modest risk for neo-aortic valve regurgitation related in part to neo-aortic root dilatation, especially in patients with a VSD [41].

Coronary stenosis or occlusion has been discovered in 5–7 % of patients after the arterial switch operation and has been associated with ventricular dysfunction and sudden death [33].

1.2.3 Truncus Arteriosus

In truncus arteriosus, both the ascending aorta and main pulmonary artery or branch pulmonary arteries arise from a common trunk, positioned over a ventricular septal defect, that supplies systemic, coronary, and pulmonary circulations (Fig. 1.7). It accounts for about 1–4 % of the congenital heart defects [10]. Associated anomalies are common, such as DiGeorge syndrome.

Symptomatology depends upon the amount of pulmonary blood flow. With increased blood flow, symptoms of congestive heart failure develop few weeks after birth. Management consists of treatment of congestive heart failure followed by surgery. Repair of truncus arteriosus includes patch closure of the VSD so that the truncal valve is aligned solely with the left ventricle, separation of the main or branch pulmonary arteries from the truncal root, and establishment of continuity between the pulmonary arteries and the right ventricle, usually by means of a

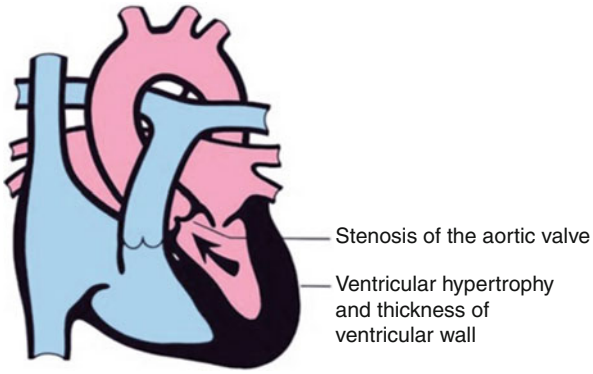

Fig. 1.8 Aortic stenosis (Courtesy of the HeartLine Association)

conduit or homograft. The prognosis is poor in untreated cases. After surgery, they will need long-term follow-up as they will eventually need to have the conduit graft replaced surgically or percutaneously.

1.2.4 Aortic Stenosis

Aortic valvar stenosis is related to either a unicuspid valve or more frequently to a congenitally bicuspid valve. The orifices of these abnormal valves are narrowed, accompanied by various degrees of aortic insufficiency in some patients. Patients with aortic stenosis are usually asymptomatic throughout childhood, even when stenosis is severe. Only 5 % of children with aortic stenosis develop congestive cardiac failure in the neonatal period, but it can develop later in childhood in patients who do not receive gradient relief. Exercise intolerance may occur. Some asymptomatic children, as they approach adolescence, may develop episodes of chest pain. These episodes signify myocardial ischemia and may precede sudden death. Syncope is another serious symptom of patients with aortic stenosis and may occur upon exercise. This symptom has also been associated with sudden death (Fig. 1.8).

Aortic valvar stenosis is progressive. Two processes probably account for this: the development of myocardial fibrosis and the decrease in size of the stenotic aortic valvar orifice by cartilaginous changes and ultimately by calcification of the valve.

Relief of the aortic stenosis gradient, either by balloon dilation or cardiac surgery, is indicated for patients with significant symptoms or for those whose catheterization data or echocardiogram indicates moderate or severe stenosis. In children, the stenotic valve is usually pliable enough for valvotomy or valvuloplasty so that an aortic valve replacement with a prosthesis or homograft is not required. Ultimately, children who have undergone aortic valvotomy may require a prosthesis or homograft in adulthood if the valve becomes calcified or rigid or, sooner, if the valve develops important insufficiency. No currently available replacement valve is perfect: mechanical prostheses are long-lived but thrombogenic, so anticoagulation is required; homograft valves, although free from thrombogenic complications, are often shorter-lived because of destruction by calcification at an unpredictable rate.

