

## **Congenital Heart Disease**

### **PREVALENCE**

Congenital heart disease occurs in 0.5–0.8% of live births. The incidence is higher in stillborns (3–4%), spontaneous abortuses (10–25%), and premature infants (about 2% excluding patent ductus arteriosus [PDA]). The diagnosis is established by 1 wk of age in 40–50% of patients with congenital heart disease and by 1 mo of age in 50–60% of patients.

### **ETIOLOGY**

The cause of most congenital heart defects is unknown. Most cases of congenital heart disease were thought to be multifactorial and result from a combination of genetic predisposition and environmental stimulus. Most congenital heart disease is a multifactorial inheritance pattern. The incidence of congenital heart disease in the normal population is  $\approx 0.8\%$ , and this incidence increases to 2–6% for a 2nd pregnancy after the birth of a child with congenital heart disease or if a parent is affected.

### **Acyanotic Congenital Heart Disease: The Left-to-Right Shunt Lesions Atrial Septal Defect**

Atrial septal defects (ASDs) can occur in any portion of the atrial septum (secundum, primum, or sinus venosus). Less commonly, the atrial septum may be nearly absent, with the creation of a functional single atrium. Isolated secundum ASDs account for  $\approx 7\%$  of congenital heart defects. The majority of cases of ASD are sporadic; autosomal dominant inheritance does occur as part of the Holt-Oram syndrome.

### **Ostium Secundum Defect**

An ostium secundum defect in the region of the fossa ovalis is the most common form of ASD and is associated with structurally normal atrioventricular (AV) valves. Mitral valve prolapse has been described in association with this defect but is rarely an important clinical consideration. Secundum ASDs may be single or multiple (fenestrated atrial septum), and openings  $\geq 2$  cm in diameter are common in symptomatic older children. Females outnumber males 3 : 1 in incidence. Partial anomalous pulmonary

venous return, most commonly of the right upper pulmonary vein, may be an associated lesion.

## **PATHOPHYSIOLOGY**

The degree of left-to-right shunting is dependent on the size of the defect, the relative compliance of the right and left ventricles, and the relative vascular resistance in the pulmonary and systemic circulations. With large defects, the ratio of pulmonary to systemic blood flow ( $Q_p : Q_s$ ) is usually between 2 : 1 and 4 : 1. The paucity of symptoms in infants with ASDs is related to the structure of the right ventricle in early life when its muscular wall is thick and less compliant, thus limiting the left-to-right shunts. The large blood flow through the right side of the heart results in enlargement of the right atrium and ventricle and dilatation of the pulmonary artery.

## **CLINICAL MANIFESTATIONS**

A child with an ostium secundum ASD is most often asymptomatic; the lesion may be discovered inadvertently during physical examination. Even an extremely large secundum ASD rarely produces clinically evident heart failure in childhood. Often, the degree of limitation may go unnoticed by the family until after surgical repair, when the child's growth or activity level increases markedly.

Examination of the chest may reveal a mild left precordial bulge. A right ventricular systolic lift is generally palpable at the left sternal border. A loud 1st heart sound and sometimes a pulmonic ejection click can be heard. In most patients, the 2nd heart sound is characteristically widely split and fixed in its splitting in all phases of respiration. A short, rumbling mid-diastolic murmur produced by the increased volume of blood flow across the tricuspid valve is often audible at the lower left sternal border. This finding, which may be subtle and is heard best with the bell of the stethoscope, usually indicates a  $Q_p : Q_s$  ratio of at least 2 : 1.

## **DIAGNOSIS**

The chest roentgenogram shows varying degrees of enlargement of the right ventricle and atrium, depending on the size of the shunt. The pulmonary

artery is large, and pulmonary vascularity is increased. These signs vary and may not be conspicuous in mild cases. Cardiac enlargement is often best appreciated on the lateral view because the right ventricle protrudes anteriorly as its volume increases. The electrocardiogram shows volume overload of the right ventricle; the QRS axis may be normal or exhibit right axis deviation, and a minor right ventricular conduction delay (rsR pattern in the right precordial leads) may be present.

