Congenital Heart Disease

The Nursing Care Handbook

Serena Francesca Flocco
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Foreword

It is our great delight and distinct honor to write the foreword to this book dedicated to the nursing care of the patient with congenital heart disease. To the best of our knowledge, this is the first book that is dedicated to nursing for the emergent population of patients with congenital heart disease. Indeed, advances in medicine have resulted in increased life expectancy, which yields a growing number of patients in need for lifelong care. In addition to the numerous medical issues that are associated with the management of people with congenital heart disease, these patients and families also face a lot of nonmedical problems. Nurses typically play an important role on the verge of both areas and are complementary to other healthcare professionals involved in the care for these patients.

The authors are to be congratulated for their prescience in editing this important contribution to the body of knowledge. This book assembles a large panel of authors with expertise in the different aspects of nursing care for patients with congenital heart disease. Doing so, well-known topics are covered, but also issues that are sometimes underestimated are addressed. The importance of this book is that it tries to make sure that such aspects are not ignored in the care of our patients.

Some chapters in this book pertain to technical aspects in nursing care for afflicted patients. They address the different paths of a daily clinical work-up. Other chapters focus on nonmedical aspects. Not surprisingly, a chapter on transfer and transition is included. Indeed, nurses play a key role in the transition of young patients and their families and in the preparation for the transfer to adult care. The book concludes with a chapter on nursing research and quality improvement initiatives, showing how deeply involved nurses are in the improvement of care to patients with congenital heart disease.

Never before has such a concentrated effort been made to provide the reader with a state-of-the-art review of nursing care in congenital heart disease. It is our pleasure and privilege to warmly recommend this textbook to all nurses who want to improve their skills in managing the problems of these patients.

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Contents

Congenital Heart Disease Classification, Epidemiology, Diagnosis, Treatment, and Outcome

1

Angelo Micheletti

Abstract

Congenital heart disease (CHD) is a problem in the structure of the heart that is present at birth. CHD is still the most common inborn defect with an approximate prevalence at birth of 5–11 per 1000 live births and incidence of 1%. The aim of this chapter is to give both a general overview on CHD and to address just a few specific issues for each type of structural disease. Congenital cardiac malformations may be classified in different ways; to highlight the underlying anatomy and pathophysiology, the diseases could be grouped as follows: (a) CHD with shunt between systemic and pulmonary circulation, (b) left heart CHD, (c) right heart CHD, (d) CHD with anomalous origin of great arteries, and (e) miscellanea.

1.1 Introduction

Congenital heart disease (CHD) is still the most common inborn defect with an approximate prevalence at birth of 5–11 per 1000 live births and incidence of 1% [1]. By definition, CHD means a disease that has been present since birth but not necessary "clinically" evident since birth: this is the case, for example, of moderate size atrial septal defect of unobstructive subaortic stenosis.

Taking into account the pathophysiology and diagnosis of CHD, it is helpful to highlight three key points: (1) the presence of shunt between arterial and venous blood, (2) the presence of cyanosis, and (3) the changes in circulation after birth [2]. A shunt consists of an abnormal communication between two cardiac chambers or vessels allowing blood to go from one side to the other. Shunting may be described

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as left-to-right, right-to-left, or bidirectional. The direction is strictly dependent on pressure gradient across the shunt; most commonly blood goes from high-pressure left-sided heart to low-pressure right-sided heart. Therefore shunt direction affects the status of pulmonary blood flow which can vary from normal, increased, or decreased. Any large left-to-right (L-R) shunt results in increased blood flow into the lungs associated with shortness of breath and prominent vascular markings on chest x-ray, volume overload of left ventricle (LV) associated with chamber dilatation, and subsequently heart failure; if untreated, it will eventually result in pulmonary artery (PA) pressure overload which, by time, will cause irreversible structural arterial wall changes ending up with pulmonary hypertension (PH), with increased pulmonary vascular resistance (PVR), and, later on, in pulmonary vascular obstructive disease (PVOD). High and fixed PVR is the feature of pulmonary vascular obstructive disease. When PVR approaches or even exceeds systemic vascular resistance, direction of the shunting becomes bidirectional or mainly right-to-left (R-L), condition known as Eisenmenger syndrome. Generally, in presence of right-to-left shunt, venous poor-oxygenated blood mixes with arterial high-oxygenated blood, causing cyanosis. Cyanosis is a bluish discoloration of the skin and mucous membranes resulting from presence of 5.0 g/dL or greater of deoxyhemoglobin in the blood.

Presence of shunts is crucial during fetal life when the placenta provides the exchange of gases and nutrients and lungs receive only about 15% of combined ventricular output. Fetal circulation is characterized by four sites of shunting: the placenta, ductus venosus through which umbilical vein drains into inferior vena cava, foramen ovale within the interatrial septum, and the arterial duct through which blood in the PA flows into descending aorta. Just after birth, placental circulation disappears and pulmonary circulation is established. Interruption of the umbilical cord results in an increase in systemic vascular resistance and closure of the ductus venosus. Concomitant lung expansion results in reduction of pulmonary artery pressure and PVR, an increase in pulmonary blood flow, functional closure of the foramen ovale, and closure of patent arterial duct due to increased arterial oxygen saturation.

With lung expansion and the resulting increase of alveolar oxygen tension, an initial significant rapid fall in PVR occurs mainly due to the vasodilating oxygen effect on pulmonary vasculature. Later on, between 4 and 8 weeks after birth, there is another slower fall in the PVR and PA pressure secondary to wall changes in pulmonary arterioles. Many neonatal conditions associated with different forms of CHD, causing inadequate oxygenation, may interfere with the normal pulmonary arteriole maturation, resulting in persistent pulmonary hypertension or delay in usual PVR fall.

Ductus arteriosus usually closes spontaneously within the first 48 h after birth, by constriction of the medial smooth muscle; after this functional closure, an anatomical closure occurs, by 2–3 weeks of age, by permanent changes in the endothelium and subintimal layers. Many factors may interfere with ductal closure such as oxygen, maturity of the newborn, prostaglandin E_2 levels, and acidosis [3].

To achieve a precise diagnosis of CHD, even in very complex cases, along with an accurate physical examination, many both noninvasive and invasive tools are currently available. Transthoracic echocardiography (TTE) is the first-line diagnostic technique in terms of anatomical and functional information in all CHD, whereas cardiac catheterization still constitutes the final definitive diagnostic test for many

of them. Over the last decade, due to the significant improvement of interventional cardiology in dealing with several forms of CHD, a new "multimodality" imaging approach has been promoted aiming at the integration rather than at the selection of the necessary details. 3D echocardiography, cardiac computed tomography (CT), and magnetic resonance (MR) have become of great interest due to their ability to generate both 3D anatomical and hemodynamic functional information.

Effective treatment of all the spectrum of CHD is currently feasible due to the amazing improvement in medical care, catheter-based interventions, and surgical procedures which have dramatically extended survival and life expectancy. Survival through childhood is now common even in the most complex and lethal malformations, such as hypoplastic left heart syndrome. As a consequence of these advances, there have been major demographic shifts, so that adult patients with CHD now outnumber children even with complex forms of CHD [4].

The aim of this chapter is to give both a general overview on CHD and to address just a few specific issues for each type of structural disease. Congenital cardiac malformations may be classified in different ways; to highlight the underlying anatomy and pathophysiology, the diseases could be grouped as follows: (a) CHD with shunt between systemic and pulmonary circulation, (b) left heart CHD, (c) right heart CHD, (d) CHD with anomalous origin of great arteries, and (e) miscellanea.

1.1.1 CHD with Shunt Between Systemic and Pulmonary Circulation

1.1.1.1 Atrial Septal Defect

Atrial septal defect (ASD) is a communication between the atrial chambers permitting left-to-right shunting (Fig. 1.1).

