Cyanotic Congenital Heart Disease:
Lesions Associated with Increased Pulmonary Blood Flow
D-Transposition of the Great Arteries

Transposition of the great vessels, a common cyanotic congenital anomaly, accounts for ≈5% of all congenital heart disease. In this anomaly, the systemic veins return normally to the right atrium and the pulmonary veins return to the left atrium. The connections between the atria and ventricles are also normal (atrioventricular concordance). The aorta arises from the right ventricle and the pulmonary artery from the left ventricle. Desaturated blood returning from the body to the right side of the heart goes inappropriately right out the aorta and back to the body again, whereas oxygenated pulmonary venous blood returning to the left side of the heart is returned directly to the lungs. The systemic and pulmonary circulations consist of two parallel circuits. Survival in these newborns is provided by the foramen ovale and the ductus arteriosus, which permit some mixture of oxygenated and deoxygenated blood. About 50% of patients with TGA also have a ventricular septal defect (VSD), which provides for better mixing. TGA is more common in infants of diabetic mothers and in males (3 : 1). TGA, especially when accompanied by other cardiac defects such as pulmonic stenosis or right aortic arch, can be associated with deletion of chromosome 22q11 (CATCH 22 [cardiac defects, abnormal facies, thymic aplasia, cleft palate, hypoplasia], DiGeorge syndrome). Before the modern era of corrective or palliative surgery, mortality was >90% in the 1st yr of life.

D-Transposition of the Great Arteries with Intact Ventricular Septum
D-TGA with an intact ventricular septum is also referred to as simple TGA or isolated TGA. Before birth, oxygenation of the fetus is nearly normal, but after birth, once the ductus begins to close, the minimal mixing of systemic and pulmonary blood via the patent foramen ovale is usually insufficient and severe hypoxemia ensues, generally within the 1st few days of life.

CLINICAL MANIFESTATIONS
Hypoxemia is usually severe, but heart failure is less common. This condition is a medical emergency, and only early diagnosis and appropriate intervention can avert the development of prolonged severe hypoxemia and acidosis, which lead to death. Murmurs may be absent, or a soft systolic ejection murmur may be noted at the mid left sternal border.

DIAGNOSIS
The electrocardiogram shows the normal neonatal right-sided dominant pattern. Roentgenograms of the chest may show mild cardiomegaly, a narrow mediastinum (hence an egg-shaped heart), and normal to increased pulmonary blood flow. Echocardiography confirms the transposed ventricular-arterial connections. Cardiac catheterization is occasionally performed in patients for whom noninvasive imaging is diagnostically inconclusive or in patients who require emergency balloon atrial septostomy.

TREATMENT
When transposition is suspected, an infusion of prostaglandin E1 should be initiated immediately to maintain patency of the ductus arteriosus and improve oxygenation (dosage, 0.01–0.20 μg/kg/min). Hypothermia intensifies the metabolic acidosis resulting from hypoxemia, and thus the patient should be kept warm. Prompt correction of acidosis and hypoglycemia is essential.Infants who remain severely hypoxic or acidotic despite prostaglandin infusion should undergo Rashkind balloon atrial septostomy. A Rashkind atrial septostomy is usually performed in all patients in whom any significant delay in surgery is necessary. The arterial switch (Jantene) procedure is the surgical treatment of choice for neonates with d-TGA and an intact ventricular septum and is usually

performed within the 1st 2 wk of life. Previous operations for d-TGA consisted of some form of atrial switch procedure (Mustard or Senning operation). In older infants, these procedures produced excellent early survival (≈85–90%), but significant long-term morbidity.

Transposition of the Great Arteries with Ventricular Septal Defect

If the VSD associated with TGA is small, the clinical manifestations, laboratory findings, and treatment are similar to those described previously for transposition with an intact ventricular septum. A harsh systolic murmur is audible at the lower left sternal border and results from flow through the defect. In equivocal cases, the diagnosis can be confirmed by cardiac catheterization. At the time of cardiac catheterization. Surgical treatment is advised soon after diagnosis, usually within the 1st months of life, because heart failure and failure to thrive are difficult to manage and pulmonary vascular disease can develop unusually rapidly. Management includes diuretics and, possibly, digitalis to lessen the symptoms of heart failure while awaiting surgical repair.

L-Transposition of the Great Arteries (Corrected Transposition)

In l-transposition, the atrioventricular relationships are discordant, with the right atrium connected to the left ventricle and the left atrium to the right ventricle (ventricular inversion). The great arteries are also transposed, with the aorta arising from the right ventricle and the pulmonary artery from the left. The aorta arises to the left of the pulmonary artery (hence the designation l for levotransposition). Because transposition is also present, desaturated blood ejected from this left ventricle enters the transposed pulmonary artery and flows into the lungs. The circulation is physiologically "corrected."

CLINICAL MANIFESTATIONS

Symptoms and signs are widely variable and are determined by the associated lesions. If pulmonary outflow is unobstructed, the clinical signs are similar to those of an isolated VSD. If the TGA is associated with pulmonary stenosis and a VSD, the clinical signs are similar to those of tetralogy of Fallot.

