Chapter 21
Commonly Encountered Congenital Heart Disease

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Abbreviations

ASD	Atrial Septal Defect
CHD	Congenital Heart Disease
ECG	Electrocardiogram
IVC	Inferior Vena Cava
PFO	Patent Foramen Ovale
PVR	Pulmonary Vascular Resistance
Qp:Qs	Ratio of pulmonary to systemic flow
SVC	Superior Vena Cava
TEE	Transesophageal Echocardiogram
TOF	Tetralogy of Fallot
TTE	Transthoracic Echocardiogram
VSD	Ventricular Septal Defect

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Atrial Septal Defect (ASD)

Introduction

- 10% of CHD
- Typically results in a left-to-right shunt.
- 4 different sub-types (Fig. 21.1). We will discuss ostium secundum and sinus venosus ASDs (isolated ostium primum ASDs and coronary sinus defects are rare and beyond the scope of this text).

Ostium Secundum ASD

- Definition/Presentation:
  - Defects of the fossa ovalis—60% of ASDs
  - May be asymptomatic for years—often first present in adulthood (depending on size)

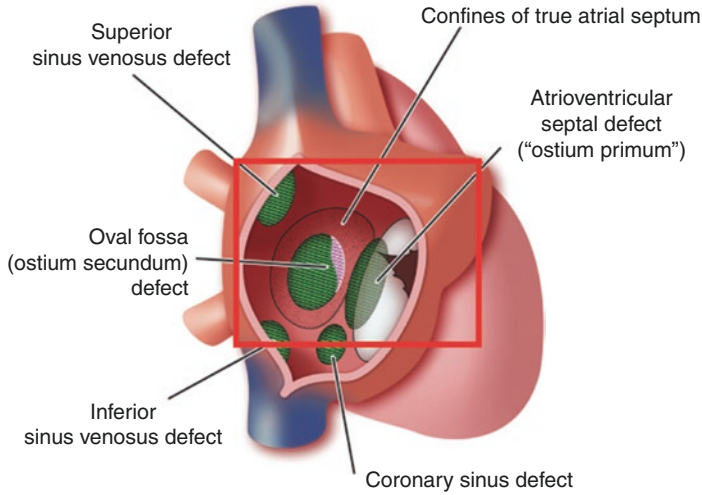

FIGURE 21.1 Schematic diagram outlines the different types of interatrial shunting that can be encountered (with permissions from Zipes et al. [2])

  - May be asymptomatic at diagnosis (diagnosed by physical exam, see below); if symptomatic, most common presentation is progressive exertional dyspnea and/or palpitations

Clinical Pearl
Consider ASD in your differential diagnosis of an adult patient with otherwise unexplained exertional dyspnea and/or palpitations.

  - Complications—atrial fibrillation/flutter, right heart failure, pulmonary hypertension, and paradoxical embolism
  - Any condition that increases left atrial pressure (i.e. left ventricular dysfunction or mitral valve disease) increases the left-to-right shunt and worsens symptoms.
- Physical Exam
  - Loud S1, a widely split and fixed S2, pulmonary systolic ejection murmur loudest at the left upper sternal border, and a tricuspid mid-diastolic murmur loudest at the left lower sternal border (due to increased flow across the tricuspid valve)
  - If right-sided heart failure/pulmonary hypertension is present, exam may show signs of elevated jugular venous pressure and right ventricular heave.
- Investigations
  - Electrocardiogram (ECG)—Look for sinus node dysfunction, prolonged PR interval, right axis deviation, rSR' pattern in V1, and large P waves.
  - Transthoracic echocardiogram (TTE)—Evaluate for color flow across the inter-atrial septum (as well as positive bubble study), and/or a dilated right ventricle. Perform a Doppler estimate of pulmonary artery pressure and an assessment for associated defects.