It is the second most common type of CHD, accounting for about 7–10% of all CHD patients, more prevalent in women (2:1), and most likely to be diagnosed in late childhood or adults. There are different types of ASD (Fig. 1.2) [5]:

- Ostium secundum ASD: The most frequent variant of ASD, 70–80% of all ASDs, results from deficiency of the flap valve of the oval fossa, ranging from incomplete development, with failed overlapping to the septum secundum, to the presence of multiple fenestrations to complete absence. Anomalous pulmonary venous return is present in about 10% of cases.
- Ostium primum ASD: Occurs in about 15% of all ASDs and is a part of atrioventricular septal defect; it is placed near the crux.
- Sinus venosus defect: 5–10%, located superiorly and posteriorly, near the entry of superior vena cava (SVC) or inferiorly and posteriorly near the entry of inferior vena cava (IVC), commonly associated with anomalous drainage of right pulmonary veins.
- Coronary sinus ASD: 1%, located in the roof of the coronary sinus which could be partially or completely missing and often associated with left SVC that drains to the left atrium.

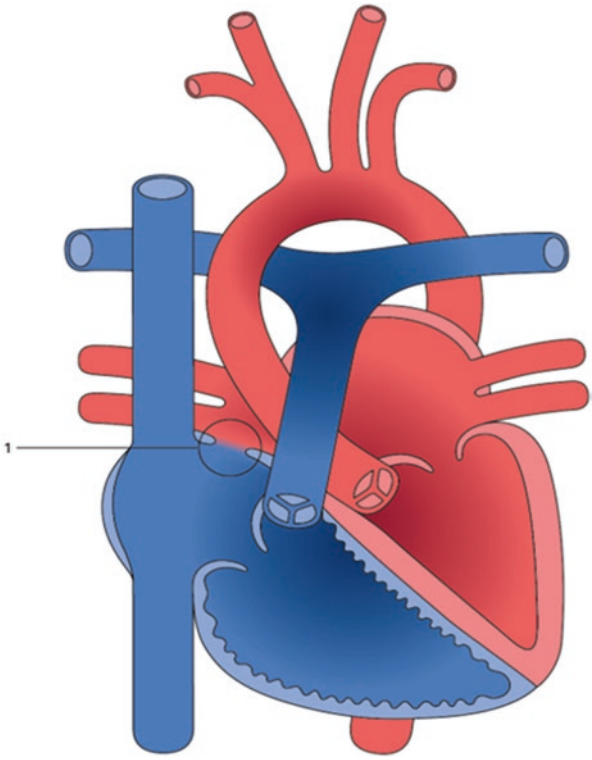

Fig. 1.1 Atrial septal defect (1)

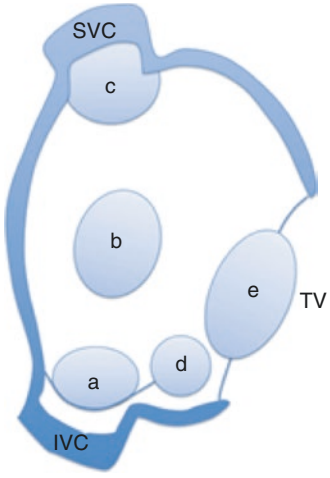

Fig. 1.2 Different types of ASD

a - Inferior sinus venosus defect,
b – ostium secundum ASD,
c – superior sinus venosus defect,
d – coronary sinus defect,
e – ostium primum ASD.
SVC: Superior vena cava.
IVC: Inferior vena cava.
TV: tricuspid valve

The L-R shunt causes volume overload of the right chambers and pulmonary circulation; the entity of shunt is related to defect size, right ventricle (RV) and LV compliance and pressure. Usually volume overload is well tolerated allowing some to go undetected until adulthood; when the shunt is significant, however, with age, symptoms can occur such as exertional dyspnea, pulmonary infections, and atrial arrhythmias [6].

Foramen ovale is an interatrial communication, during fetal life, between septum secundum (limbus of fossa ovalis) and septum primum (the valve of fossa ovalis) which allows venous return from IVC to shunt across to the left atrium. Normally it closes at birth functionally, whereas a complete anatomic closure occurs in 70–75% of adults [7]. Patent foramen ovale (PFO) refers to persistence of this normal communication postnatally; it assumes clinical importance in certain CHD and in patients with cerebral vascular accident due to paradoxical emboli [8].

Diagnosis. From childhood, the classic clinical findings include widely split and fixed second heart sound associated with a low-pitched systolic ejection murmur. TTE is the diagnostic test of choice. In case of PH, diagnostic cardiac catheterization is mandatory to calculate pulmonary vascular resistance and to assess pulmonary circulation vasoreactivity.

Treatment. The presence of hemodynamically significant L-R shunt (a ratio of pulmonary blood flow to systemic blood flow of >1.5:1.0) and/or right chamber volume overload without significant PH represents the indication for ASD closure. Closure can be accomplished by surgery or, in case of secundum ASD and adequate anatomic rims, by device implantation during interventional catheterization [9].

Outcome. Atrial arrhythmias are the most frequent late complication, and the risk increases with advancing age at repair. PH is another late complication affecting survival but is rare in patients operated before 25 years of age, and the risk increases with advancing age at repair as well [10]. If closure is performed in adolescence or childhood, life expectancy returns to normal [11].

1.1.1.2 Ventricular Septal Defect

Ventricular septal defect (VSD) is defined as a communication between the two ventricles (Fig. 1.3). It is the most common CHD, almost 20% of all defects, and may occur in isolation or as a part of a complex cardiac malformation [10]. According to the anatomical position, different types of VSD can be described (Fig. 1.3):

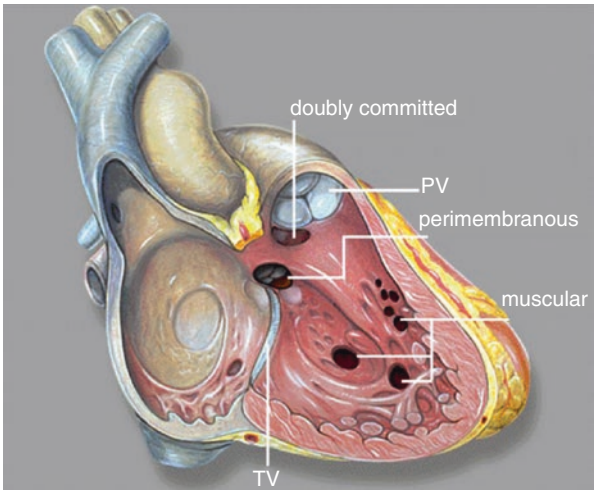

Fig. 1.3 Different types of VSD according to anatomical position within the septum viewed from RV side. *TV* tricuspid valve. *PV* pulmonary valve

- Perimembranous VSD: The most frequent variant, placed within the membranous septum with variable anatomical extension toward the other components of interventricular septum (IVS), inlet, outlet, and apical. It is in close relationship with the septal leaflet of the tricuspid valve (TV) and the his bundle of electrical conduction system. Aneurysm of membranous septum is frequently associated and may result, over time, in partial or complete closure.
- Muscular VSD: About 15–18% of all VSDs, could be placed within any muscular component of IVS, and is surrounded by complete muscular rims. Quite often small defects close spontaneously; rarely the defects can be multiple, the so-called "Swiss-cheese" septum.
- Doubly committed, juxta-arterial VSD: More frequent in Asian countries, it is localized just beneath the arterial valves which are, as a consequence, in fibrous continuity.
- Inlet defect: Located in the inlet component of the septum, immediately inferior to the atrioventricular valve apparatus; typically occurring in down syndrome.

Shunt direction and magnitude are determined by the size of the defect, ventricular systolic and diastolic function, the presence of right ventricular outflow tract obstruction (RVOT), and PVR.

