with the blood reentering the systemic circulation. Cyanotic defects are those in which poorly oxygenated blood mixes with the blood reentering the systemic circulation.

Search the American Heart Association website for congenital heart defects (go to http://www.heart.org). About 40,000 babies in the United States are born with congenital heart defects.

Acyanotic Defects

Acyanotic defects occur when the blood flows from the left side of the heart to the right side of the heart due to a hole in the interventricular septum. Acyanotic defects do not normally interfere with the oxygen or blood reaching the body's tissues. The most common acyanotic defects include the following:

- *Ventricular septal defect* (VSD; ICD-10: Q21.0) is the most commonly occurring congenital heart defect, where there is an abnormal opening between the wall, or **septum,** of the right and left ventricles (Fig. 6.1). The extent of the opening may vary from the size of a pin to a complete absence of the ventricular septum, creating one common ventricle. Blood from the left ventricle flows back into the right ventricle, causing too much blood to be pumped to the lungs. This results in lung congestion. This defect typically accompanies other congenital anomalies, especially trisomy 21 (Down syndrome), renal defects, or other cardiac defects. This defect occurs in 2% to 7% of live births.
- *Atrial septal defect* (ASD; ICD-10: Q21.1) is an abnormal opening between the right and left atria (Fig. 6.2). The size and location of the opening determine the severity of the defect. If the hole is large, a fair amount of oxygen-rich blood leaks back to the right atria and is then pumped back into the lungs, which are already rich in oxygen.
- *Coarctation of the aorta* (ICD-10: Q25.1) is a malformation in a portion of the aorta wall that causes narrowing of the aortal opening, or **lumen,** at the point of the defect (Fig. 6.3). Consequently, blood pressure is increased proximal to the defect and decreased distal to it. Congestive heart failure may result. This defect can range from mild to severe and may not be recognized until the patient becomes an adult. It usually occurs with other heart defects.
- *Patent ductus arteriosus* (PDA; ICD-10: Q25.0) is a defect resulting from the failure of the **ductus arteriosus,** a connection between the aorta and the pulmonary artery in the fetus, to close after birth (Fig. 6.4). During the prenatal period, much of the fetal circulation bypasses the lungs through this blood vessel, which connects the pulmonary artery to the aorta. When this fetal structure fails to close

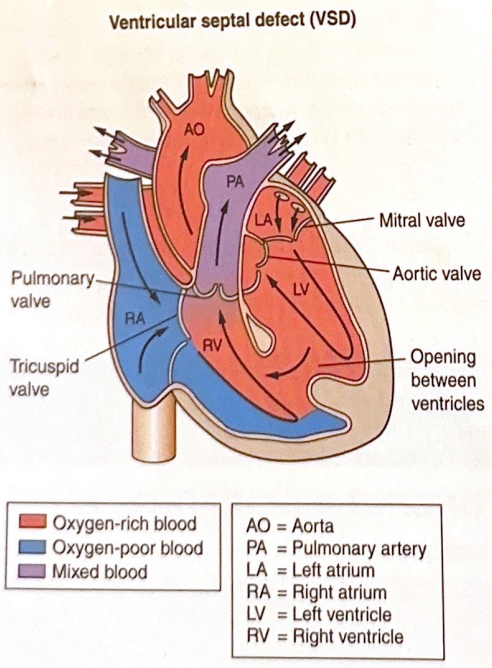

FIGURE 6.1 Ventricular septal defect (VSD). In VSD, a hole in the ventricular septum occurs, and blood from the left ventricle flows back into the right ventricle, due to higher pressure in the left ventricle. This backflow causes an extra volume of blood to be pumped into the lungs by the right ventricle, creating lung congestion.