An alternative operation is the Ross autograft procedure, in which the patient's normal pulmonary valve is excised and placed in the aortic position. A homograft valve is placed in the pulmonary position, where performing balloon dilation or future surgical revision is less risky because of its more accessible anterior location and presence on the pulmonary side of the circulation. Limitations with the Ross operation are higher operative risk and longevity of the patient's native pulmonary valve functioning in the aortic position.

Subaortic stenosis is the second most common form of left ventricular outflow obstruction. This obstruction is a fibromuscular membrane with a small central orifice located in the left ventricle, usually within few centimeters of the aortic valve. A jet of blood passes through the orifice and strikes the aortic valve. The energy of the jet frequently results in alterations in the aortic valve and aortic insufficiency. Excision of the membrane is indicated in all patients to relieve the elevated left ventricular systolic pressure and to reduce the trauma to the aortic valve. The operative risk approaches that of operation for valvar aortic stenosis. The major hazard of the operation is the possibility of damage to the septal leaflet of the mitral valve, since the membrane is often attached to this leaflet. The results are generally very good, with near-normal left ventricular systolic pressure postoperatively. The subaortic membrane has a high recurrence rate after surgical correction mainly in patients operated sooner and with high peak pressure gradients, suggesting a more severe form of disease [34, 35].

Obstruction to left ventricular outflow can also result from supravalvar stenosis. In most of these patients, the ascending aorta narrows in an hourglass deformity. Although the abnormality is usually limited to the ascending aorta, other arteries, such as the brachiocephalic and even the renal arteries, may also be narrowed. Peripheral pulmonary arterial stenosis and hypoplasia may coexist and indeed represent the most important problem. The systolic pressure is elevated in the ascending aorta proximal to the obstruction. Therefore, the coronary arteries are submitted to an elevated systolic pressure that can lead to tortuosity of the coronary arteries and to premature atherosclerosis. Two factors have been implicated in the etiology of this condition. The first is Williams syndrome, in which a defect in the elastin gene is present. The second is familial supravalvar aortic stenosis, which occurs in patients who do not have Williams syndrome but they probably carry a mutated elastin gene as well. Surgery may be indicated for a lesser gradient compared with aortic valvar stenosis or if symptoms related to myocardial ischemia are present. Operative relief of the obstruction in the ascending aorta can be accomplished by surgical widening of the narrowing with a patch. Over the long term, reobstruction can occur because of progressive medial thickening of affected vessels.

1.2.5 Pulmonary Stenosis

Pulmonary stenosis occurs at three sites in the right heart outflow area: below the pulmonary valve (infundibular), at the level of the valve (valvar), or above the valve (supravalvar).

Fig. 1.9 Pulmonary stenosis (Courtesy of the HeartLine Association)

Infundibular pulmonary stenosis rarely occurs as an isolated lesion. Supravalvar stenosis or stenosis of the individual pulmonary arteries is also uncommon. In most patients, obstruction occurs at the level of the pulmonary valve.

Regardless of the anatomic type of stenosis, the right ventricular systolic pressure must increase to maintain a normal cardiac output. With the elevation of right ventricular systolic pressure, right ventricular hypertrophy develops (Fig. 1.9).

In the usual form of pulmonary stenosis, the valve cusps are fused; and the valve appears domed in systole. A small central orifice and poststenotic dilation are found. In less than 10 % of valvar pulmonary stenosis, the pulmonary valve leaflets are dysplastic: they do not show commissural fusion, the commissures are rather open, but each leaflet is greatly thickened and redundant. In most patients, dysplastic pulmonary valve is associated with various noncardiac abnormalities of Noonan and similar genetic syndromes. Critical pulmonary stenosis presents with cyanosis and failure in the neonatal period or within the first year of life. However, many of the patients are completely asymptomatic throughout childhood. Cardiac catheterization is required to perform balloon dilation, which is so far the procedure of choice for gradient relief. This procedure almost always results in a favorable outcome and reduction of right ventricular systolic pressure to normal or near normal. Even though pulmonary valvar insufficiency regularly results from valvuloplasty, it is well tolerated because pulmonary arterial pressure is low. Operative valvotomy is indicated for those patients who have failed dilation (e.g., Noonan syndrome patients with dysplastic valves) or who are not candidates for balloon dilation (e.g., the neonate with critical stenosis and an extremely hypoplastic pulmonary annulus instead requires outflow tract widening by use of a patch).