The clinical spectrum of this defect is wide: large L-to-R shunt causes high pulmonary blood flow, LV volume overload presenting with heart failure, and, particularly in infancy and childhood, failure to thrive and repeated respiratory infections. Large VSD with large L-R shunt is associated with PH and, if untreated, ends up in pulmonary vascular disease as in Eisenmenger syndrome. In case of doubly committed VSD, less common perimembranous, there is a risk for prolapse of the right coronary or noncoronary cusp of the aortic valve, resulting in progressing aortic regurgitation [12]. Subaortic discrete obstruction and/or right mid-ventricular discrete stenosis (double-chambered RV) may develop, the latter being the result of a high-velocity VSD jet lesion on RV endothelium. Finally arrhythmias can occur although less frequently than other forms of CHD.

Diagnosis. The classic pansystolic murmur, which is best audible along the left mid to lower sternal border, may not be heard until a few weeks of age when the shunt becomes maximal. Generally, the smaller is the defect, the higher is the murmur intensity. A second sound physiologically split with respiration is indicative of normal pulmonary pressure. In POVD associated with VSD, varying degrees of cyanosis may be noted. On chest x-ray, the degree of cardiomegaly and the increase in pulmonary vascular markings directly relate to the magnitude of the L-R shunt. TTE is mandatory to assess all the characteristics in terms of position, size, number, association with other congenital defects or aortic valve dysfunction, direction of the shunting, relationship with tricuspid valve and its cordal apparatus, the presence of aneurysmal tissue, and the distance between VSD and aortic valve [13]. In some selected cases, a diagnostic cardiac catheterization is needed to calculate PVR and to assess pulmonary circulation vasoreactivity.

Treatment. Spontaneous closure of small, restrictive VSD may occur. Closing moderate to large VSD by open heart surgery is usually performed during infancy to

prevent complications later [14]. Although surgery remains the treatment of choice, with low operative mortality, transcatheter closure has become a valid and alternative procedure to be considered only in selected cases either in children or in adults [15].

Outcome. The long-term prognosis for a patient with a successful surgically repaired VSD is excellent with life expectancy as good as in the general population [16]. Acute and/or late-onset complete atrioventricular block (cAVB) due to conduction system injury remains a crucial issue after surgical or transcatheter closure of perimembranous VSD; the incidence of cAVB, after device closure, requiring permanent pacemaker implantation is 2.6%, and the risk is higher in children aged <6 years [17–19]. The prognosis for patients with Eisenmenger syndrome is the worst among the VSD population.

1.1.1.3 Atrioventricular Septal Defect

Atrioventricular septal defect (AVSD) refers to a spectrum of cardiac malformations characterized by abnormal development of atrioventricular junction which normally derives from endocardial cushion tissue. It is clearly associated with chromosomal abnormalities, mainly trisomy 21, Down syndrome.

The key anatomic feature is the presence of common AV junction guarded by a 5-leaflet common AV valve, associated with deficient septation. Arrangement of common AV valve leaflets differs significantly from the normal tricuspid and mitral valves. Anatomy can vary from:

- Partial AVSD, with an ostium primum ASD and two separate valve orifices and without inlet VSD
- Complete AVSD, with an ostium primum ASD, single valve orifice, and inlet VSD

The predominant hemodynamic consequence of this abnormal anatomy is the presence of intracardiac shunting between either the atriums or the ventricles or from ventricles to atriums. This will result in volume and/or pressure overload of cardiac chambers which may be exacerbated by the regurgitation of one or more components of the common AV valve. The incidence of associated lesions is high, approaching 50% of the cases. Symptoms are usually the result of intracardiac left-to-right shunting and/or regurgitation of AV valve; patients with only an atrial level shunt are rarely symptomatic, whereas signs of congestive heart failure (CHF) as tachypnea, dyspnea, diaphoresis, failure to thrive, and frequent chest infections are typical of significant ventricular level shunt and/or significant AV valve regurgitation.

Diagnosis. Due to the presence of a common AV junction, the AV conduction system is typically displaced posteriorly and inferiorly. On ECG, it results in superior and rightward QRS axis deviation frequently associated with prolonged PR interval and QRS duration.

On chest x-ray, cardiomegaly and increased pulmonary vascular markings reflect significant left-to right intracardiac shunting and/or left AV valve regurgitation; the presence of prominent central pulmonary arteries combined with peripheral “pruning” and right heart enlargement reflects Eisenmenger syndrome.

Comprehensive and sequential 2D and 3D TTE [20] should be focused on all the anatomical details and to rule out unbalanced ventricles, outflow tract obstructions, and associated lesions.

Diagnostic cardiac catheterization is usually reserved for all the cases in whom there is concern about PVR.

Treatment. Medical management with diuretics and ACE inhibitors is usually required for complete AVSD while waiting for surgical repair although the use of afterload reduction with ACE inhibitors is still controversial. In most infants, failure to thrive is an indication for repair which is usually performed within the first 3–6 months. Patients with unrepaired AVSD and Eisenmenger syndrome respond to pulmonary vasodilator therapy after diagnostic catheterization [21].

Outcome. Currently, excellent short-term survival after AVSD repair is the norm.

In the postoperative AVSD patients, the most common causes of late morbidity and mortality and consequently need of reoperation are dysfunction of the left AV valve (mainly regurgitation), subaortic stenosis, residual septal defects, and heart block [22]. Early mortality has been reported at 4% after reoperation during childhood; the highest risk is in those patients requiring left AV valve replacement [23]. Atrial arrhythmias appear to have an earlier age of onset after AVSD repair than in other atrial shunts, likely due to concomitant left AV valve dysfunction [24].

1.1.1.4 Patent Ductus Arteriosus

Patent ductus arteriosus (PDA) is a vascular structure which connects the descending aorta to the roof of pulmonary arterial trunk near the origin of left pulmonary artery. Usually it closes spontaneously within the first 48 h after birth, by constriction of the medial smooth muscle due to the acute increase in oxygen tension and reduction in prostaglandin E_2 and I_2 levels. Subsequently, during the following 2–3 weeks, muscle fibers are replaced by connective tissue resulting in formation of ligamentum arteriosum. If this does not occur, there is a patent ductus arteriosus (PDA). It accounts for 5–10% of all CHD in children, more frequent in females than in males [25]. Considering the anatomy, it can vary significantly in its shape, size, and attachment to the aorta; usually its narrowest part is the pulmonary arterial end. The most common type is a funnel-shaped duct with a localized narrowing at the pulmonary artery junction. PDA results in L-R shunt with potential left heart volume overload and pulmonary artery pressure overload. Clinical features depend on size, length, and age at presentation; especially the size should be correlated to the age and weight of the patient. Small PDA is usually not associated with either symptoms or signs of volume/pressure overload. Moderate and large PDA may present predominantly with left heart failure due to volume overload or right heart failure due to PA and RV pressure overload. Large PDA, if not closed in time, generally develops in Eisenmenger syndrome.

Diagnosis. In case of large PDA, peripheral arterial pulses are bounding with a rapid upstroke; pulse pressure is wide because of rapid runoff of the blood from the systemic circulation to the pulmonary circulation. In a patient with small to moderate PDA, only a systolic murmur may be heard, whereas, in a large shunt, a continuous machinery-type murmur is audible at the upper left sternal edge; the systolic

component sound is a crescendo, and the diastolic component sound is a decrescendo. A normal ECG or left ventricular hypertrophy (LVH) is seen with small to moderate PDA. Biventricular hypertrophy is seen with large PDA and pulmonary hypertension. Classically, chest x-ray reveals enlarged cardiac silhouette when the relative pulmonary-to-systemic flow ratio is >2:1; cardiomegaly occurs with the enlargement of left atrium (LA) and left ventricle and ascending aorta. Pulmonary vascular markings are increased. TTE evaluation gives accurate information about ductal anatomy and physiology and is practically feasible in almost all infants, in children, and in many adults.