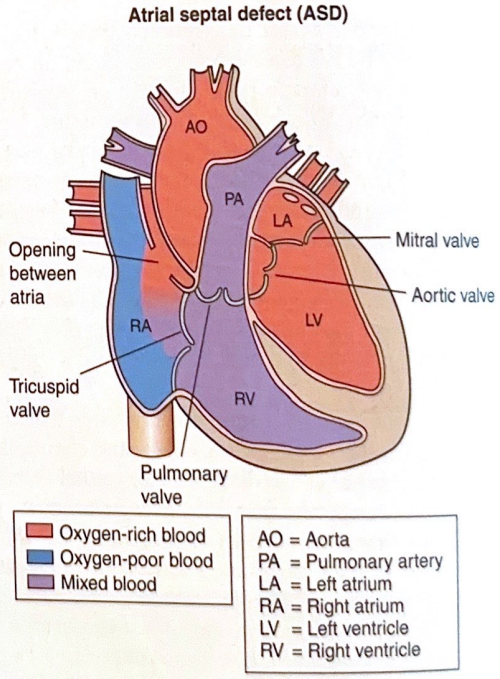

FIGURE 6.2 Atrial septal defect (ASD). In ASD, there is an abnormal opening between the two atria of the heart, causing an abnormal blood flow through the heart. Some infants have no symptoms and appear healthy. If the ASD is large and permits a large amount of blood to pass through the right side, symptoms will be noted.

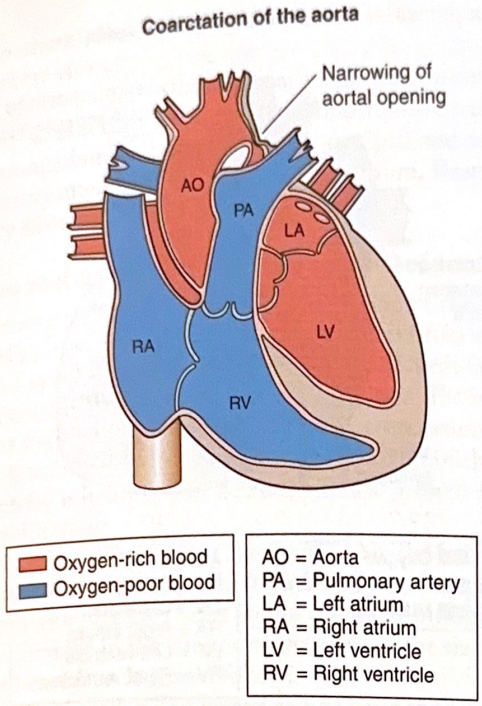

FIGURE 6.3 Coarctation of the aorta (coarct). In this condition, the aorta is narrowed or constricted, obstructing blood flow to the lower part of the body and increasing blood pressure above the constriction. Usually there are no symptoms at birth, but they can develop as early as the first week of life. If severe symptoms of high blood pressure and congestive heart failure develop, surgery may be considered.

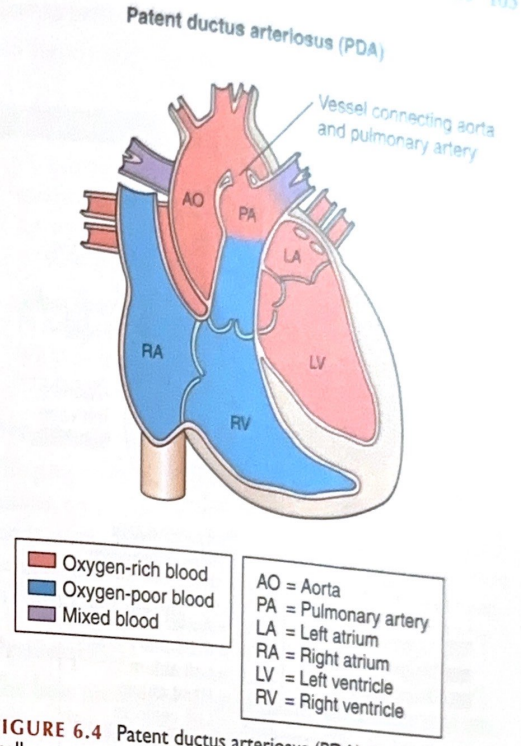

FIGURE 6.4 Patent ductus arteriosus (PDA). This defect normally occurs during fetal life. PDA short-circuits the normal pulmonary vascular system and allows blood to mix between the pulmonary artery and the aorta. Before birth, there is an open passageway between the two blood vessels that closes soon after birth. When it does not close, some blood returns to the lungs. PDA is often seen in premature infants.

