Congenital Heart Disease

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KEYWORDS
- Pediatric
- Congenital heart disease
- Echocardiography
- Cardiology

Congenital heart disease is estimated to affect up to 1% of live births, although the spectrum of disease severity varies greatly.1 Severe lesions, such as hypoplastic left heart syndrome or truncus arteriosus, are relatively rare compared with atrial or ventricular septal defects and some lesions, such as a small patent ductus arteriosus, may resolve spontaneously.2 Despite advances in diagnosis and surgical care, congenital cardiac malformations remain one of the leading cause of death in infants.3 The incidence of specific congenital heart disease lesions is reflected in Table 1. Most congenital heart disease is diagnosed in utero by ultrasound during routine prenatal care. However, some lesions may escape detection and not manifest until after birth (Table 2), and these are the ones that are most likely to present to the emergency department. A significant proportion of lesions is dependent on mixing of oxygenated and deoxygenated blood and relies on a patent ductus arteriosus for communication between the systemic and pulmonic circulations. Thus, a child with congenital heart disease may present with severe cyanosis or shock at 1 to 2 weeks of age as the ductus arteriosus closes, making an understanding of these lesions crucial for any clinician who treats children in the neonatal period. Some nonobstructive lesions (ie, ventricular septal defects and patent ductus arteriosus) present later in the newborn period with congestive heart failure (CHF). The cardinal presentations of cyanosis, shock, and CHF can mimic those of pulmonary disease or sepsis; thus, the emergency physician must establish a broad differential and consider congenital heart disease when treating a critically ill neonate. Along with structural abnormalities, congenital rhythm disturbances, such as supraventricular tachycardia, congenital complete heart block, and long QTc syndrome, can also present during the newborn period with signs and symptoms that suggest other, more common conditions.

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ANATOMY AND PATHOPHYSIOLOGY

Circulation in the fetus is designed to transport oxygen-rich blood from the placenta to the systemic circulation. To accomplish this feat, the fetal lungs must be bypassed and blood shunted from the right side of the heart to the left. After leaving the placenta, oxygen-rich blood bypasses the liver through the ductus venosus and is returned to the right atrium. Most of this oxygenated blood then travels from the right atrium to the left atrium through the foramen ovale and is then pumped to the systemic circulation by the left ventricle. A small amount of blood enters the high-resistance pulmonary circuit and then is shunted right to left across the ductus arteriosus, from the pulmonary artery to the aortic arch.

At birth, the lungs fill with air, and the pulmonary vascular resistance drops dramatically. This leads to a complex series of changes that results in the normal circulatory pattern seen in adults. As the pulmonary vascular resistance falls, more blood enters...
the pulmonary circuit and oxygenated blood is delivered in larger quantities to the left atrium. This leads to a reversal of pressure forces and functional closure of the foramen ovale. During this time of transition, systemic vascular resistance rises and results in a reversal of shunting through the ductus. The blood flowing left to right across the ductus arteriosus is now highly oxygenated and this results in closure of this pathway within the first 2 weeks of life. The increased workload on the left ventricle results in an increased left ventricular mass while the right ventricle mass decreases. Normal fetal circulatory anatomy is depicted in **Fig. 1.** The development of the heart is a complex embryologic process involving an organized series of molecular and morphogenetic events. Any genetic, molecular, or cellular error in this process can result in gross structural abnormalities of the heart, heart valves, blood vessels, or conduction system with functional significance.\(^4\) There are many risk factors for the development of congenital heart disease. Family history plays a role; 1% to 4% of babies born to parents with congenital heart disease are affected.\(^5\) Maternal diabetes is associated with up to a 30% chance of structural heart disease in the newborn, particularly hypertrophic cardiomyopathy, ventricular septal defect, or transposition of the great vessels (TOGV).\(^6\) Maternal alcohol or drug use (ie, lithium use and associated Ebstein anomaly) also increases the risk of heart defects (**Table 3**).

**Fig. 1.** Normal fetal circulatory anatomy. Ao, aorta; LA, left atrium; LV, left ventricle; PA, pulmonary artery; RA, right atrium; RV, right ventricle. (**From** Mick N. Pediatric cardiac disorders. In: Adams JG, Barton ED, Collings J, et al, editors. Emergency medicine. 1st edition. Philadelphia: Saunders Elsevier; 2008. p. 618; with permission.)
Neonates presenting to the emergency department with undiagnosed congenital heart disease, whether it be structural or arrhythmogenic, typically exhibit signs of shock, cyanosis, CHF, or a combination of the three and typically present at 2 weeks of age as the ductus arteriosus closes. Those with left-sided obstructive lesions present with inadequate systemic perfusion and shock, whereas those with right-sided lesions present with cyanosis because of inadequate blood reaching the lungs. The differential diagnosis of shock in a neonate is broad with infectious etiologies being far and away the most common (Table 4). A blood pressure differential between the preductal right arm and the postductal left arm may suggest a ductal-dependent left-sided obstructive lesion, such as critical coarctation of the aorta or congenital aortic stenosis.