Treatment. PDA should be closed in patients with signs of LV volume overload even if asymptomatic and in patients with PAH but PAP < 2/3 of systemic pressure or PVR < 2/3 of systemic vascular resistance (SVR). Closure should be avoided in "silent" duct, very small with no murmur, and in PDA-Eisenmenger. Percutaneous closure can be performed in those cases who meet certain criteria concerning body weight and ductal size; it is feasible and safe in children weighing more than 5 kg with PDA diameter of 2.5–3 mm. In smaller-weight babies, this option is still under debate [26, 27], whereas larger PDAs are currently best managed with surgery (via thoracotomy).

Percutaneous closure is the first choice treatment in adults, even if cardiac surgery is indicated due to other concomitant cardiac lesions, because calcification of the duct increases surgical complications. The PDA anatomical characteristics guide the choice of proper occlusion device because different types are worldwide currently available. Small ducts can be closed using coils such as Cook detachable coil, whereas large PDAs (Fig. 1.4) can be addressed, according to the anatomy, using different devices such as an Amplatzer Duct Occluder (ADO) I device or its modification ADO II (Fig. 1.5) [28] or a mVSD Amplatzer Occluder or an ASD Amplatzer Occluder [29] or the new Occlutech(®) PDA Occluder [30].

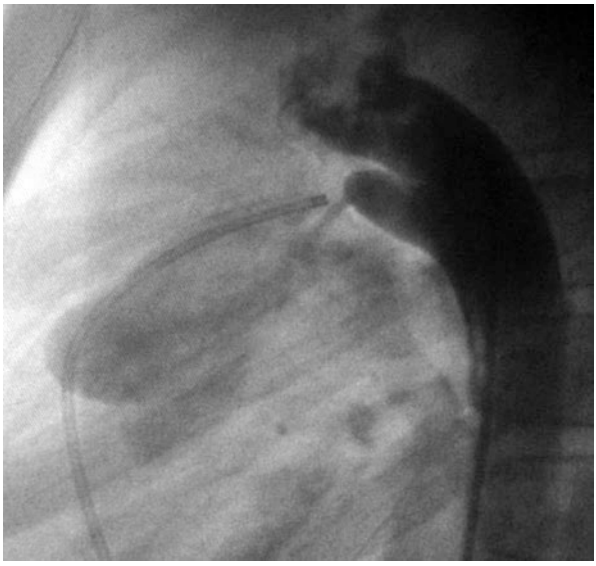

Fig. 1.4 Fluoroscopy, lateral view, aortic angiogram: PDA

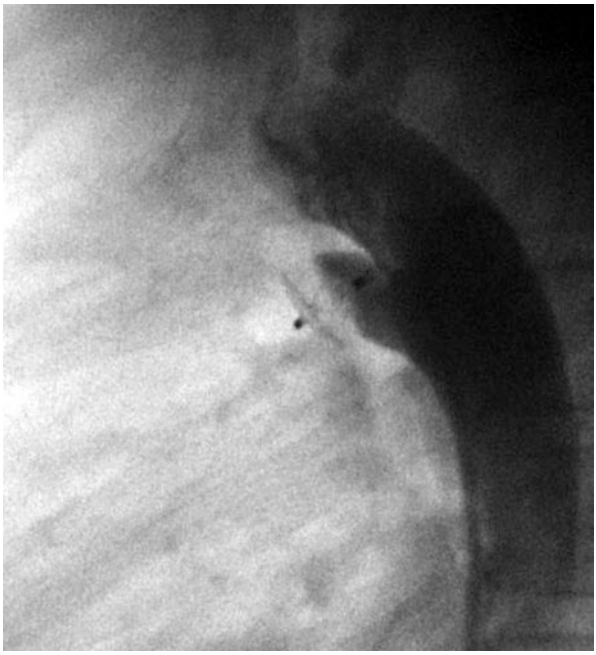

Fig. 1.5 Percutaneous closure with Amplatzer Duct Occluder II device

Outcome. Early and mid-term complications after transcatheter closure may include device embolization or dislocation. Embolization can occur more often after coil release; according to the size, coil can embolize in the pulmonary arteries or in the aorta and subsequently in all the arterial vessels as the intracranial arteries. In case of dislocation or malposition, device can cause left pulmonary artery stenosis or aortic stenosis associated, quite often, to residual shunting. Hemolysis is another rare and serious complication usually after coil PDA occlusion, related to a residual flow at the end of the procedure. If it happens, it is difficult to eliminate as long as the residual shunt is closed. At present, overall device occlusion rate is about 95%. Early complications after surgical closure may include recurrent laryngeal nerve palsy, chylothorax, pneumothorax, and immediate LV dysfunction. Untreated PDA may lead to POVD and Eisenmenger condition. Aneurysm of the ductus is a rare complication that develops gradually.

1.1.1.5 Aortopulmonary Window

Aortopulmonary window (APW) is a large defect between the ascending aorta and the main PA which results from failure of the spiral septum to completely divide the embryonic truncus arteriosus (Fig. 1.6). It represents 0.2–0.6% of all CHDs. In approximately half of the patients, there are associated cardiac abnormalities, most commonly VSD, interrupted aortic arch, aortic coarctation, and tetralogy of Fallot [31]. In isolated APW, clinical features are similar to those of large PDA.

Diagnosis. In isolated APW, peripheral pulses are bounding, but the heart murmur is of the systolic ejection type rather than continuous murmur at the base. On

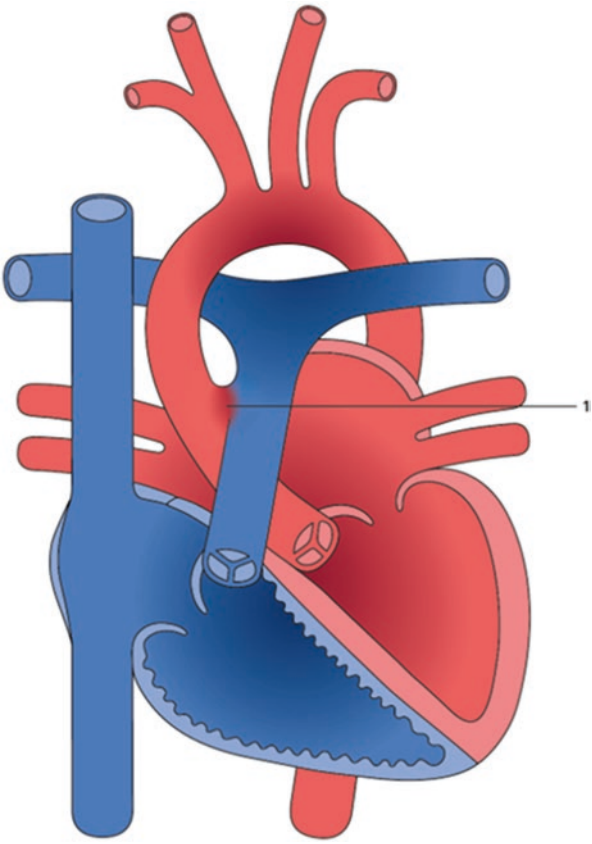

Fig. 1.6 Aortopulmonary window (1)

ECG, biventricular hypertrophy is seen. Classically, chest x-ray reveals enlarged cardiac silhouette due to the enlargement of left chambers; pulmonary vascular markings are markedly increased. TTE evaluation gives accurate information about the anatomy of APW which always should be ruled out in case of unexplained pulmonary hypertension. Diagnostic cardiac catheterization is usually reserved for all the cases in whom there is concern about PVR.

Treatment. This defect has no tendency to reduce or close spontaneously; therefore prompt surgical closure is indicated when the diagnosis is made as long as PVR is normal. In some selected cases of isolated APW with favorable anatomy, percutaneous device closure can be achieved [32].