  - Transesophageal echocardiogram (TEE)—often best characterizes the ASD (precise size, margins, etc.) and identifies the pulmonary veins (to assess for associated partial anomalous pulmonary venous return).
  - Cardiac catheterization—performed to calculate pulmonary vascular resistance and determine the ratio of pulmonary to systemic flow (Qp:Qs); also often performed to evaluate for concomitant coronary artery disease prior to ASD repair.
- ASD Closure
  - Indications for ASD closure—evidence of right heart volume overload, left-to-right shunt ratio greater than or equal to 1.5:1, size greater than 10 mm, or prevention of recurrent paradoxical embolism
  - Contradictions to ASD closure—significant pulmonary vascular disease, severe left ventricular dysfunction, and significant mitral valve disease
  - Often can be closed percutaneously or surgically

Sinus Venosus ASD

- Caused by defects of the infolding of the atrial wall at the site of the superior vena cava (SVC) or inferior vena cava (IVC)—superior is more common than inferior
- Superior sinus venosus ASD—the defect in the atrial wall leads to the SVC communicating with both atria—invariably associated with anomalous right-sided pulmonary venous return whereby the right-sided pulmonary veins drain into the SVC near its junction with the right atrium.
- 2–3% of ASDs
- Similar presentation to ostium secundum ASDs
- TEE—performed both to evaluate the defect and to identify anomalous pulmonary venous drainage; MRI or CT may be required if the pulmonary venous return is not clearly seen on TEE

- Same indications for closure as ostium secundum ASDs; however, sinus venosus ASDs are not suitable for percutaneous closure because of the lack of rim around the defect as well as the associated anomalous pulmonary venous return.

Patent Foramen Ovale (PFO)

- PFO is common—found in up to 25% of the population—it is usually clinically silent
- Results from failure of fusion of the valve of the foramen ovale with the inter-atrial septum after birth when the left atrial pressure exceeds that of the right atrium; unlike ASDs—there is no defect in atrial septal tissue
- Most are found incidentally; however, paradoxical embolism can be a cause of cryptogenic stroke in young adults
- Detected on contrast echocardiogram—agitated saline is injected intravenously while an echocardiogram is obtained (can be TTE or TEE). A PFO is likely if, with Valsalva, bubbles appear within the left atrium within 5 heartbeats
- Patients with PFO who have had prior embolic stroke and have risk factors for venous thrombosis appear to benefit from PFO closure (which can be done percutaneously similar to ostium secundum ASD closure). However, a thorough evaluation of risk factors for alternative etiology of stroke should be performed prior to PFO closure.

Clinical Pearl
PFO's are commonly diagnosed on echocardiogram during the inpatient workup for stroke. However, one must complete a comprehensive evaluation to identify possible alternative etiologies for stroke before referral for PFO closure. If an alternative etiology is found, the patient is unlikely to benefit from PFO closure.

Ventricular Septal Defect (VSD)

- *Definition/Prevalence*
  - Interventricular septum is composed of 4 parts: inlet septum, outlet septum, muscular (also known as trabecular) septum, and membranous septum. VSDs can arise from defects in any of these components (Fig. 21.2).
  - Most common congenital heart defect—membranous VSDs are the most common type

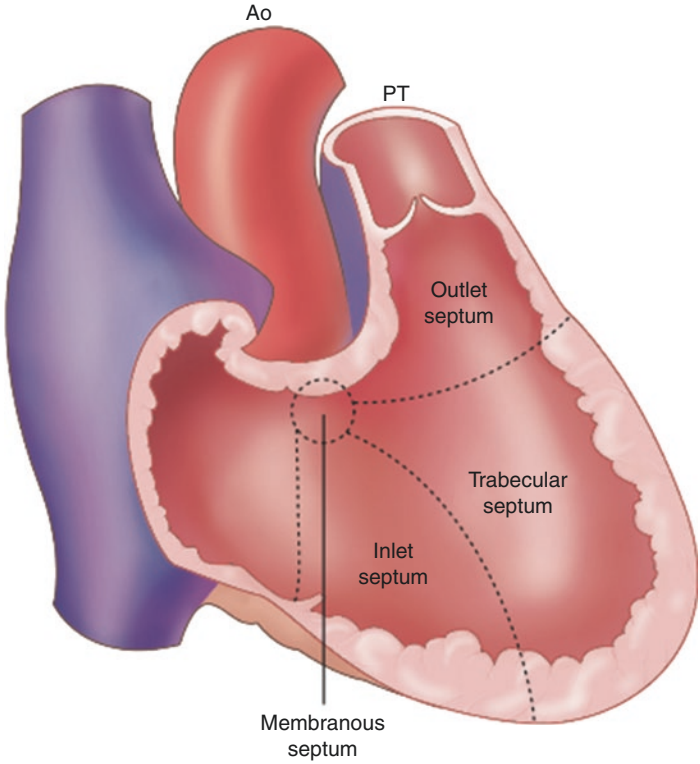

FIGURE 21.2 Four components of the ventricular septum are shown here from the right ventricular aspect. *Ao*, aorta; *PT*, pulmonary trunk (With permissions from Zipes et al. [2])