Cyanosis is caused by the presence of deoxygenated blood at the level of the capillaries. Detection of cyanosis requires a systemic oxygen saturation of 80% to 85%, depending on the patient’s hemoglobin level. Transient peripheral cyanosis (acrocyanosis) can occur in children with normal cardiac anatomy and function as a result of benign conditions, such as cold exposure, and more serious noncardiac conditions, such as sepsis. Central cyanosis, involving the mucous membranes, the lips, or the trunk, is always pathologic and should raise red flags. In children with cyanotic congenital heart disease, cyanosis can occur if blood flow to the lungs is insufficient or if a large percentage of the deoxygenated blood is pumped to the systemic circulation and a large percentage of the oxygenated blood is pumped back to the lungs.7

<table>
<thead>
<tr>
<th>Risk Factor</th>
<th>Lesions Associated</th>
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<tr>
<td>Maternal diabetes</td>
<td>Ventricular septal defect, transposition of the great vessels, hypertrophic cardiomyopathy</td>
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<td>Maternal lithium use</td>
<td>Ebstein anomaly</td>
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<td>Maternal phenytoin use</td>
<td>Aortic stenosis, pulmonic stenosis</td>
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<tr>
<td>Maternal alcohol use</td>
<td>Ventricular septal defect, atrial septal defect</td>
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<th>Type of Shock</th>
<th>Etiology</th>
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<tr>
<td>Hypovolemia</td>
<td>Dehydration, Blood loss</td>
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<tr>
<td>Cardiogenic</td>
<td>Coarctation of the aorta, Aortic stenosis, Interrupted aortic arch, Hypoplastic left heart syndrome, Congenital arrythmias, Myocarditis, Tension pneumothorax, Tamponade</td>
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<td>Distributive</td>
<td>Sepsis, Anaphylaxis</td>
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Pulmonary causes of cyanosis typically improve with supplemental oxygen administration or agitation, whereas cardiac cyanosis does not improve with oxygen and generally worsens with activity or crying. Neonates who present with cyanosis generally have ductal-dependent right-sided obstructive lesions and are intolerant of the transition to postnatal circulation.

CHF occurs with structural heart disease and congenital rhythm abnormalities. Typically, the infant presents with respiratory distress, tachypnea, and rales on pulmonary examination, although more subtle signs, such as poor feeding and sweating with feeds, may be the only clue to the diagnosis. Unlike adults with CHF, peripheral edema is rare in children. Physical examination findings suggestive of heart failure include signs of respiratory distress with grunting, flaring, and pulmonary rales. Hepatomegaly is also a common sign caused by engorgement of the hepatic vasculature, and examination of the liver edge should be a standard part of the examination in any child presenting in the first month of life with respiratory distress. Heart failure can be caused by volume overload of the right ventricle as is seen in total anomalous pulmonary venous return (TAPVR) with a large ventricular septal defect or lesions with large left-to-right shunts, such as critical aortic stenosis or coarctation of the aorta as blood preferentially flows into the low resistance pulmonary circuit. Congenital rhythm disturbances, particularly supraventricular tachycardia, can also lead to CHF because of poor cardiac output. For the emergency physician, initial management should be focused on the stabilization of these cardinal presentations.

STABILIZING DUCTAL-DEPENDENT LEFT-SIDED OBSTRUCTIVE LESIONS PRESENTING AS SHOCK

Ductal-dependent left-sided obstructive lesions include coarctation of the aorta, critical aortic stenosis, interrupted aortic arch, and hypoplastic left heart syndrome. These lesions are similar in that blood flow to the systemic circulation is contingent on the patency of the ductus arteriosus (Fig. 2). Many of these lesions are detected in utero by the 20-week anatomic ultrasound during the prenatal period. If undetected, they become apparent during the first 2 weeks of life when the ductus arteriosus closes and these infants typically present with profound shock and without cyanosis. The differential diagnosis of shock in the neonate is broad and includes cardiac, infectious, and metabolic causes. All neonates in shock should be treated aggressively with intubation if necessary; fluid resuscitation (20 mL/kg of normal saline); and empiric antibiotics and antivirals (ampicillin, gentamicin, and acyclovir). If the infant fails to