Outcome. Without intervention, 40–50% of all patients will die due to congestive heart failure during the first year of life, and a large number of survivors will suffer from sequelae of congestive heart failure or pulmonary vascular disease later on. Outcomes of early repair of APW are excellent, including infants with complex associated cardiac lesions which should contemporarily be repaired. Cardiac reoperation can be required in complex APW, mainly with concomitant arch repair, and is usually related to aortic obstruction [33].

1.1.2 Left-Heart Congenital Heart Disease

1.1.2.1 With Inflow Obstruction

Cor Triatriatum Sinister

Cor triatriatum sinister (CTS) is a rare congenital abnormality characterized by the left atrium being divided into two chambers by a fibrous membrane, a proximal chamber that receives the pulmonary venous drainage and a distal chamber that contains the mitral valve and the left atrial appendage. CTS can occur in isolation (classic) or in association with other congenital cardiac anomalies (atypical) such as an ASD [34]. The atypical form occurs in 50–85% of all patients with CTS, although it is rare in adolescents and adults. Older patients more often have an isolated type of CTS. Symptoms are related to the size of fenestrations within the fibrous membrane and therefore to the degree of obstruction; presenting symptoms can mimic those seen in mitral stenosis and are related to both pulmonary venous and pulmonary arterial hypertension [35].

Diagnosis. Physical findings include dyspnea, a loud S2 and basal lung crackles. ECG usually shows right axis deviation and right ventricular hypertrophy (RVH). On chest x-ray, there is evidence of pulmonary venous congestion or pulmonary edema, prominent PA, and varying degree of right heart enlargement. TTE is mandatory to assess the intra-atrial membrane characteristics and the presence of associated CHD. In the differential diagnosis, a supramitral membrane is distinguished from the CTS membrane by its location below the left atrial appendage. This is an important differentiation, because, as a result of its proximity to the mitral valve and left circumflex coronary artery, the resection of a supramitral membrane is more difficult than in case of CTS membrane.

Treatment. Surgery is the definitive treatment and should be considered at any age if there are any associated symptoms or complications.

Outcome. Surgery in patients with CTS is performed with good outcomes. Mortality and reoperation rate are nearly 0% in patients with classic CTS and are determined by the repair of associated anomalies in atypical CTS [36]. The risk of recurrent intra-atrial obstruction postoperatively is low [37]. Pulmonary hypertension regresses rapidly in survivors if the correction is made early.

Congenital Mitral Valve Stenosis

Different anatomic types are described [38]:

- Congenital mitral valve (MV) stenosis: Extremely rare, it usually involves abnormalities of one or more components of the valve apparatus (Fig. 1.7).
- Supravalvar mitral ring: A thin fibrous membrane partially or completely encircles the mitral orifice and adheres to the leaflets. It is most commonly associated with “shone complex” which comprises parachute MV, subaortic stenosis, and coarctation of the aorta.
- Parachute MV: In the most typical and frequent form, all the chordal attachments are to a single papillary muscle (Fig. 1.8).

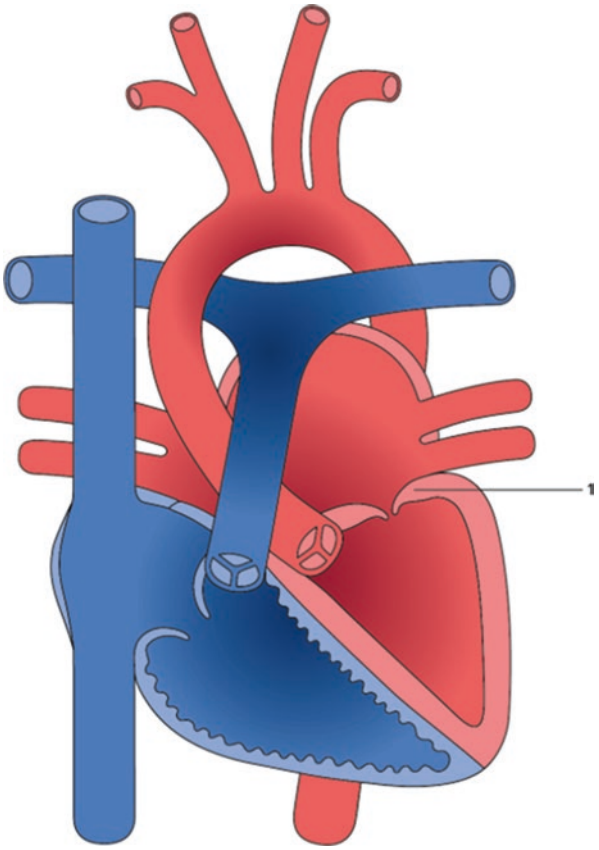

Fig. 1.7 Congenital mitral valve dysplasia and stenosis (1)

- Double-orifice MV: Two complete mitral orifices are supported by their own tension apparatus. The orifices are usually unbalanced with the smaller one directed to the anterolateral commissure. Rarely it may be associated with AVSD or VSD and coarctation.
- Arcade MV: The leaflets insert directly into the papillary muscles or the ventricular wall without chordal attachments, resulting in stenosis and regurgitation.

Symptoms and clinical signs clearly reflect the degree of pulmonary venous congestion, pulmonary hypertension, and right heart failure. The onset strictly depends on the severity of the stenosis: severe stenosis becomes evident in the neonatal period (failure to thrive, diaphoresis, cough, and recurrent respiratory infections), whereas mild or moderate disease usually presents later on in infancy, childhood, or adolescence.

Diagnosis. The typical murmur is an apical low-pitched rumbling mid-diastolic murmur. In presence of PH, a right ventricular heave is usually palpable. ECG during infancy may be normal; as the disease progresses, LA enlargement, RVH, and RA enlargement gradually appear. With ongoing atrium dilatation, atrial

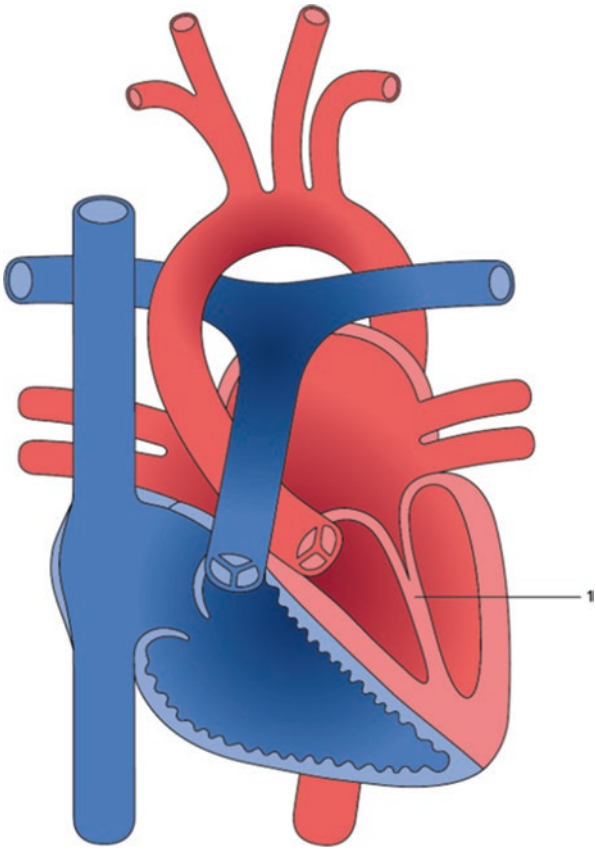

Fig. 1.8 Parachute mitral valve (1)

arrhythmias may occur. Chest x-ray shows dilatation of LA, PA, and RV; in severe disease, Kerley's B lines may be evident. TTE is diagnostic of the condition in terms of anatomic and functional MV details. Cardiac catheterization is indicated when there is discrepancy between echo measurements, hemodynamics, and clinical status or to ascertain the presence of POVD and its reversibility.