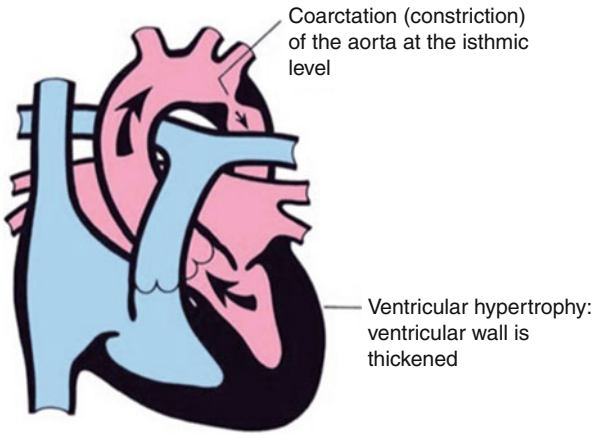

Fig. 1.10 Coarctation of the aorta (Courtesy of the HeartLine Association)

Stenosis also occurs above the pulmonary valve and in the branches of the pulmonary arteries. One or more major branches may be involved, showing either a long area of narrowing or a discrete narrowing. Peripheral pulmonary artery stenosis occurs in children with supravalvar aortic stenosis, particularly those with Williams syndrome, in patients with Alagille syndrome, or appears without apparent cause. Hypoplastic pulmonary arteries frequently accompany ToF with pulmonary valve atresia; these patients often have DiGeorge syndrome. Most patients with this condition are asymptomatic. The prognosis is extremely variable. Since the degree of stenosis is often mild and does not increase with age in most patients, it has been considered a benign condition. Apparent growth of the pulmonary arteries does occur in some; rarely, especially in Williams syndrome patients, stenosis may progress in severity. Most patients do not require surgery as the degree of stenosis is not severe. Treatment with catheter balloon dilation, sometimes with placement of endovascular metal stents, is widely used, although with variable results that depend greatly on the etiology and severity of the stenosis.

1.2.6 Coarctation of the Aorta

Coarctation of the aorta results from constriction of the tissue of the distal aortic arch at the junction with the descending aorta and near the insertion of the ductus arteriosus [36]. Often, discrete coarctation is associated with tubular hypoplasia of the isthmus and/or aortic arch (Fig. 1.10). The bicuspid aortic valve is frequently associated with aortic coarctation (85 %) [37]. Obstruction of the arch causes hypertension proximal to the obstruction and reduced blood flow and pressure distal. In adults, large collateral vessels develop that bypass the obstruction and maintain adequate resting flow to the lower body. There are several surgical techniques used to repair a coarctation of the aorta. If the coarcted segment is short and discrete, resection and end-to-end anastomosis of the proximal and distal ends is possible. If the coarctation is a long tubular obstruction, resection with interposition of a tube

graft would be necessary. In the young infant, sacrificing the left subclavian artery, and using the transected blood vessel as a graft by turning it down and sewing it into the aortic wall, was popular at one time [38]. Percutaneous balloon dilation eventually with stent positioning is indicated for recurrent and or native coarctation in adolescent and adult patients with discrete lesions [39]. Late complications include systemic hypertension, recurrence of coarctation, premature coronary atherosclerosis, stroke, aortic aneurysm formation or dissection, and endocarditis [40, 41]. Hypertension persists in up to one-third of patients after successful resolution of coarctation [42].

1.2.7 Ebstein

Ebstein anomaly is characterized by downward displacement of the septal and posterior leaflets of the tricuspid valve that are attached to the right ventricular septum. The anterior leaflet is elongated and is displaced downward within the right ventricular cavity causing "atrialization of the right ventricle" (i.e., the right ventricle is small). The "atrialized" ventricle is enlarged and thin [43–46], while the functional RV distal to the valve is variably hypoplastic [45]. The result most often is tricuspid regurgitation (TR), but in some cases, tricuspid stenosis is predominant [46].