![Fig. 2.](A) Preductal closure of left-sided obstructive lesion showing systemic circulation dependent on patent ductus arteriosus and mixing occurring at the level of the ductus. (B) Closure of the ductus results in decrease in systemic perfusion and shock.)
respond to these interventions or a cardiac cause is suspected, prostaglandin E₁ should be given. Prostaglandin E₁ is given as an infusion starting at 0.1 μg/kg/min and works by preventing closure of the ductus arteriosus, which results in improved systemic blood flow in left-sided obstructive lesions. The infusion is titrated to palpable femoral pulses and positive results occur in minutes. The major side effect of the infusion is apnea, which can occur in up to 10% of patients and intubation should be considered if an infant is to be transferred to another facility. Once a child has been stabilized on prostaglandins, definitive management involves consultation with a pediatric cardiologist for echocardiography and transfer to a center capable of repairing the lesion in question.

STABILIZING DUCTAL-DEPENDENT RIGHT-SIDED OBSTRUCTIVE LESIONS PRESENTING AS CYANOSIS

Ductal-dependent right-sided obstructive lesions include tricuspid atresia, severe pulmonic stenosis, tetralogy of Fallot, TOGV, Ebstein anomaly, and many others. In right-sided obstruction, pulmonary blood flow is dependent on the combined volume of that coming from the ductus arteriosus and that coming through the obstructed right ventricular output. As the ductus closes, at 1 to 2 weeks of age, the infant becomes cyanotic (Fig. 3). The differential diagnosis of cyanosis includes cardiac and pulmonary conditions, and chest radiographs and the hyperoxia test may be useful in making the distinction. A chest radiograph is obtained looking for signs of pulmonary disease including pneumothorax, lung collapse, or pneumonia, and to evaluate the heart size and shape. The hyperoxia test takes advantage of the fact that supplemental oxygen does not raise the PaO₂ of the blood in congenital heart disease to the same degree as in pulmonary disease because of the presence of an intracardiac shunt. To perform the test, have the infant breath 100% oxygen for 10 minutes and then measure a postductal arterial blood gas. Congenital heart disease is suggested if the PaO₂ is less than 150 mm Hg during hyperoxia. Pulmonary disease is more likely if the PaO₂ is greater than 150 mm Hg during hyperoxia.

Stabilizing right-sided obstructive lesions involves the administration of a prostaglandin E₁ infusion to keep the ductus arteriosus open. The dosing of prostaglandins is identical to that used in left-sided obstruction and the infusion should be titrated to oxygen saturations of 80% to 85%. Once the oxygen saturations have been stabilized,

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*Fig. 3.* (A) Preductal closure of right-sided obstructive lesion showing pulmonary circulation dependent on patent ductus arteriosus and mixing occurring at the level of the ductus. (B) Closure of the ductus results in severe cyanosis because oxygenated blood from the lungs has no way to enter the systemic circuit.
pediatric cardiology consultation should be obtained and transfer to an appropriate tertiary care center effected.

STABILIZATION OF LESIONS PRESENTING WITH CHF

Treatment of CHF in the newborn suspected of having congenital heart disease may require intubation and positive pressure ventilation, and gentle diuresis. Because structural heart disease and congenital rhythm abnormalities can cause CHF, electrocardiography early in the resuscitation may aid in the diagnosis. Supraventricular tachycardia in infants is suggested by a narrow complex tachycardia with rates greater than 220 beats per minute.

LESION-SPECIFIC PRESENTATIONS

Left-sided Obstructive Lesions

Hypoplastic left heart syndrome

Hypoplastic left heart syndrome is a spectrum of cardiac anomalies including varying degrees of underdevelopment of the aorta, aortic valve, left ventricle, mitral valve, and left atrium. The right ventricle is the dominant pumping chamber of the heart and systemic blood flow relies on right-to-left shunting via the ductus arteriosus to the aorta.

Before closure of the ductus arteriosus, patients may have subtle signs including mild cyanosis, tachypnea, and tachycardia. When the ductus arteriosus begins to constrict, systemic cardiac output is significantly reduced and profound CHF, cardiovascular collapse, and shock rapidly develops.

Chest radiographs may reveal cardiomegaly with pulmonary edema. Electrocardiogram is generally nonspecific but may show evidence of right ventricular hypertrophy. Definitive diagnosis requires echocardiography, which can elucidate the character and severity of the anatomic anomalies. Initial stabilization involves prostaglandin E₁ infusion and endotracheal intubation is frequently required.¹⁰

Coarctation of the aorta

Coarctation of the aorta represents a spectrum of disease ranging from mild narrowing of the descending aorta to complete interruption of the aortic arch. It most often presents as a discrete narrowing in the region of the ligamentum arteriosum. The descending aorta just proximal to the coarcted section is frequently aneurismatic, and a bicuspid aortic valve is present in between 22% and 42% of cases.¹¹ After closure of the ductus arteriosus, infants with severe coarctation or an interrupted aortic arch present in shock with acidosis and evidence of end-organ damage.