  - Can occur in isolation, in association with other lesions, or as part of a more complex condition (such as tetralogy of Fallot)
- *Presentation*
  - Depends upon the size and hemodynamic effects of the defect
  - Restrictive VSDs describe small defects with a high pressure gradient between the left and right ventricles and a small shunt of no hemodynamic significance.
  - Larger defects are hemodynamically significant and cause left ventricular volume overload and progressive increase in pulmonary vascular resistance (PVR).
  - In adults, un-operated restrictive VSDs are often asymptomatic but cause a loud murmur. Survivors of large un-operated VSDs are likely to have developed significant pulmonary hypertension and possibly Eisenmenger syndrome.
- *Physical Exam*
  - Small restrictive VSDs cause a high-frequency pansystolic murmur loudest at the left sternal border, with palpable thrill.
  - Moderate-to-large nonrestrictive VSDs cause a displaced cardiac apex and a pansystolic murmur, as well as an apical diastolic murmur and S3 (from increased flow through the mitral valve).

Clinical Pearl
The intensity of the systolic murmur due to a VSD is inversely proportional to the size of the VSD because of the increased turbulence and flow velocity produced by a smaller defect.

- *Investigations*
  - ECG—large VSDs cause right axis deviation and biventricular hypertrophy; ECG often unremarkable in small defects

  - TTE—characterizes the size, location, and hemodynamic consequences of the VSD and identifies associated lesions; moderate-sized VSDs cause left atrial and left ventricular volume overload, which leads to dilation of these chambers. Large VSDs that have already resulted in increased PVR cause right ventricular pressure overload, which leads to right ventricular hypertrophy.
  - Cardiac catheterization—performed to calculate the size of the shunt as well as PVR.
- *VSD Closure*
  - Indications for closure—symptoms and Qp:Qs greater than 1.5:1, ventricular dysfunction with right ventricular pressure or left ventricular volume overload, or a previous episode of endocarditis.
  - Selected membranous and muscular VSDs are suitable for percutaneous closure; however, surgical repair is usually necessary when indicated.
  - Damage to the conduction system is relatively common during VSD closure (either percutaneous or surgical), especially for membranous VSDs. Right bundle branch block is common post-procedure and complete heart block can occur, and if it is persistent, a pacemaker is required.

Tetralogy of Fallot (TOF)

Introduction

- The underlying abnormality in TOF is the antero-cephalad deviation of the outlet ventricular septum. This leads to the four features (Fig. 21.3):
  - VSD
  - Sub-pulmonary stenosis
  - Aorta that overrides the inter-ventricular septum
  - Secondary right ventricular hypertrophy
- TOF—most common cyanotic congenital heart defect