The functional impairment of the right ventricle and regurgitation of the tricuspid valve retard forward flow of blood through the right heart. The overall effect is right atrium dilation and increased right atrial pressure, thus favoring a right-to-left shunt across the interatrial communication and/or reduced systemic cardiac output. Cyanosis depends upon the right-to-left shunt.

Associated lesions include an interatrial communication and less commonly VSD or PDA. In more complex forms of Ebstein anomaly, pulmonary valve and pulmonary artery stenosis or atresia or left-sided abnormalities such as mitral stenosis or regurgitation can be seen [47]. The conduction system is often abnormal. Transcatheter ablation is the standard treatment of an accessory pathway or other arrhythmia substrate, but success rates tend to be lower and recurrence rates higher than in the structurally normal heart [48].

While fetal or neonatal clinical presentation of Ebstein anomaly is associated with a poor outcome [49, 50], adults have a much better prognosis [51, 52]. Prognosis depends on the severity of the lesion: it is good with mild lesions and poor with severe lesions with other associated anomalies/malformations (the most frequent situation when clinical symptoms appear in neonates). Treatment is mainly palliative, and there are no good surgical options. Medical therapy of Ebstein anomaly is limited to management of complications. In older patients, tricuspid annuloplasty and rarely tricuspid valve replacement may be performed. Surgical mortality in adult patients is low, under 3 % in the current era [37]. Atrial arrhythmia is the most frequent early complication occurring in one-third of postoperative patients [54]. Recurrent hospitalizations are frequent with arrhythmia being the most common indication for readmission [54].

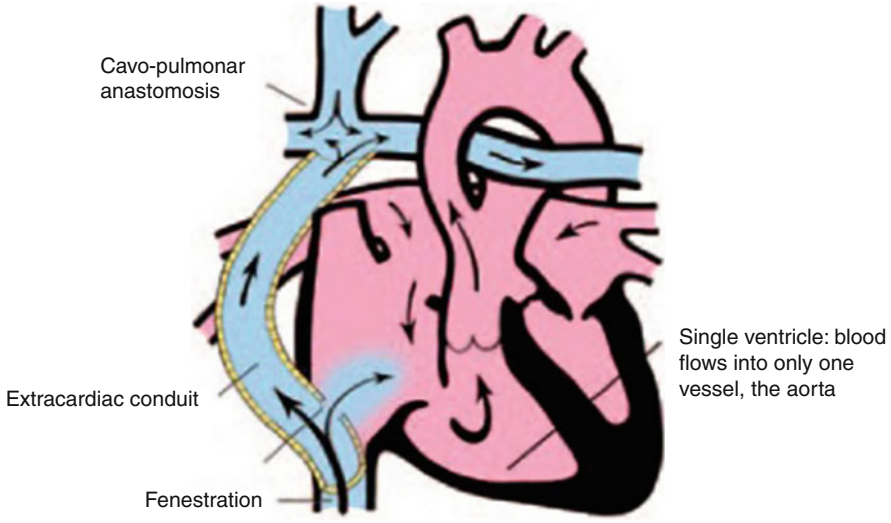

Fig. 1.11 Univentricular heart (Courtesy of the HeartLine Association)

1.2.8 Univentricular Hearts

Hearts with one functional ventricle are rare, comprising 1–2 % of all congenital heart defects. The physiology in infancy depends on several factors, including pulmonary blood flow, systemic outflow obstruction, pulmonary venous anomalies, and the atrioventricular valve function (Fig. 1.11). The Fontan procedure is the established final palliative cardiac surgical procedure in the treatment of patients with a functionally univentricular heart. In the current era, a bidirectional cavopulmonary shunt is created during infancy. Then the Fontan procedure is completed during childhood by using either a lateral tunnel or an extracardiac conduit, which is placed to redirect inferior vena caval blood to the pulmonary arteries [52]. Congenital heart defects typically staged toward this type of palliation include tricuspid or mitral atresia, double-inlet left ventricle, common AV canal with unbalanced ventricles, hypoplastic left or right heart syndrome, hypoplastic right heart syndrome, and other more rare defects.