The most significant clue to diagnosis is a pulse differential between the upper and lower extremities. A normal patient has an increase of 5 to 10 mm in systolic blood pressure in the lower extremities compared with the upper extremities. Absence of this differential or a decrease in the systolic blood pressure in the lower extremities compared with the upper extremities should prompt further evaluation for coarctation. Chest radiograph may demonstrate characteristic rib notching, indicative of significant arterial collateral formation bypassing the coarcted area. Echocardiography is again the diagnostic modality of choice and initial stabilization involves prostaglandin E₁ infusion, inotropic support with dopamine or dobutamine, and correction of acidosis with bicarbonate.

Aortic stenosis

Aortic stenosis is defined as narrowing across the aortic valve. Left untreated, this results in increased afterload on the left ventricle, left ventricular hypertrophy, and
eventual dilatation and failure of the left ventricle. In neonates with critical aortic valvular stenosis, shock caused by an obstructed aortic valve puts these infants at extreme risk for early sudden death.3

Echocardiography is used to confirm the diagnosis and prostaglandin E1 is a life-saving intervention in patients with critical aortic stenosis. Initial stabilization also includes airway support, inotropic augmentation with dopamine or dobutamine, and correction of acidosis.8

Right-sided Obstructive Lesions

Transposition of the great vessels

TOGV accounts for 5% of congenital heart disease in children.4 Half of the patients with this anomaly present within the first hour of life and 90% present within the first day.12 With this lesion, the aorta arises from the right ventricle and receives deoxygenated blood returning from the systematic circulation. This results in oxygen saturations in the main pulmonary artery that exceed those in the aorta. Infants with transposition of the great arteries may be at higher risk of cerebral damage than others because of chronically decreased oxygen delivery to the brain in fetal life as oxygenated blood is pumped preferentially to the lower body at the expense of the brain.13 With the two components of the circulation working in parallel instead of in series, a connection between the two, at the level of the atria, the ventricles, or the ductus, is required for survival.

Neonates with TOGV develop cyanosis soon after birth as the mixing between venous and arterial blood diminishes with the closure of the foramen ovale and the ductus arteriosus. If an atrial septal defect, ventricular septal defect, or patent ductus arteriosus is present, mixing is improved, and cyanosis may not present immediately. Respirations are typically not labored, the liver is not enlarged, and pulses are generally normal. There is usually not a murmur.

Chest radiographs in TOGV typically show a normal or minimally enlarged cardiac silhouette. The superior mediastinum is usually narrow and the thymus is involuted, which leads to an “egg-on-a-string” appearance (Fig. 4). Pulmonary vasculature is prominent, and the aortic arch is left-sided. A normal chest radiograph does not exclude the diagnosis of transposition, and the classic findings are seen less than 50% of the time.14

Echocardiography is the test of choice for diagnosing TOGV and can also be helpful in identifying associated lesions, such as pulmonic valvular stenosis, a patent foramen ovale, and ventricular and atrial septal defects. Significant aortic arch hypoplasia, interruption, or coarctation occurs in 19% of infants with TOGV and an associated ventricular septal defect and in 0.6% of patients with an intact ventricular septum.13 Echocardiography can also help the clinician visualize the response of the patent ductus arteriosus to a prostaglandin infusion.

Tetralogy of Fallot

Tetralogy of Fallot is the most common cyanotic congenital heart defect. Hallmarks of this lesion are (1) large, nonrestrictive ventricular septal defect; (2) severe right ventricular outflow tract obstruction; (3) overriding of the aorta; and (4) right ventricular hypertrophy. Timing of presentation and degree of cyanosis correlates precisely with the amount of right ventricular outflow tract obstruction. Patients with severe outflow tract obstruction (blue tets) present early in the neonatal period. Those patients with adequate pulmonary blood flow at birth (pink tets) gradually develop increasing cyanosis during the first few weeks and months of life. These patients may present to the emergency department with hypercyanotic “tet” spells, which occur as a consequence of transient reductions in pulmonary blood flow.15
Cyanosis is the most common presenting complaint in infants with tetralogy who are not diagnosed in utero. Hypercyanotic tet spells are the most dramatic presentation of this condition and are characterized by profound cyanosis, respiratory distress and grunting, and fussiness and agitation. These spells are brought on by increased right-to-left shunting and should be treated in a stepwise fashion with oxygen, knee-chest position, morphine sulfate, β-blockade, and phenylephrine. Knee-chest positioning of the infant is thought to increase systemic vascular resistance and decrease right-to-left shunting. Morphine acts to calm agitation and decrease venous return. β-Blockade slows the heart rate and allows more blood to enter the pulmonary circuit across the right ventricular outflow tract, and phenylephrine infusion increases systemic vascular resistance. Tetralogy of Fallot may also be suggested on chest radiography by the presence of a “boot-shaped” heart, which is caused by the absence of the small, atretic pulmonary arteries (Fig. 5), although diagnosis is most often made by echocardiography.