Treatment. Patients may require diuretics to alleviate pulmonary venous congestions. In case of severe obstruction, surgical valve repair or replacement with a prosthetic valve may be indicated but carries morbidity and mortality [39]. Only a few selected patients with hypoplastic annulus may benefit from balloon angioplasty which delays the need for surgical valve repair or replacement [40]. Valve choices for infants are limited: a mechanical valve is preferred with the caveat that long-term anticoagulation is necessary. For supravalvar mitral ring, early surgical repair should be considered in the presence of severe heart failure. Isolated double-orifice MV may never require intervention. MV arcade is usually repairable making the need of replacement quite rare.

Outcome. The prognosis after valve repair or replacement depends on many factors: patient age and size, the degree of annual hypoplasia, ventricular size and

performance, severity of PH, and presence of other lesions [41]. MV replacement in neonates and infants is associated with an early mortality rate of approximately 10–20% at 6 months [42]; furthermore, prosthetic valves need to be replaced within 5–10 years due to the infant's growth and prosthetic stenosis or pannus formation. Similar data are reported in children: 50% of patients require prosthesis replacement by 10 years post-valve implantation [43].

In adults following percutaneous valvuloplasty, the survival rate is 80–90% in those with favorable MV morphology. Bioprosthetic valves last approximately 5–10 years, whereas mechanical valves last a lifetime. Five-year survival rate in adults after valve replacement exceed 70% unless they have coinciding complex CHD.

1.1.2.2 Congenital Mitral Valve Regurgitation

It is most often associated with other congenital cardiac abnormalities. Mitral valve prolapse (MVP) is rare in infants and extremely rare in neonates; exceptions are cases associated with Marfan syndrome or other connective tissue disorders as Ehlers-Danlos syndrome or osteogenesis imperfecta [44]. In MVP leaflets extend beyond the annular plane during ventricular systole; any portion of MV leaflets can be affected resulting in varying degrees of regurgitation.

Along with aortic dilation, MVP is the cardiovascular abnormality typical of Marfan syndrome which is a heritable autosomal dominant connective tissue disorder causing histologic and morphologic changes in fibrillin structure. Both the anterior and posterior leaflets become elongated and redundant; chordal rupture, progressive annular dilation, and calcification may occur.

Timing of MVP presentation is variable and depends on the degree of regurgitation. Due to left atrial dilation, left main stem bronchial compression, elevated pulmonary capillary hydrostatic pressure, atrial arrhythmias, and respiratory symptoms occur. Children with severe MV regurgitation may present with orthopnea, dyspnea, chest pain, palpitations, reduced exercise tolerance, or syncope.

Diagnosis. A regurgitant holosystolic murmur is audible at the apex with radiation to the left axilla and left back. A loud S3 and an apical rumble may be present as well. In case of moderate to severe regurgitation, ECG and CXR may show hypertrophy and dilation of left chambers. The main tool used for diagnosing congenital MV regurgitation is echocardiography for both qualitative and quantitative assessment.

Treatment. Patients presenting with congestive heart failure require diuretics and ACE inhibitors to reduce afterload and to improve cardiac output [45]. Isolated MVP and cleft mitral valve rarely require intervention in infants or children unless Marfan syndrome coexists; in this case, MV replacement may be indicated depending on the severity of regurgitation. Percutaneous MV repair has been emerging as an option for the treatment of MV regurgitation in adults; current modalities include MV repair by placing metal clip on the leaflets and MV annuloplasty by placing into the coronary sinus to the great cardiac vein in order to circle three-fourths of the entire annulus [46].

MV is typically repaired prior to the need for replacement; multiple techniques are nowadays available for repair. Replacement is most commonly performed using mechanical valves, necessitating lifelong anticoagulation.

Outcome. Cleft MV and MVP, without intervention, have excellent short-term outcomes because they feature no progression of insufficiency except in the presence of connective tissue abnormalities. Short- and immediate-term outcomes after repairing a cleft MV are excellent, and most patients require no additional interventions. Patients with Marfan syndrome and MV involvement should be closely monitored keeping in mind the associated complications, in particular of the aortic valve, aorta, ventricular function, and arrhythmias; aortic aneurysm dissection or rupture is rare in childhood and more typically occurs in the third and fourth decades of life [47].

1.1.2.3 With Outflow Obstruction

Aortic Stenosis

Aortic stenosis (AS) represents 3–6% of all patients with CHD, and it occurs more often in males (male-female ratio of 4:1). Stenosis may be at the valvular (70%), subvalvular (23%), or supravalvular (7%) level.

Valvular AS is usually caused by bicuspid aortic valve with a fused commissure and an eccentric orifice [48] (Fig. 1.9). Less common is the unicuspid valve with

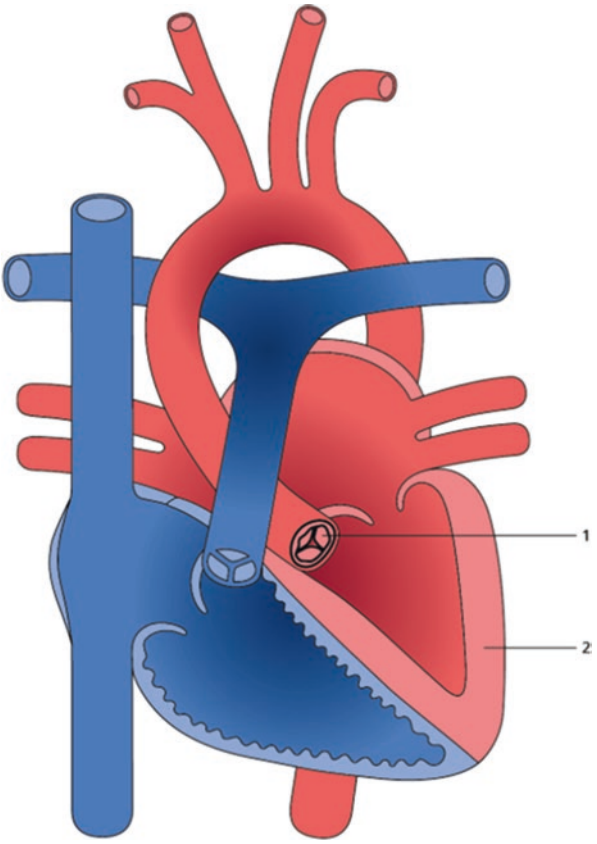

Fig. 1.9 Congenital aortic valve stenosis: (1) Valvular AS. (2) Left ventricular hypertrophy

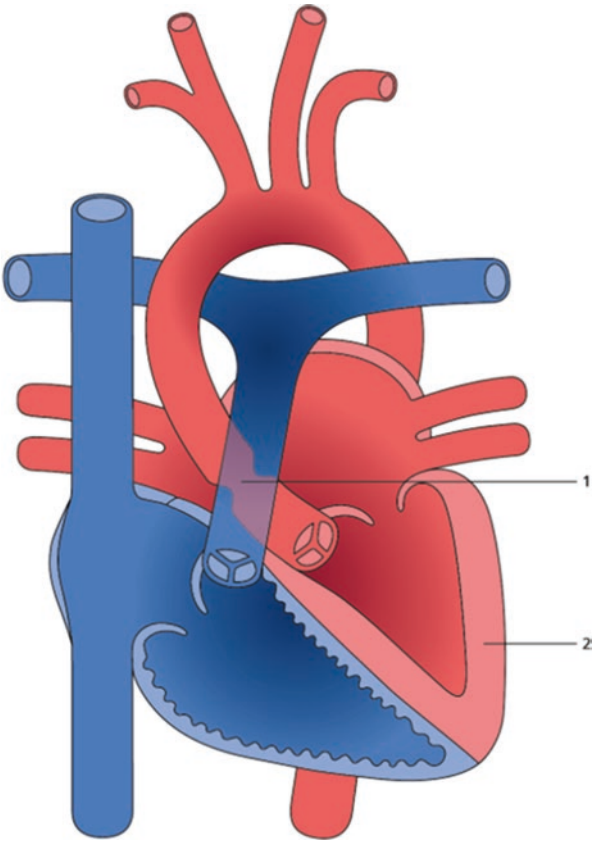

Fig. 1.10 Supravalvular aortic stenosis: (1) Supravalvular AS. (2) LV hypertrophy

one lateral attachment, while a tricuspid valve with a central stenotic orifice is the least common form.