after birth, blood from the aorta flows back into the pulmonary artery. This defect is common in premature infants and puts a strain on the heart, causing **tachypnea,** or fast breathing. A small patent ductus arteriosus may not require treatment; however, larger ones left untreated can cause weakening of the heart muscle, in turn causing heart failure.

Cyanotic Defects

Cyanotic defects cause the oxygen-rich blood and the oxygen-poor blood to mix, allowing less oxygen-rich blood to reach the body tissues. Often a bluish tint to the skin results. The most common cyanotic defects follow:

- *Tetralogy of Fallot* (ICD-10: Q21.3) is a combination of four congenital heart defects: (1) pulmonary stenosis, a narrowing of the opening into the pulmonary artery from the right ventricle; (2) VSD, an abnormal opening in the septum between the left and right ventricles; (3) dextroposition of the aorta, in which the opening of the aorta bridges the ventricular septum, receiving blood from both the left and right ventricles; and (4) right ventricular **hypertrophy,** an increase in size or volume (Fig. 6.5). Diagnosis of this defect most often occurs in infancy.
- *Transposition of the great arteries* (TGA; ICD-10: Q20.3) is a condition in which the two major arteries of the heart are reversed, with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle (Fig. 6.6). The result is two noncommunicating circulatory systems—one circulating blood in a closed loop between the heart and lungs and the other between the heart and systemic circulation. Why this defect occurs is not known. This is a rare defect found at birth.
- *Tricuspid atresia* (ICD-10: Q22.4) is a condition in which the valve between the right atrium and the right ventricle fails to develop (Fig. 6.7). This defect may be combined with ASD, VSD, and transposition of the great arteries. This defect is found at birth and is treated with surgery. The symptoms become pronounced immediately after birth and are characterized by cyanosis, fatigue, breathing difficulties, and slow growth rate.

Etiology

The etiology of congenital heart defects is mostly unknown, but there may be a genetic link. Predisposing factors may include maternal infections, use of certain drugs during gestation, diabetes, alcoholism, and poor maternal nutrition. In roughly 85% of cases, however, there is no

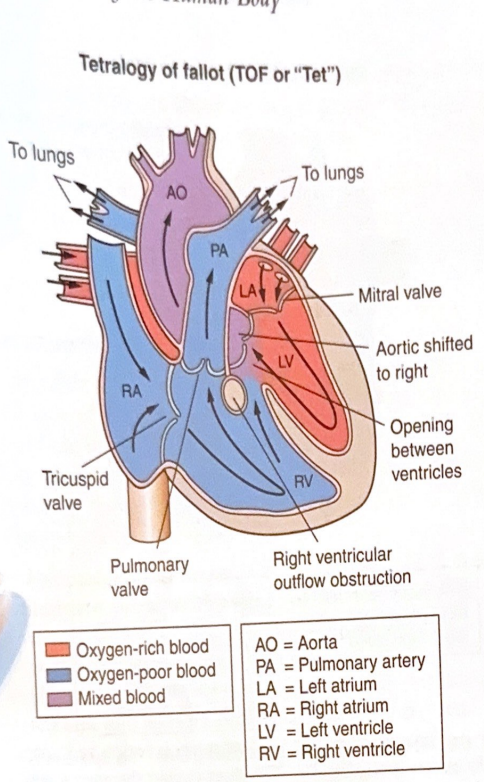

FIGURE 6.5 Tetralogy of Fallot. This condition is characterized by four defects: (1) An abnormal opening or ventricular septal defect allows blood to pass from the right ventricle to the left ventricle without going through the lungs. (2) A narrowing or stenosis at or just beneath the pulmonary valve partially blocks the flow of blood from the right side of the heart to the lungs. (3) The right ventricle is more muscular than normal. (4) The aorta lies directly over the ventricular septal defect. Tetralogy of Fallot results in cyanosis due to lack of oxygen.