**Ebstein anomaly**

Ebstein anomaly is a cardiac abnormality of the tricuspid valve and right ventricle in which the valve is displaced downward and there is “atrialization” of a portion of the ventricle. This lesion is associated with tricuspid valvular insufficiency and stenosis, and patients with these anomalies often have accessory atrioventricular conduction pathways or intraventricular conduction delays that predispose them to arrhythmias. Patients can also have other structural cardiac anomalies, the most common of which is an atrial septal defect, which occurs in 80% to 94% of patients with Ebstein anomaly. Patients presenting with Ebstein anomaly have a high mortality rate, with one study showing that 18% of newborns presenting with Ebstein anomaly died in the neonatal period.

Ebstein anomaly may present with cyanosis, right-sided heart failure, arrhythmias, or sudden cardiac death. Prognosis in these patients is determined by degree of cyanosis and presence or absence of arrhythmias.
Chest radiographs reveal dramatic cardiomegaly, sometimes so extreme that the heart occupies most of the thoracic cavity. The heart is described as “globe-shaped” and may have a narrow waist. Echocardiography is the test of choice for diagnosing Ebstein anomaly and demonstrates the degree of inferior displacement of the proximal attachments of the tricuspid valvular septal leaflet.

An electrocardiogram should be obtained, because patients with Ebstein anomaly often have associated arrhythmias. Other electrocardiographic abnormalities seen with Ebstein anomaly include tall and broad P waves, prolonged PR interval, and complete or incomplete bundle branch block.

Initial treatment of these infants is first directed at decreasing pulmonary vascular resistance and preserving pulmonary blood flow. Prostaglandin E\textsubscript{1} should be initiated in an infant suspected of having Ebstein anomaly for maintenance of the patent ductus arteriosus.

Lesions Presenting as CHF

Truncus arteriosus

Patients with truncus arteriosus have only one great artery leaving the heart. This gives rise to the coronary arteries, aorta, and pulmonary arteries. There is a single semilunar valve, which is frequently regurgitant or stenotic. Patients with truncus arteriosus always have an associated large ventricular septal defect, and left and right ventricular pressures are equal. In addition, systemic and pulmonary arterial pressures are equal, resulting in pulmonary hypertension. Pulmonary arterial flow is frequently increased leading to pulmonary congestion and interstitial edema.

Physical examination findings in truncus arteriosus include tachypnea and diaphoresis, especially with feedings. As the pulmonary vascular resistance falls over the first few days of life, these symptoms become more prominent and patients frequently have a component of CHF as a result of the increased pulmonary blood flow. Mild cyanosis may be present. Pulses are bounding because of diastolic runoff into the
pulmonary arteries. A right ventricular impulse is palpable at the left lower sternal border. Usually, a murmur and systolic ejection click is heard within the first several days of life because of the high-flow state.6

Chest radiography shows cardiomegaly and evidence of increased pulmonary blood flow, including pulmonary vascular congestion and interstitial edema. A right-sided aortic arch may be present. Truncus arteriosus should be strongly suspected in the setting of a right-sided aortic arch, a systolic ejection click, increased pulmonary vascular markings, and mild cyanosis.

Echocardiography is the modality of choice for diagnosis and reveals the large ventricular septal defect, single great artery, dysplastic truncal valve, and the divergence of the pulmonary arteries from the truncus. Doppler echocardiography can identify stenosis or regurgitation across the truncal valve. Initial stabilization of these patients involves treatment of CHF, as outlined previously.

**Total anomalous pulmonary venous return**
TAPVR occurs when the pulmonary veins do not connect to the left atrium and instead drain into other embryologic venous structures that would typically involute in the setting of correct pulmonary vein attachment to the left atrium. Anomalous pulmonary venous return is total or partial. Partial anomalous pulmonary venous return has some pulmonary veins draining normally to the left atrium and some “anomalous” pulmonary veins draining to other venous structures in the thorax. In patients with TAPVR, none of the pulmonary veins drain into the left atrium. The four forms of TAPVR are (1) supracardiac, (2) cardiac, (3) infracardiac, and (4) mixed. With all forms of TAPVR, an atrial septal defect is required for any blood to reach the left side of the heart. There is mixing of oxygenated and deoxygenated blood in the right atrium, and thus the blood that crosses the atrial septal defect to the left heart is not fully oxygenated. There is an increase volume load to the right atrium, which results in dilation of the right atrium, right ventricle, and pulmonary arteries.