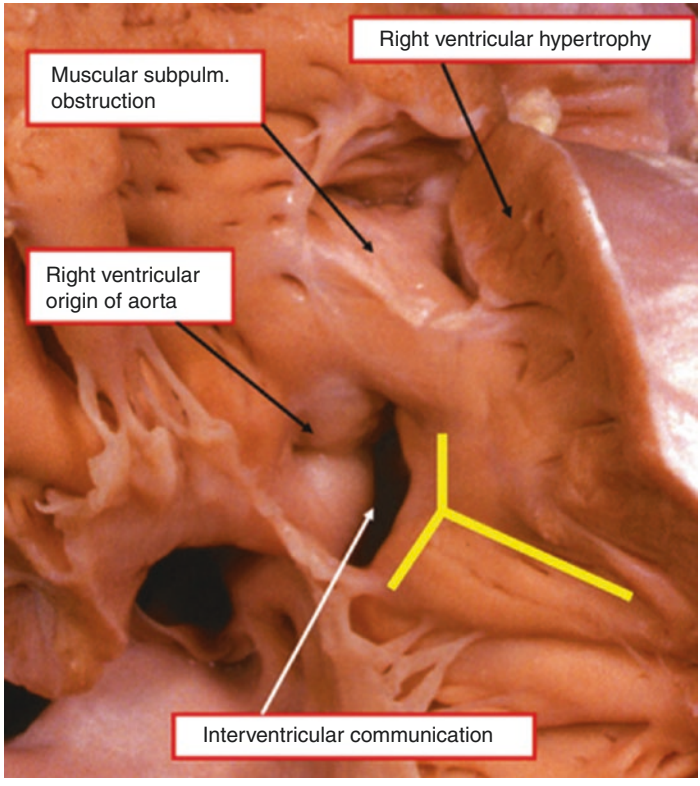

FIGURE 21.3 The photograph, taken from the apex of the right ventricle looking towards the base, shows the features of tetralogy of Fallot. The infundibulum is inserted to the antero-cephalad limb of the septomarginal trabeculation (*yellow* Y). (With permissions from Anderson [3])

Natural History (Un-Operated)

- Survival past 40 years of age is rare.
- Survival into adulthood without repair is dependent upon mild right ventricular outflow tract obstruction in childhood. Patients with severe obstruction have cyanosis early in life and don't survive to adulthood without repair.

- Complications
  - Cyanosis
  - Arrhythmia (atrial or ventricular)
  - Ascending aortic dilation and aortic regurgitation
  - Endocarditis
- Physical exam
  - Cyanosis and clubbing
  - Right ventricular heave
  - Palpable thrill and loud systolic ejection murmur (from the right ventricular outflow tract obstruction)
  - Soft P2
- Investigations
  - ECG—right axis deviation and right ventricular hypertrophy
  - Chest x-ray—look for boot-shaped heart and decreased pulmonary vascularity.
  - Echocardiography makes the diagnosis.

TOF Repair

- Surgical repair involves:
  - Patch closure of the VSD
  - Resection of the infundibular stenosis
  - Transannular patch to enlarge the pulmonary valve annulus in most patients
- Most adults with TOF have undergone repair and have a good prognosis. However, lifetime follow-up is required due increased risk of complications, particularly relating to the pulmonary valve and the need for pulmonary valve replacement.
- Possible sequelae of repair
  - Right bundle branch block
    Occurs in almost all patients because the right bundle runs in the floor of the VSD and is damaged during surgery
  - Pulmonary regurgitation (more on this below)
  - Late complete heart block

  - Residual right ventricular outflow tract obstruction
  - Aortic dilation and aortic regurgitation
  - Endocarditis
  - Arrhythmia and arrhythmogenic sudden death

Pulmonary Regurgitation After TOF Repair

- Most patients with TOF repair have severe pulmonary regurgitation that results from the relief of pulmonary stenosis and RVOT obstruction at the time of repair (transannular patch). Severe pulmonary regurgitation is usually well tolerated for several years or decades but most will eventually require pulmonary valve replacement. Pulmonary valve replacement is not a usual part of initial TOF repair.
- Signs of severe pulmonary regurgitation
  - Loss of sinus rhythm
  - Signs of right heart failure (elevated jugular venous pressure, hepatomegaly, peripheral edema, etc.)
  - Right ventricular heave
  - Soft or absent P2 (single S2)
  - To and fro systolic and diastolic murmur of pulmonary regurgitation (the diastolic murmur is often missed when it is short with rapid decrescendo)
- Investigations
  - With few exceptions, ECG shows RBBB with QRS widening. Varying degrees of atrioventricular block can also be present. Longer QRS duration is associated with more severe RV dilation and arrhythmia risk.
  - In addition to demonstrating the severe pulmonary regurgitation, echocardiography is important to evaluate for other abnormalities, including ventricular dysfunction, paradoxical inter-ventricular septal motion, residual VSD, aortic root dilation, aortic regurgitation, and residual right ventricular outflow tract obstruction.