Patients with Fontan physiology are at risk for multiple complications. Those with an atriopulmonary connection are at particularly high risk for right atrium dilation with thrombus formation and atrial arrhythmias. Both problems lead to diminished cardiac output, reduced capacity, and diminished quality of life. Heart transplantation is effective in Fontan patients with intractable arrhythmias, advanced heart failure, and protein-losing enteropathy.

Acknowledgment The figures were first published in *Heart Children*: *a practical handbook for parents* [55] and are published here with the permission of Heartline Families, which holds intellectual property rights.

References

1. Hoffman JI, Kaplan S (2002) The incidence of congenital heart disease. J Am Coll Cardiol 39(12):1890–1900
2. Pacifico AD, Soto B, Bargeron LM (1979) Surgical treatment of straddling tricuspid valves. Circulation 60:655–664
3. Green M (1998) Chapter 48: Cyanosis. In: Green M (ed) Pediatric diagnosis. W.B. Saunders, Philadelphia, p 389–395
4. Connuck D, Sun JP, Super DM et al (2002) Incidence of patent ductus arteriosus and patent foramen ovale in normal infants. Am J Cardiol 89:244–247
5. Fisher DC, Fisher EA, Budd JH et al (1995) The incidence of patent foramen ovale in 1,000 consecutive patients. A contrast transesophageal echocardiography study. Chest 107:1504–1509
6. Schneider B, Zienkiewicz T, Jansen V et al (1996) Diagnosis of patent foramen ovale by transesophageal echocardiography and correlation with autopsy findings. Am J Cardiol 77:1202–1209
7. Koenig P, Hijazi Z, Zimmerman F (2004) Essential pediatric cardiology. McGraw-Hill Publishing, New York
8. John Sutton MG, Tajik AJ, McGoon DC (1981) Atrial septal defect in patients ages 60 years or older: operative results and long-term postoperative follow-up. Circulation 64:402–409
9. Butera G, Romagnoli E, Carminati M, Chessa M, Piazza L et al (2008) Treatment of isolated secundum atrial septal defects: impact of age and defect morphology in 1,013 consecutive patients. Am Heart J 156:706–712
10. Hoffman JI, Kaplan S, Liberthson RR (2004) Prevalence of congenital heart disease. Am Heart J 147:425–439
11. Gatzoulis MA, Hechter S, Webb GD, Williams WG (1999) Surgery for partial atrioventricular septal defect in the adult. Ann Thorac Surg 67:504–510
12. Bergin ML, Warnes CA, Tajik AJ, Danielson GK (1995) Partial atrioventricular canal defect: long-term follow-up after initial repair in patients > or = 40 years old. J Am Coll Cardiol 25:1189–1194
13. Mas MS, Bricker JT (1990) Clinical physiology of left-to-right shunts. In: Garson Jr A, Bricker JT, McNamara DG (eds) The science and practice of pediatric cardiology. Lea and Febiger, Philadelphia, p 999–1001
14. Keane JF, Plauth WH, Nadas AS (1977) Ventricular septal defect with aortic regurgitation. Circulation 56(1):172–177
15. Rhodes LA, Keane JF, Keane JP et al (1990) Long follow-up (to 43 years) of ventricular septal defect with audible aortic regurgitation. Am J Cardiol 66:340–345
16. Chessa M, Butera G, Negura D, Bussadori C, Giamberti A, Fesslova V, Carminati M (2009) Transcatheter closure of congenital ventricular septal defects in adult: mid-term results and complications. Int J Cardiol 133(1):70–73
17. Flyer DC (1992) Chapter 30. Tetralogy of Fallot. In: Flyer DC (ed) Nadas' pediatric cardiology. Hanley & Belfus, Inc, New York, p 471–491
18. Farouk A, Zahka K, Siwik E, Erenberg F, Al-Khatib Y, Golden A, Karimi M, Uddin M, Hennein HA (2009) Individualized approach to the surgical treatment of tetralogy of Fallot with pulmonary atresia. Cardiol Young 19(1):76–85
19. Warnes CA, Williams RG, Bashore TM, Child JS, Connolly HM et al (2008) ACC/AHA 2008 guidelines for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Practice Guidelines (writing committee to develop guidelines on the management of adults with congenital heart disease). Circulation 118:e714–e833
20. Graham TP Jr, Cordell D, Atwood GF, Boucek RJ Jr, Boerth RC et al (1976) Right ventricular volume characteristics before and after palliative and reparative operation in tetralogy of Fallot. Circulation 54:417–423
21. Dave HH, Buechel ER, Dodge-Khatami A, Kadner A, Rousson V et al (2005) Early insertion of a pulmonary valve for chronic regurgitation helps restoration of ventricular dimensions. Ann Thorac Surg 80:1615–1620