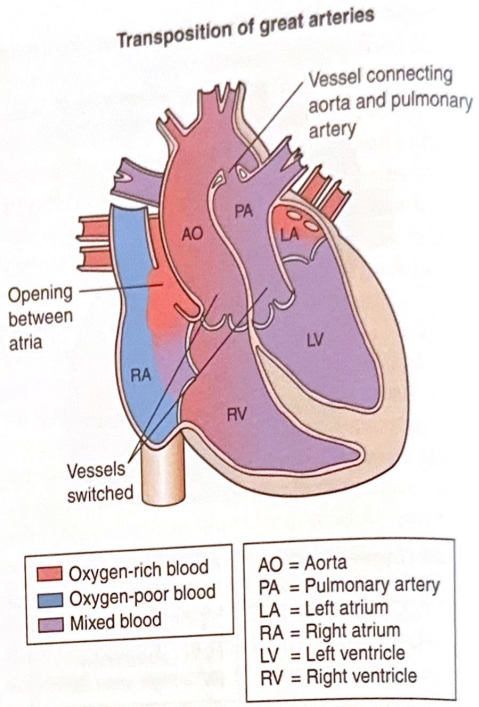

FIGURE 6.6 Transposition of the great arteries. With this congenital defect, the positions of the pulmonary artery and the aorta are reversed. Thus, the aorta originates from the right ventricle, so most of the blood returning to the heart from the body is pumped back out without first going to the lungs, and the pulmonary artery originates from the left ventricle, so that most of the blood returning from the lungs goes back to the lungs again.

one identifiable cause of a congenital heart defect. The abnormalities are thought to be multifactorial—both genetic and environmental. Clinical features vary with age and seriousness of the defect.

Signs and Symptoms of Acyanotic Defects

- *VSD:* The classic clinical feature is a loud, early systolic murmur heard during **auscultation,** or listening for sounds produced by the internal organs. The typical murmur is described as blowing or rumbling. There may be signs of rapid, heavy breathing; poor feeding; poor weight gain; and sweating.
- *ASD:* There are very few symptoms of ASD. The classic clinical feature is a crescendo-decrescendo type of systolic ejection murmur. A large hole can cause **arrhythmias** and congestive heart failure in middle age.
- *Coarctation of the aorta:* The clinical features vary with age. A murmur may or may not be present. An infant may exhibit dyspnea, pulmonary edema, **tachycardia** (an abnormally rapid heartbeat), and failure to thrive. Symptoms appearing after adolescence may include dyspnea, **claudication**

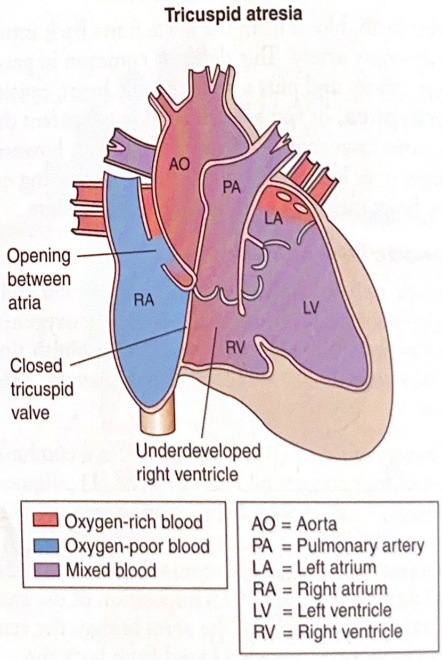

FIGURE 6.7 Tricuspid atresia. In tricuspid atresia, there is no tricuspid valve; therefore, no blood flows from the right atrium to the right ventricle. Tricuspid atresia defect is characterized by a small right ventricle, a large left ventricle, diminished pulmonary circulation, and cyanosis. A surgical shunting procedure is often necessary to increase the blood flow to the lungs.