Physical examination findings in neonates with TAPVR include mild to moderate cyanosis. As the pulmonary vascular resistance decreases after birth, signs of CHF develop with tachypnea and diaphoresis. Pulmonary rales and a hepatomegaly may be found. Patients with infradiaphragmatic TAPVR typically present more acutely than other subtypes and are more likely to become hemodynamically unstable.

Findings on chest radiograph include an enlarged cardiac size with increased pulmonary vascular markings. In cases of supracardiac TAPVR the cardiac silhouette resembles a figure eight or snowman in shape because of the dilated vertical vein and superior vena cava. As with other lesions, echocardiography is the standard diagnostic modality for diagnosing TAPVR and determining the location and drainage of all four pulmonary veins is essential.

Initial treatment of TAPVR should focus on management of heart failure. Some infants present with cyanosis as the chief complaint and may be started on prostaglandin E\(_1\) as a stabilizing maneuver. TAPVR is the only cause of neonatal cyanosis that may be worsened by administration of prostaglandin E\(_1\). In the presence of a prostaglandin E\(_1\) infusion, pulmonary blood flow increases further through the patent ductus arteriosus causing worsening pulmonary vascular congestion and edema. If a cyanotic child with suspected congenital heart disease has worsening respiratory distress after initiation of prostaglandins, TAPVR should be suspected.

**Ventricular septal defect**
Ventricular septal defect, defined as a hole in the ventricular septum, is the most common congenital heart defect in children. At birth, the pressures in the right and
left ventricles are equal, there is no shunting, and the ventricular septal defect may not be detected. As the pulmonary vascular resistance decreases after birth, blood begins to flow through the defect from the left ventricle to the right ventricle and recirculate through the lungs. As the shunt volume increases, both ventricles and the left atrium dilate, and right ventricular volume overload may cause hepatomegaly or pulmonary congestion. In the setting of a large defect, tachypnea and failure to thrive ensue. Pulmonary hypertension occurs when the high left ventricular pressure is transmitted through the defect to the pulmonary circulation.

History and physical examination components vary depending on the size and resulting hemodynamic significance of the defect. Patients with a small ventricular septal defect have a loud, holosystolic murmur, usually audible starting shortly after birth. They are generally otherwise asymptomatic. Patients with a moderate-to-large ventricular septal defect typically present with poor weight gain. They are often dyspneic and diaphoretic, especially with feedings. Feeding problems and irritability are common, and infants may be misdiagnosed with colic or gastroesophageal reflux. These infants also have hepatomegaly, and systemic perfusion can be compromised.21

Chest radiographs may demonstrate cardiomegaly and increased pulmonary blood flow as CHF develops. Echocardiography is the most accurate tool for diagnosing and characterizing a ventricular septal defect. An echocardiogram can also evaluate the hemodynamic significance of the lesion, including direction and volume of flow across the ventricular septum.22

**Patent ductus arteriosus**

In this abnormality, the ductus arteriosus remains persistently open after birth. Because the aortic pressure exceeds the pulmonary arterial pressure throughout the cardiac cycle, there is a continuous flow of blood from the aorta to the pulmonary artery. Postnatal increases in pulmonary blood flow in the setting of prematurity can lead to pulmonary edema, loss of lung compliance, and deterioration of respiratory function, which ultimately leads to chronic lung disease. Large amounts of left-to-right shunting through the ductus may also increase the risk of intraventricular hemorrhage, necrotizing enterocolitis, and death.23 Persistent patent ductus arteriosus is much more common in premature infants and in patients with lung disease.24 Genetic factors, environmental exposures, such as medications, and prenatal infections, such as rubella, may also play a role in persistent patency.25

Patients with symptomatic patent ductus arteriosus typically present as dyspneic, especially with activity. The hallmark physical finding in patients with a patent ductus arteriosus is a murmur, described as continuous and “machinery-like.” Peripheral pulses are typically bounding. Some patients present primarily in atrial fibrillation because of chronic and progressive left atrial enlargement.

Chest radiographs in patients with patent ductus arteriosus can be normal or may display cardiomegaly with increased pulmonary vascular markings. Electrocardiogram is often normal in patients with small shunts but may show sinus tachycardia, atrial fibrillation, left ventricular hypertrophy, or left atrial enlargement with moderate-to-large shunts. Diagnosis can be made and the degree of shunting can be verified by echocardiography.