22. Oosterhof T, van Straten A, Vliegen HW, Meijboom FJ, van Dijk AP et al (2007) Preoperative thresholds for pulmonary valve replacement in patients with corrected tetralogy of Fallot using cardiovascular magnetic resonance. Circulation 116:545–551
23. Stulak JM, Dearani JA, Burkhart HM, Park SJ, Suri RM, Schaff HV (2012) The surgical treatment of concomitant atrial arrhythmias during redo cardiac operations. Ann Thorac Surg 94(6):1894–1899
24. Niwa K, Siu SC, Webb GD, Gatzoulis MA (2002) Progressive aortic root dilatation in adults late after repair of tetralogy of Fallot. Circulation 106:1374–1378
25. Rieker RP, Berman MA, Stansel HC Jr (1975) Postoperative studies in patients with tetralogy of Fallot. Ann Thorac Surg 19:17–26
26. Khairy P, Aboulhosn J, Gurvitz MZ, Opotowsky AR, Mongeon FP, Kay J, Valente AM, Earing MG, Lui G, Gersony DR, Cook S, Ting JG, Nickolaus MJ, Webb G, Landzberg MJ, Broberg CS (2010) Alliance for Adult Research in Congenital Cardiology (AARCC). Arrhythmia burden in adults with surgically repaired tetralogy of Fallot: a multiinstitutional study. Circulation 122(9):868–875
27. Alexander ME, Cecchin F, Walsh EP, Triedman JK, Bevilacqua LM et al (2004) Implications of implantable cardioverter defibrillator therapy in congenital heart disease and pediatrics. J Cardiovasc Electrophysiol 15:72–76
28. Nollert G, Fischlein T, Bouterwek S, Bohmer C, Klinner W et al (1997) Long-term survival in patients with repair of tetralogy of Fallot: 36-year follow-up of 490 survivors of the first year after surgical repair. J Am Coll Cardiol 30:1374–1383
29. Hoffman JIE, Christianson R (1978) Congenital heart disease in a cohort of 19,502 births with long-term follow-up. Ann J Cardiol 42:461
30. Gelatt M, Hamilton RM, McCrindle BW, Connelly M, Davis A et al (1997) Arrhythmia and mortality after the mustard procedure: a 30-year single-center experience. J Am Coll Cardiol 29:194–201
31. Brown JW, Ruzmetov M, Huynh D, Rodefeld MD, Turrentine MW et al (2011) Rastelli operation for transposition of the great arteries with ventricular septal defect and pulmonary stenosis. Ann Thorac Surg 91:188–193
32. Prifti E, Crucean A, Bonacchi M, Bernabei M, Murzi B et al (2002) Early and long term outcome of the arterial switch operation for transposition of the great arteries: predictors and functional evaluation. Eur J Cardiothorac Surg 22:864–873
33. Mayer JE Jr, Sanders SP, Jonas RA, Castaneda AR, Wernovsky G (1990) Coronary artery pattern and outcome of arterial switch operation for transposition of the great arteries. Circulation 82:IV139–IV145
34. Lopes R, Lourenco P, Goncalves A, Cruz C, Maciel MJ (2011) The natural history of congenital subaortic stenosis. Congenit Heart Dis 6(5):417–423
35. Geva A, McMahon CJ, Gauvreau K, Mohammed L, del Nido PJ, Geva T (2007) Risk factors for reoperation after repair of discrete subaortic stenosis in children. J Am Coll Cardiol 50(15):1498–1504
36. Brom AG (1965) Narrowing of the aortic isthmus and enlargement of the mind. J Thorac Cardiovasc Surg 50:166
37. Duffie ER Jr, Wilson JF et al (1968) Coarctation of the aorta. In: Moss AJ, Adams FH (eds) Heart disease in infants, children, and adolescents. The Williams and Wilkins Company, Baltimore, p 386
38. Reddy M, McElhinney DB, Silverman NH et al (1998) Partial biventricular repair for complex congenital heart defects: an intermediate option for complicated anatomy or functionally borderline right complex heart. J Thorac Cardiovasc Surg 116:21–27
39. Tanous D, Benson LN, Horlick EM (2009) Coarctation of the aorta: evaluation and management. Curr Opin Cardiol 24:509–515
40. Vriend JW, Mulder BJ (2005) Late complications in patients after repair of aortic coarctation: implications for management. Int J Cardiol 101:399–406
41. Krieger EV, Stout K (2010) The adult with repaired coarctation of the aorta. Heart 96:1676–1681