Subvalvular stenosis may result from discrete membranous diaphragm, usually associated with other CHDs like VSD, PDA, or coarctation, or, less commonly, from tunnel-like fibromuscular narrowing of left ventricular outflow tract. The latter is usually associated with other LV anomalies including Shone complex.

In supravalvular AS the constriction occurs at the upper margin of the sinus of Valsalva, and aorta narrows in an hourglass deformity (Fig. 1.10). This is often associated with Williams-Beuren syndrome, an elastin protein gene disorder, which includes characteristic facies, mental retardation, and multiple PA stenoses.

Patients with AS are usually asymptomatic throughout childhood, even when stenosis is severe. Only 5% of children develop congestive heart failure in the neonatal period due to critical AS. Some children, as they approach adolescence, may become symptomatic for exercise intolerance; episodes of chest pain, due to myocardial ischemia; and syncope on exertion. Both chest pain and syncope are serious symptoms which may precede sudden death.

Aortic valvular stenosis is a progressive disease; two processes probably account for this: the development of myocardial fibrosis and the decrease in size of the stenotic valvar orifice by fibrosis and, later on, calcification of the valve. Bicuspid aortic valves often go undetected in early life; however, up to 70% of the cases develop some degree of stenosis or regurgitation by age 30.

Diagnosis. Infants with critical AS usually present in poor general condition, poor pulses, S3 and/or S4, and hyperdynamic precordium with a right ventricular tap. They may have either no murmur or a soft systolic ejection-type murmur due to the severe cardiac function impairment; differential oxygen saturations between the right arm (higher) and lower limbs (lower) are often detected.

In children and adolescents with severe valvular AS, a harsh grade 3–4/6, ejection systolic murmur is best heard at the second right or left intercostal space with good transmission to the neck and apex. In case of stenotic and regurgitant bicuspid valve, a high-pitched, early diastolic decrescendo murmur may be audible as well. Correlation of the severity of AS and the ECG abnormalities is relatively poor; LVH with or without strain pattern may be present in severe cases. Echocardiography is the preferred method for diagnosis and decision-making; it allows one to visualize directly the valve, to quantify the degree of stenosis, to assess cardiac function, and to detect associated CHD. In infant with severe AS, endocardial fibroelastosis can be visualized by the presence of a bright white layer in the endocardial tissue. Cardiac catheterization is rarely performed solely for diagnostic purposes; low cardiac output state can underestimate the degree of AS; therefore the aortic valve area can be calculated using the Gorlin and Hakki equations which include cardiac output and pressure gradient.

Treatment. For critical, neonatal valvular AS, inotropic agents and diuretics should be started to treat CHF; prostaglandin E_1 infusion should be given to reopen the arterial duct. Relief of the AS, either by percutaneous balloon valvuloplasty (PBV) or surgical valvotomy, should be achieved as soon as possible. Percutaneous valvuloplasty has become the preferred treatment at many centers, also in children and young adults [49]. Children who have undergone aortic valvotomy may require valve replacement later on if the valve becomes calcified or rigid or, sooner, if important regurgitation occurs; 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. Another option for aortic valve replacement, even in neonates and especially in small children with dysplastic and obstructed aortic valves, is the Ross operation [50] in which the patient's native pulmonary valve is excised and placed in aortic position and a homograft valve is placed in pulmonary position. The major advantages are related to the neoaortic valve which, being the patient's own tissue, degenerates very slowly, continues to grow with the patient, and does not require anticoagulation. Major disadvantages are high operative rates of acute and chronic morbidity and mortality when performed in neonates and infant, RV-PA conduit degeneration and dysfunction, and the potential dilation of the neoaorta with subsequent development of aortic regurgitation.

In subvalvar AS, surgical obstruction removal is indicated to relieve the LV pressure overload and to reduce the mechanical trauma to the aortic valve. The operative risk approaches that of operation for valvular AS; the major hazard is the possibility of damage to the mitral valve anterior leaflet since the membrane is quite often attached to that.

In supravalvular AS, surgery may be indicated for a lesser pressure gradient compared with valvular AS due to the potential concomitant disease of coronary arteries. Surgical obstruction relief is accomplished by different techniques: patch enlargement of the narrowing or slide aortoplasty techniques, according to the anatomy and the degree of ascending aorta hypoplasia.

Outcome. Mortality rate for infants and small children with valvular AS ranges from 15 to 20%. Sick neonates with poor preoperative general conditions have a mortality rate as high as 40%. The hospital mortality in older children is 1–2%.

Overall, PBV achieves a 60% reduction in the aortic valve gradient with a procedure-related mortality of 1.9% and complication rate strongly correlated with the age of the child. Repeat PBV and valve replacement occurred in 15% of patients after initial PBV [51], whereas 25% of patients require valve replacement 15–20 years after the original surgery.

Subaortic membrane has a high recurrence rate after surgical removal mainly in patients operated sooner and with high peak pressure gradients, suggesting a more severe form of disease [52].

Over the long term after supravalvular stenosis surgical relief, reobstruction and aortic valve progressive regurgitation can occur [53].

Coarctation of the Aorta

Coarctation of the aorta (CoA) is considered part of a generalized arteriopathy and not only a circumscript narrowing of the aorta. It occurs as a discrete stenosis most commonly seen in periductal region or as a long, hypoplastic segment (Fig. 1.11). It accounts for 5–8% of all CHD and may occur in isolation or in association with other anomalies: bicuspid aortic valve (up to 80%); subvalvular, valvular, or supravalvular AS; MV stenosis (as seen in Shone complex); hypoplastic left heart syndrome; and aberrant subclavian artery. CoA can be associated with Turner, Williams-Beuren [54], and congenital rubella syndromes or neurofibromatosis.

CoA causes significant increase in LV afterload resulting in increased wall stress, compensatory hypertrophy, and LV dysfunction; from the aorta a variable degree of arterial collateral circulation can develop. Elastic fiber rupture and fibrosis are typical of cystic medial necrosis which is found in the ascending and descending aorta determining an increased arterial stiffness [55]. Clinical signs and symptoms depend on the severity of CoA; when severe, closure of the arterial duct after birth will result in critical aortic obstruction, LV dilatation and dysfunction, low cardiac output state, and, if untreated, death. In less severe forms, CoA may be picked up during childhood when a murmur may be heard and femoral pulses found to be weak; in adulthood the most common sign is systemic hypertension [56]. Symptoms may include nosebleeds, dizziness, headache, shortness of breath, abdominal angina, leg cramps, claudication, exertional leg fatigue, and cold feet. Some life-threatening