Stabilization of a symptomatic patient with a patent ductus arteriosus can generally be achieved with digoxin and diuretics. Afterload-reducing agents, such as angiotensin-converting enzyme inhibitors, may also improve clinical status. Patients presenting with arrhythmias, such as atrial fibrillation or flutter, should be treated with antiarrhythmics or cardioversion and maintained on antiarrhythmics, β-blocking medications, and anticoagulants as indicated.
Most patients have spontaneous closure of their ductus in the first days to months of life. However, if it does not close within the first year, it is very unlikely to do so spontaneously. At that point, intervention to promote closure is recommended to prevent long-term pulmonary complications and infective endocarditis, for which these patients are at increased risk. Nonselective cyclooxygenase inhibitors, such as indomethacin or ibuprofen, are generally used to induce ductus closure, but these may be less effective in severely premature infants.

**Congenital Cardiac Arrhythmias**

**Supraventricular tachycardia**

Supraventricular tachycardia is the most common symptomatic arrhythmia in the pediatric population, and 60% to 80% of these patients present in the first year of life. The tachycardia is most commonly narrow complex, and the rate is typically greater than 220 beats per minute, but it can be up to 300 beats per minute. Atrioventricular reentrant tachycardia, including the Wolff-Parkinson-White syndrome, can result from an accessory bypass tract between the atria and ventricles. Atrioventricular nodal reentrant tachycardia occurs when there is a reentrant pathway within the atrioventricular node. Most patients with supraventricular tachycardia have structurally normal hearts, but 25% have an associated congenital heart defect. Presenting complaints with sustained tachycardia include irritability and feeding intolerance. If the duration of tachycardia is prolonged, the patient may develop CHF. Diagnosis is established by characteristic findings on electrocardiogram. A narrow-complex tachycardia with a rate of 220 to 300 beats per minute or regular sinus rhythm with a Wolff-Parkinson-White pattern, characterized by a short PR interval and a slurred, delta-shaped initial segment of the QRS complex, on electrocardiogram is highly suggestive of the diagnosis.

Initial treatment for supraventricular tachycardia includes attempts at blocking the atrioventricular node, because most of the supraventricular tachycardias involve the atrioventricular node as part of the electrical circuit. Vagal maneuvers can be attempted first, such as application of a bag of ice to the face. Adenosine, a potent atrioventricular nodal blocker, is the drug of choice for supraventricular tachycardia. It is successful at terminating supraventricular tachycardia in 80% to 90% of children. The initial dose is 0.1 mg/kg to a maximum of 6 mg and subsequent doses should be 0.2 mg/kg to a maximum of 12 mg. Other medication options include digoxin and propranolol. Verapamil should not be used in infants. An unstable child in supraventricular tachycardia should be rapidly direct-current cardioverted using 0.5 to 1 J/kg.

Stable children in sinus rhythm can be discharged home with pediatric cardiology follow-up. Patients may go home on an antiarrhythmic, such as digoxin or propranolol, but this decision should be made in consultation with pediatric cardiology. Many infants outgrow their supraventricular tachycardia by 1 year of age. Patients with persistent episodes of tachycardia after 1 year and despite aggressive medical therapy may require radiofrequency catheter ablation.

**Long QT syndrome**

Congenital long QT syndrome is a genetic disorder characterized by prolongation of the QT interval on electrocardiogram. This leads to a propensity for developing torsades de pointes polymorphic ventricular tachycardia in the setting of adrenergic stimulation. Long QT syndrome is highly genetically linked and is principally caused by mutations of the genes encoding potassium and sodium ion channels or proteins associated with these channels.
Patients with long QT syndrome often present primarily with palpitations, syncope, cardiac arrest, or sudden death. These patients are typically young and otherwise healthy. Any patient with palpitations, syncope, cardiac arrest, or sudden death requires that a thorough family history be obtained. In the setting of a family history of cardiac events at a young age or unexplained syncope or death, long QT syndrome needs to be strongly considered.

A 12-lead electrocardiogram should be obtained to confirm the diagnosis. QT interval should be measured, and an age- and gender-specific corrected QT interval (QTc) should be calculated. Patients with a QTc of greater than 500 milliseconds and symptomatic episodes are highly likely to have disease. A symptomatic patient with a borderline QTc of 440 to 500 milliseconds needs further evaluation. β-Blockers are the therapy of choice for long-term management of long QT syndrome to blunt the adrenergic surges associated with deterioration of sinus rhythm to torsades de pointes and then to ventricular fibrillation. Patients should be counseled to avoid activities associated with increased catecholamine release including exercise and swimming, high emotions, loud noise, or sudden awakening from sleep. In symptomatic patients with a QTc greater than 440 milliseconds, urgent referral to pediatric cardiology is imperative. Patients with a QTc greater than 500 milliseconds require cardiology consultation while still in the emergency department. In consultation with pediatric cardiology, these patients should be admitted to the hospital pending placement of the cardioverter-defibrillator or may possibly be discharged home with a portable external defibrillator and family education regarding its use. All first-degree relatives of definitively diagnosed patients should be screened for the disorder with electrocardiogram.