42. Musto C, Cifarelli A, Pucci E, Paladini S, De Felice F et al (2008) Endovascular treatment of aortic coarctation: long-term effects on hypertension. Int J Cardio 130:420–425
43. Lev M, Liberthson RR, Joseph RH, Seten CE, Eckner FA et al (1970) The pathologic anatomy of Ebstein's disease. Arch Pathol 90:334–343
44. Attenhofer Jost CH, Connolly HM, Dearani JA, Edwards WD, Danielson GK (2007) Ebstein's anomaly. Circulation 115:277–285
45. Schreiber C, Cook A, Ho SY, Augustin N, Anderson RH (1999) Morphologic spectrum of Ebstein's malformation: revisitation relative to surgical repair. J Thorac Cardiovasc Surg 117:148–155
46. Dearani JA, Danielson GK (2000) Congenital heart surgery nomenclature and database project: Ebstein's anomaly and tricuspid valve disease. Ann Thorac Surg 69:S106–S117
47. Attenhofer Jost CH, Connolly HM, Edwards WD, Hayes D, Warnes CA et al (2005) Ebstein's anomaly – review of a multifaceted congenital cardiac condition. Swiss Med Wkly 135:269–281
48. Chetaille P, Walsh EP, Triedman JK (2004) Outcomes of radiofrequency catheter ablation of atrioventricular reciprocating tachycardia in patients with congenital heart disease. Heart Rhythm 1:168–173
49. Celermajer DS, Bull C, Till JA, Cullen S, Vassillikos VP et al (1994) Ebstein's anomaly: presentation and outcome from fetus to adult. J Am Coll Cardiol 23:170–176
50. Kumar AE, Fyler DC, Miettinen OS, Nadas AS (1971) Ebstein's anomaly. Clinical profile and natural history. Am J Cardiol 28:84–95
51. Attie F, Rosas M, Rijlaarsdam M, Buendia A, Zabal C et al (2000) The adult patient with Ebstein anomaly. Outcome in 72 unoperated patients. Med (Baltimore) 79:27–36
52. World J (2013) Functionally univentricular heart and the fontan operation: lessons learned about patterns of practice and outcomes from the congenital heart surgery databases of the European association for cardio-thoracic surgery and the society of thoracic surgeons. Pediatr Congenit Heart Dis 4(4):349–355
53. Brown ML, Dearani JA, Danielson GK, Cetta F, Connolly HM et al (2008) The outcomes of operations for 539 patients with Ebstein anomaly. J Thorac Cardiovasc Surg 135(1120–1136):1136
54. Brown ML, Dearani JA, Danielson GK, Cetta F, Connolly HM et al (2008) Functional status after operation for Ebstein anomaly: the Mayo Clinic experience. J Am Coll Cardiol 52:460–466
55. Rees P, Tunstill A, Pope T, Kinnear D (2002) Heart children: a practical handbook for parents. HeartLine Association, Camberley