The echocardiogram shows findings characteristic of right ventricular volume overload movement. The location and size of the atrial defect are readily appreciated by two-dimensional scanning. The shunt is confirmed by pulsed and color flow Doppler. If pulmonary vascular disease is suspected, cardiac catheterization confirms the presence of the defect and allows measurement of the shunt ratio and pulmonary pressure.

## **TREATMENT**

Surgical or transcatheter device closure is advised for all symptomatic patients and also for asymptomatic patients with a Qp : Qs ratio of at least 2 : 1. The timing for elective closure is usually after the 1st yr and before entry into school. Closure carried out at open heart surgery is associated with a mortality rate of <1

## **PROGNOSIS**

ASDs detected in term infants may close spontaneously. Secundum ASDs are well tolerated during childhood, and symptoms do not usually appear until the 3rd decade or later. Infective endocarditis is extremely rare, and antibiotic prophylaxis for isolated secundum ASDs is not recommended.

### **Atrioventricular Septal Defects (Ostium Primum and Atrioventricular Canal or Endocardial Cushion Defects)**

An ostium primum defect is situated in the lower portion of the atrial septum and overlies the mitral and tricuspid valves. In most instances, a cleft in the anterior leaflet of the mitral valve is also noted. The tricuspid valve is usually functionally normal, although some anatomic abnormality of the septal leaflet is generally present. The ventricular septum is intact.

An AV septal defect, also known as an AV canal defect or an endocardial cushion defect, consists of contiguous atrial and ventricular septal defects

with markedly abnormal AV valves. The lesion is common in children with Down syndrome and may occasionally occur with pulmonary stenosis.

### **Ventricular Septal Defect**

VSD is the most common cardiac malformation and accounts for 25% of congenital heart disease. Defects may occur in any portion of the ventricular septum, but most are of the membranous type. VSDs in the midportion or apical region of the ventricular septum are muscular in type and may be single or multiple (Swiss cheese septum).

### **Clinical Feature**

The clinical findings of patients with a VSD vary according to the size of the defect and pulmonary blood flow and pressure. Small VSDs with trivial left-to-right shunts and normal pulmonary arterial pressure are the most common. These patients are asymptomatic, and the cardiac lesion is usually found during routine physical examination. Characteristically, a loud, harsh, or blowing holosystolic murmur is present and heard best over the lower left sternal border, and it is frequently accompanied by a thrill.. Large VSDs with excessive pulmonary blood flow and pulmonary hypertension are responsible for dyspnea, feeding difficulties, poor growth, profuse perspiration, recurrent pulmonary infections, and cardiac failure in early infancy. Cyanosis is usually absent, but duskeness is sometimes noted during infections or crying. Prominence of the left precordium is common, as are a palpable parasternal lift, a laterally displaced apical impulse and apical thrust, and a systolic thrill. The holosystolic murmur of a large VSD is generally less harsh than that of a small VSD and more blowing in nature because of the absence of a significant pressure gradient across the defect. It is even less likely to be prominent in the newborn period. The pulmonic component of

### **PATHOPHYSIOLOGY**

The physical size of the VSD is a major, but not the only determinant of the size of the left-to-right shunt. When a small communication is present (usually <0.5 cm), the VSD is called restrictive and right ventricular pressure is normal. In large nonrestrictive VSDs (usually >1.0 cm), right and left ventricular pressure is equalized.

When the ratio of pulmonary to systemic resistance approaches 1:1, the shunt becomes bidirectional, the signs of heart failure abate, and the patient becomes cyanotic (Eisenmenger physiology).

## **TREATMENT**

The natural course of a VSD depends to a large degree on the size of the defect. A significant number (30–50%) of small defects close spontaneously, most frequently during the 1st 2 yr of life. Small muscular VSDs are more likely to close (up to 80%) than membranous VSDs are (up to 35%). The vast majority of defects that close do so before the age of 4 yr, although spontaneous closure has been reported in adults.