**Complete heart block**

Congenital complete heart block in infants and children is relatively uncommon and in up to 30% patients there are associated structural heart defects. Most of these patients are diagnosed en utero. Maternal risk factors include systemic lupus erythematosus. Acquired complete heart block is most typically a result of cardiac surgery for congenital heart disease; myocarditis; or infectious etiologies, such as Lyme disease.

Patients with symptomatic complete heart block present with signs and symptoms of hypoperfusion, including syncope or sudden death. Physical findings include bradycardia and cannon waves in the neck, generated by atrial contraction against closed tricuspid valves.

Diagnosis is achieved by electrocardiogram. Complete heart block as a result of Lyme carditis has a low morbidity, resolves spontaneously, and does not require any specific treatment. Definitive treatment for other causes of complete heart block is placement of a permanent pacemaker. In a hemodynamically unstable patient in the emergency department, sedation and transcutaneous pacing may be required.

**SPECIAL CONSIDERATIONS IN THE TREATMENT OF ADULTS WITH CONGENITAL HEART DISEASE**

As our surgical techniques, understanding of multisystem effects, and medical management of congenital heart disease have improved, the duration of survival with congenital heart disease has increased. The prevalence of severe congenital heart disease in adults increased 85% between 1985 and 2000, significantly outpacing the prevalence in the pediatric population. Special circumstances, including pregnancy in a patient with congenital heart disease, must be considered as women with congenital heart disease live to childbearing age and beyond. In addition, adult
patients with congenital heart disease may present to the emergency department with
decompensation of their chronic cardiac conditions. Most of the acute presentations
of adults with congenital heart disease involve heart failure, respiratory compromise,
and dysrhythmias.

Patients with long-standing congenital heart disease may develop CHF. Left-sided
and right-sided lesions may be treated very differently in the setting of heart failure.
Patients with primarily left-sided disease, including left ventricular dysfunction and
aortic or mitral valve disease with either low cardiac output or left atrial hypertension,
may benefit substantially from positive pressure ventilation. Positive pressure
increases intrathoracic pressure, decreasing afterload and allowing for more effective
forward flow of blood. In contrast, patients with right-sided lesions likely decline
further with the initiation of positive pressure ventilation. As intrathoracic pressure is
increased, systemic vascular return to the heart decreases. This lowers cardiac pre-
load and leads to decreased cardiac output and circulatory collapse. The remainder
of supportive measures applicable to all other patients presenting with CHF including
nitrates, diuretics, supplemental oxygen, and positive inotropy also apply to patients in
heart failure with congenital heart disease.

Adult patients with congenital heart disease may develop pulmonary complications,
such as pulmonary artery hypertension. Unrepaired systemic-to-pulmonary vascular
connection, such as ventricular septal defect, patent ductus arteriosus, or atrioventric-
ular canal, can result in increased pressure through the pulmonary vasculature. Known
as Eisenmenger syndrome, these patients go on to develop cyanosis and its associ-
ated complications, such as polycythemia, headaches, blurred vision, and strokes.
Shortness of breath ensues, and hemoptysis is not uncommon with advanced
disease. Supplemental oxygen, digitalis, diuretics, phlebotomy, and anticoagulation
may help mitigate some of the symptoms and complications of the disease. However,
one the pathophysiologic changes have occurred, the process is generally progres-
sive and irreversible.31

More than 50% of patients with congenital heart disease develop arrhythmias in
their lifetime. Arrhythmias are the leading cause of hospital admissions for adults
with congenital heart disease, and they constitute a significant predictor of mortality.30
For patients with compromised cardiac output at baseline, loss of synchronous atrio-
ventricular activity can cause hemodynamic compromise and collapse. Treatment is
supportive, and intervention should be in collaboration with an electrophysiologist
comfortable in the management of patients with congenital heart disease.

SUMMARY

Pediatric congenital heart disease comprises a wide spectrum of structural defects.
These lesions, although many, tend to present in a limited number of ways. An infant
presenting with profound shock, cyanosis, or evidence of CHF should raise the suspi-
cion of congenital heart disease. Although most congenital lesions are diagnosed in
utero, the emergency physician must be aware of these cardinal presentation because
patients present in the postnatal period around the time that the ductus arteriosus
closes. Aggressive management of cardiopulmonary instability combined with empiric
use of prostaglandin E1, and early pediatric cardiology consultation is essential for
positive outcomes.

REFERENCES