(lameness), headache, **epistaxis** (nosebleed), and hypertension.
- *PDA:* The clinical feature is a "machinery" murmur usually associated with an abnormal tremor accompanying a cardiac murmur or thrill and often accompanied by a widened pulse pressure. Respiratory distress and cyanosis are common.

Signs and Symptoms of Cyanotic Defects

- *Tetralogy of Fallot:* A bluish discoloration of the skin and mucous membranes, or **cyanosis,** is often evident at birth or within several months of birth and is considered the hallmark of the disorder. The infant may exhibit other signs of poor oxygenation, such as increasing dyspnea on exertion, diminished exercise tolerance, and delayed physical growth and development.
- *Transposition of the great arteries* (TGA): The infant is typically severely cyanotic at birth and has tachypnea. Signs of congestive heart failure and **cardiomegaly** (an increase in the volume of the heart or the size of the heart muscle tissue) follow.
- *Tricuspid atresia:* The extent of VSD and the relationship of the great arteries will determine the symptoms. The neonate will have low oxygen levels and cyanosis. There can also be tachypnea and poor feeding.

Diagnostic Procedures

Signs and symptoms often point to a diagnosis, but a history and physical examination are essential and may be all that are necessary to diagnose some congenital heart abnormalities. Other diagnostic procedures may include an echocardiogram, chest x-rays, an electrocardiogram (ECG), magnetic resonance imaging (MRI), pulse oximetry, and heart catheterization. Laboratory studies may be ordered to determine the degree of cyanosis and to detect possible acidosis.

Treatment

Some congenital heart defects require no treatment because there is spontaneous closure of the defects, or some medications may be effective in closing defects. If surgery is necessary, it usually is done during the first year of life. Surgery may include closing holes in the heart with patches or stitches, repairing or replacing heart valves, or widening arteries or openings to heart valves. When the defect is complex, however, more than one surgical procedure may be required. Some surgical procedures may be delayed until the child is old enough to better withstand the surgery. Heart catheterizations are also used in treatment. A needle puncture is made in the skin to insert a catheter into a vein or artery to repair some defects. Because this procedure is much easier than surgery on an individual with a birth defect, it is the preferred treatment when possible.

Complementary Therapy

No significant complementary therapy is indicated.

CLIENT COMMUNICATION

Any heart defect is frightening to all involved. Reassurance is important and should include explanations to the parents and caregivers of any procedures to be performed. Search the Internet for the March of Dimes website for information helpful to parents who are trying to understand what is happening and why.

Prognosis

The prognosis is dependent on the type of defect, its location, and its severity. If the defect is small and treatment is successful, the prognosis is quite good. Some defects, however, are so severe as to put the infant's life in immediate danger.

Prevention

The best prevention begins before pregnancy and continues through the first trimester. The following steps are recommended:

- Confirm immunity to rubella.
- Take a daily multivitamin that contains folic acid.
- Avoid use of all recreational drugs.
- Avoid viral infections, especially of the upper respiratory tract.
- Avoid alcohol.
- Do not take prescription medications, such as lithium or drugs that treat acne and seizures. (*Note:* Women with diabetes or seizure difficulties are advised to consult their primary care provider (PCP) before pregnancy to minimize risk to the fetus while maintaining proper management of their health issues.)
- Avoid x-rays, strong chemicals, and solvents.
- Consider genetic counseling if another child or a relative has a congenital heart defect.

REALITY EPISODE

Expectant parents were shocked when they learned from their ultrasound at 30 weeks' gestation that their baby had a heart defect—tetralogy of Fallot. Fortunately, they had some time to prepare. They knew there would be a long hospital stay, transfer to a children's hospital 3 hours away, and surgery to correct the defect. They named their unborn child Rachael. Rachael was born just 3 weeks later.

1. Describe events or circumstances that Rachael's parents might not have expected or anticipated.
2. Is their location a consideration?