It is less common for moderate or large VSDs to close spontaneously, although even defects large enough to result in heart failure may become smaller and up to 8% may close completely. More commonly, infants with large defects have repeated episodes of respiratory infection and heart failure despite optimal medical management. Heart failure may be manifested in many of these infants primarily as failure to thrive. Pulmonary hypertension occurs as a result of high pulmonary blood flow. These patients are at risk for pulmonary vascular disease with time if the defect is not repaired.

## **Patent Ductus Arteriosus**

During fetal life, most of the pulmonary arterial blood is shunted through the ductus arteriosus into the aorta. Functional closure of the ductus normally occurs soon after birth, but if the ductus remains patent when pulmonary vascular resistance falls, aortic blood is shunted into the pulmonary artery. The aortic end of the ductus is just distal to the origin of the left subclavian artery, and the ductus enters the pulmonary artery at its bifurcation. Female patients with PDA outnumber males 2 : 1. PDA is also associated with maternal rubella infection during early pregnancy. It is a common problem in premature infants, where it can cause severe hemodynamic derangements and several major sequelae.

When a term infant is found to have a PDA, the wall of the ductus is deficient in both the mucoid endothelial layer and the muscular media. In a premature infant, the PDA usually has a normal structure; patency is the result of hypoxia and immaturity. Thus, a PDA persisting beyond the 1st few weeks of life in a term infant rarely closes spontaneously or with pharmacologic intervention, whereas if early pharmacologic or surgical intervention is not required in a premature infant, spontaneous closure occurs in most instances. A PDA is seen in 10% of patients with other congenital heart lesions and often plays a critical role in providing pulmonary blood flow when the right ventricular outflow tract is stenotic or atretic or in providing systemic blood flow in the presence of aortic coarctation or interruption.

### **PATHOPHYSIOLOGY**

As a result of the higher aortic pressure, blood shunts left to right through the ductus, from the aorta to the pulmonary artery. The extent of the shunt depends on the size of the ductus and on the ratio of pulmonary to systemic vascular resistance. In extreme cases, 70% of the left ventricular output may be shunted through the ductus to the pulmonary circulation. If the PDA is small, pressure within the pulmonary artery, the right ventricle, and the right atrium is normal. If the PDA is large pulmonary artery pressure may be elevated to systemic levels during both systole and diastole. Patients with a large PDA are at extremely high risk for the development of pulmonary vascular disease if left unoperated.

### **CLINICAL MANIFESTATIONS**

A small patent ductus does not usually have any symptoms associated with it. A large PDA will result in heart failure similar to that encountered in infants with a large VSD. Retardation of physical growth may be a major manifestation in infants with large shunts.

### **DIAGNOSIS**

If the left-to-right shunt is small, the electrocardiogram is normal; if the ductus is large, left ventricular or biventricular hypertrophy is present..

Cardiac size depends on the degree of left-to-right shunting; it may be normal or moderately to markedly enlarged. The chambers involved are the left atrium and ventricle. The aortic knob is normal or prominent.

The echocardiographic view of the cardiac chambers is normal if the ductus is small. Color and pulsed Doppler examinations demonstrate systolic or diastolic (or both) retrograde turbulent flow in the pulmonary artery and aortic retrograde flow in diastole.

## **PROGNOSIS AND COMPLICATIONS**

Patients with a small PDA may live a normal span with few or no cardiac symptoms, but late manifestations may occur. Spontaneous closure of the ductus after infancy is extremely rare. Cardiac failure most often occurs in early infancy in the presence of a large ductus but may occur late in life even with a moderate-sized communication. The chronic left ventricular volume load is less well tolerated with aging.

Infective endarteritis may be seen at any age. Pulmonary or systemic emboli may occur. Pulmonary hypertension (Eisenmenger syndrome) usually develops in patients with a large PDA who do not undergo surgical treatment.

## **TREATMENT**

In patients with a small PDA, the rationale for closure is prevention of bacterial endarteritis or other late complications. Once the diagnosis of a moderate to large PDA is made, treatment should not be unduly postponed after adequate medical therapy for cardiac failure has been instituted.