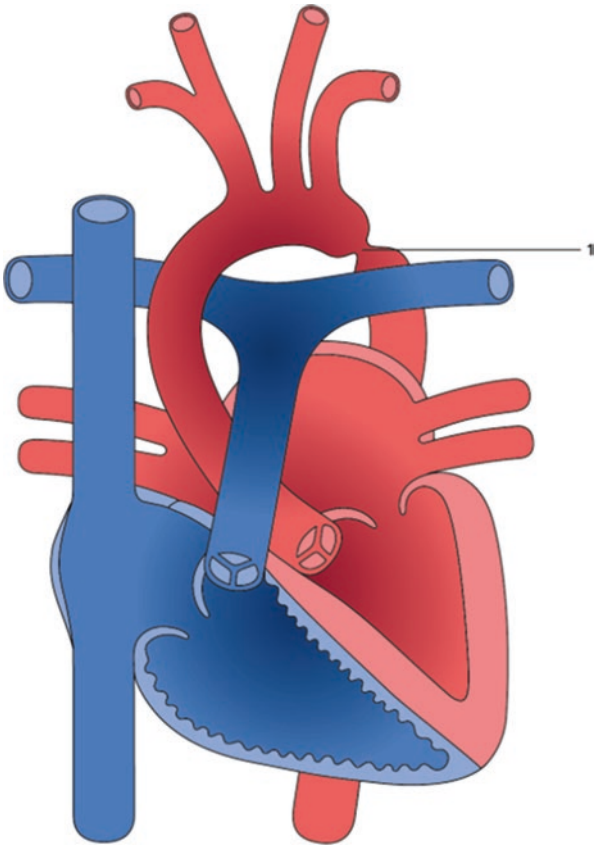

Fig. 1.11 Aortic isthmic coarctation (1)

conditions may occur during "natural history" of CoA: left heart failure, intracranial hemorrhage (from berry aneurysm of circle of Willis), aortic rupture/dissection, premature coronary and cerebral artery disease, and infective endocarditis.

Diagnosis. Neonates with severe CoA are pale and experience varying degree of respiratory distress and low cardiac output state; differential cyanosis may be present due to a right-to-left ductal shunt causing cyanosis only in the lower half of the body. Peripheral pulses may be weak and thready; the S2 is single and loud; a gallop is usually present, whereas a heart murmur is not audible in 50% of sick babies as long as the cardiac function improves. On ECG, RVH or right bundle branch block (RBBB) is often seen rather than LVH. On x-ray, marked cardiomegaly and pulmonary venous congestion are usually present.

In adolescent and adult patients, a significant CoA is diagnosed when there is a systolic blood pressure gradient between the upper and lower limb of at least 20 mmHg or less than 20 mmHg in the presence of systemic hypertension; common physical findings are weak or absent lower limb pulses. A continuous murmur in the left back near the scapula signals the presence of large collateral arteries; a systolic murmur sometimes can be heard in the left infraclavicular area or in the left upper

back. ECG generally shows LVH in patients with chronic hypertension. In some cases, chest-x-ray shows some typical findings which strongly suggest CoA: prominent ascending aorta contour, the change in aortic caliber at the coarctation site producing the "3" sign, and notching of the inferior margin of ribs 3–8 owing to erosion by large and developed collateral arteries.

Echocardiography is the definitive imaging modalities for diagnosing CoA and for identifying all associated cardiac defects; a sagittal view from the suprasternal notch should be used to visualize the posterior wedge-shaped shelf that characterizes true CoA. In neonates and infants, it is sometimes difficult to establish a proper diagnosis in the presence of PDA, and CoA may become evident once the duct closes. A typical Doppler flow pattern with persistent forward flow into diastole usually confirms a significant stenosis.

Key features, prior to any treatment, are the aortic arch morphology and the branching pattern of the head and neck vessels. If echocardiography is inconclusive, MR and CT are the preferred noninvasive techniques to evaluate the entire aorta, while cardiac catheterization is still the gold standard in case of complex CoA associated with other CHD: a peak-to-peak gradient >20 mmHg is indicative of significant CoA in the absence of well-developed collaterals.

Treatment. For sick neonates, PGE_1 infusion should be given to reopen the arterial duct; inotropic agents, diuretics, and, if necessary, mechanical ventilation should be started to treat CHF. All these measures aim at stabilizing the patient in preparation for surgery. In fact, the standard management of native CoA in neonates, infants, and young children is surgical repair [57]. Some different surgical techniques have been used: subclavian flap repair, patch aortoplasty, and, currently, resection with end to end anastomosis, via a left lateral thoracotomy or, in the presence of severely hypoplastic aortic arch, via a median sternotomy on cardiopulmonary bypass. For older children, adolescents, and adults with native CoA, in many specialized centers, balloon angioplasty (BA) with covered or non-covered stent implantation has become first choice treatment if anatomy is appropriate [57, 58] (Figs. 1.12 and 1.13). Procedure is generally performed under general anesthesia which reduces sympathetic drive and may result in a falsely low gradient across the CoA site.

Outcome. Surgical short-term complications include recurrent laryngeal or phrenic nerve damage, Horner syndrome, pleural effusion, and chylothorax. Short-term outcomes relate mostly to the clinical condition of the patient prior to surgery and the severity of associated cardiac lesions. For children and adolescents operated on, spinal cord injury with paraplegia is quite rare but is more common in patients with poor collateral circulation. Initially, long-term incidence of recurrent CoA after surgery approached 30%, whereas, nowadays, percentage has gone down to about 10% [59]. BA is mainly indicated in recoarctation in infants and young children providing excellent acute relief of obstruction but being associated with high rate of coarctation recurrence and big concerns regarding aneurysm formation and dissection. The use of covered stents has reduced significantly the incidence of dissection and aneurysm formation at the site of treated coarctation; furthermore, stents can be re-dilated later in life in case of recoarctation or residual stenosis.

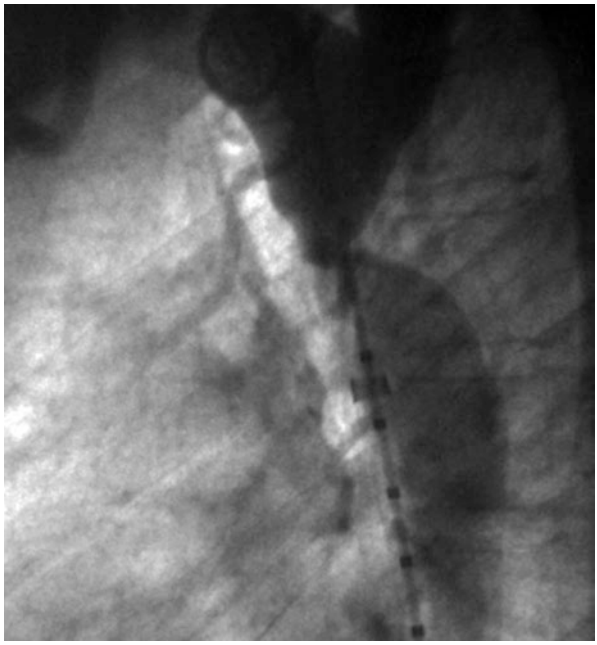

Fig. 1.12 Angiography: severe isthmic aortic coarctation

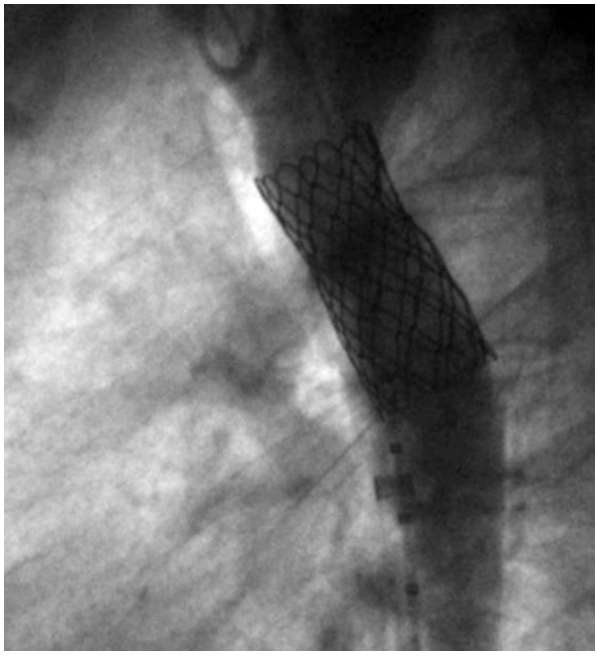

Fig. 1.13 Angiography post-covered stent implantation