- Indications for pulmonary valve replacement
  - Severe pulmonary regurgitation and any of the following:
    Increasing symptoms
    Impaired exercise tolerance on cardio-pulmonary exercise testing
    Arrhythmias
    Progressive right ventricular dilation or dysfunction

Clinical Pearl
Severe pulmonary regurgitation after TOF repair can be asymptomatic for years. Indications for referral for pulmonary valve replacement include worsening symptoms, objective evidence of exercise intolerance, or right ventricular dilation or dysfunction.

Cyanotic Heart Disease—The Basics

Introduction

- There are many specific defects that cause cyanosis other than TOF. Detailed discussion of each specific condition is beyond the scope of this text. Nevertheless, there are some general principles that apply to the care of the hospitalized cyanotic patient with which adult providers should be familiar. These patients are typically followed at tertiary referral centers; however, they often live far away from these centers and when acutely ill may present and be admitted locally initially for stabilization. This underlies the need for the adult inpatient provider to have a basic understanding of cyanotic heart disease.

- Cyanosis is present when there is a right-to-left shunt. The shunt can occur at any level—intra-cardiac, between the great vessels, or intrapulmonary.
- Cyanosis is typically clinically detectable when oxygen saturations are less than 85%.

General Principles for Inpatients with Cyanotic Heart Disease

- Supplemental oxygen should be used primarily for symptom relief and/or to maintain baseline oxygen saturations; oxygen is a pulmonary vasodilator and depending on the patient's anatomy, excessive oxygen may cause pulmonary edema.
- Use 0.22 micron air filters on all intravenous lines and use infusion pumps with a bubble detector to prevent paradoxical bubble emboli
- Maintain adequate hemoglobin concentration to optimize oxygen-carrying capacity
- Only perform phlebotomy if there are signs and symptoms of hyperviscosity (headache, confusion, etc.) due to very high hemoglobin concentration; phlebotomy can cause a relative iron deficiency anemia, which can increase thromboembolic risk
- Vasodilators can worsen hypoxia and should only be used with caution and under the supervision of an adult congenital cardiologist.

Clinical Pearl
One must be aware of the common pitfalls in caring for cyanotic patients—avoid the following: 1) excessive supplemental oxygen, 2) vasodilators, and 3) excessive phlebotomy.

Common Inpatient Emergencies in Cyanotic Heart Disease

- Tachyarrhythmias
  - Atrial arrhythmias are often atypical.
  - Tachyarrhythmias are generally not well tolerated.
  - Electrical cardioversion is often safer and more effective than anti-arrhythmic therapy.
- Hemoptysis
  - Can be life-threatening and warrants inpatient admission
  - One of the leading causes of death in cyanotic patients, especially in those with pulmonary hypertension.
  - Chest CT should be obtained.
  - Systemic blood pressure may need to be lowered.

    In patients with Eisenmenger syndrome, the systemic blood pressure is essentially equal to the pulmonary arterial pressure, and it is important to lower the pressure in a bleeding pulmonary vessel.

    IV beta blockers are typically the most appropriate pharmacologic agent; as noted above, vasodilators generally should be avoided.
  - Emergency management for severe hemoptysis is complicated and requires emergent adult congenital cardiology consultation. In general, bronchoscopy, intubation, and mechanical ventilation should be avoided if at all possible. Urgent transfer to a tertiary center is usually required.
  - Possible sources of hemoptysis—pulmonary artery thrombus, bleeding from a pulmonary arteriovenous malformation, bleeding collateral vessels, or chest infection

Key Learning Points

1. There are currently more adults than children living with CHD in the United States; therefore, adult inpatient providers need to be familiar with commonly encountered congenital heart defects.
2. ASDs comprise about 10% of CHD, and the diagnosis should be considered in patients with otherwise unexplained exertional dyspnea.
3. VSD is the most common congenital heart defect, and auscultation of the systolic murmur can lead to the diagnosis in asymptomatic patients.
4. Severe pulmonary regurgitation is common after TOF repair, and pulmonary valve replacement will eventually be needed in most patients, once indications arise, which include signs or symptoms of RV dilation and dysfunction.
5. For patients with cyanotic CHD, excessive oxygen supplementation, unnecessary phlebotomy, and vasodilators should be avoided, and 0.22 micron air filters should be used on all IVs. Arrhythmias should be treated aggressively, and hemoptysis can be life-threatening and warrants transfer to an adult congenital heart disease center.

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