DIAGNOSIS

The chest roentgenogram may suggest the abnormal position of the great arteries. The echocardiogram is diagnostic. Surgical treatment of the associated anomalies, most often the VSD, is complicated by the position of the bundle of His, which can be injured at the time of surgery and result in heart block

Total Anomalous Pulmonary Venous Return

PATHOPHYSIOLOGY

Abnormal development of the pulmonary veins may result in either partial or complete anomalous drainage into the systemic venous circulation. Partial anomalous pulmonary venous return is usually an acyanotic lesion. Total anomalous pulmonary venous return (TAPVR) allows total mixing of systemic venous and pulmonary venous blood flow within the heart and thus produces cyanosis.
In TAPVR, the heart has no direct pulmonary venous connection into the left atrium. The pulmonary veins may drain above the diaphragm into the right atrium directly, into the coronary sinus, or into the superior vena cava via a "vertical vein," or they may drain below the diaphragm and join into a "descending vein" that enters into the inferior vena cava or one of its major tributaries, often via the ductus venous. This latter form of anomalous venous drainage is most commonly associated with obstruction, usually as the ductus venosus closes soon after birth, although supracardiac anomalous veins may also become obstructed. All forms of TAPVR involve mixing of oxygenated and deoxygenated blood before or at the level of the right atrium (total mixing lesion). The

manifestations of TAPVR depend on the presence or absence of obstruction of the venous channels.

Anomalous Pulmonary Venous Return

CLINICAL MANIFESTATIONS

Three major clinical patterns of TAPVR are seen. Some are manifested in the neonatal period as severe obstruction to pulmonary venous return, most prevalent in the infracardiac group . Cyanosis and severe tachypnea are prominent, but murmurs may not be present. Rapid diagnosis and surgical correction are necessary for survival. Another group is characterized by heart failure in early life, but these infants have only mild or moderate obstruction to pulmonary venous return and a large left-to-right shunt. Cyanosis is mild. The third group of patients with TAPVR consists of those in whom pulmonary venous obstruction is not present; these patients have total mixing of systemic venous and pulmonary venous blood and a large left-to-right shunt.

% AND SITE OF CONNECTION	% WITH SEVERE OBSTRUCTION
Supracardiac (50)	
superior vena cava	40
ght superior vena c (10)	75
Cardiac (25)	
Coronary sinus (20	10
Right atrium (5)	5
Infracardiac (20)	95–100
Mixed (5)	

DIAGNOSIS

The electrocardiogram demonstrates right ventricular hypertrophy. Roentgenograms are. A large supracardiac shadow can be seen, which together with the normal cardiac shadow forms a "snowman" appearance. The echocardiogram demonstrates a large right ventricle and usually identifies the pattern of abnormal pulmonary venous connections. The demonstration of a vessel in the abdomen with Doppler venous flow away from the heart is pathognomonic of TAPVR below the diaphragm. Cardiac catheterization shows that the oxygen saturation of blood in both atria, both ventricles, and the aorta is more or less similar, indicative of a total mixing lesion. MRI and CT may be alternative methods of confirming the diagnosis.

TREATMENT

Surgical correction of TAPVR is indicated during infancy. Before surgery, infants may be stabilized with prostaglandin E1 to dilate the ductus venosus and the ductus arteriosus, although with significant obstruction this is usually not effective. If surgery cannot be performed urgently. To date, the long-term prognosis in these patients is poor and heart-lung transplantation may be the only option.

Truncus Arteriosus

In truncus arteriosus, a single arterial trunk (truncus arteriosus) arises from the heart and supplies the systemic, pulmonary, and coronary circulations. A VSD is always present, with the truncus overriding the defect and receiving blood from both the right and left ventricles. The number of truncal valve cusps varies from two to as many as six. The pulmonary arteries may arise together from the posterior left side of the persistent truncus arteriosus and then divide into left and right pulmonary arteries (type I truncus arteriosus). In types II and III truncus arteriosus, no main pulmonary artery is present, and the right and left pulmonary arteries arise from separate orifices in the posterior (type II) or lateral (type III) aspects of the truncus arteriosus. Type IV truncus has no identifiable connection between the heart and pulmonary arteries, and pulmonary blood flow is derived from major aortopulmonary collateral arteries arising from the transverse or descending aorta.

CLINICAL MANIFESTATIONS
The clinical signs of truncus arteriosus vary with age and depend on the level of pulmonary vascular resistance. In the immediate newborn period, signs of heart failure are usually absent; a murmur and minimal cyanosis are the initial signs. Truncus arteriosus is a conotruncal malformation and may be associated with DiGeorge syndrome, which has been linked to a deletion of chromosome 22q11.

DIAGNOSIS
The electrocardiogram shows right, left, or combined ventricular hypertrophy. The chest roentgenogram also shows considerable variation. The truncus may produce a prominent shadow that follows the normal course of the ascending aorta and aortic knob; the aortic arch is to the right in 50% of patients. Echocardiography demonstrates the large truncal artery overriding the VSD and the pattern of origin of the branch pulmonary arteries. Pulsed and color Doppler studies are used to evaluate truncal valve regurgitation.

PROGNOSIS AND COMPLICATIONS
Without surgery, many of these patients succumb during infancy or by the 1st or 2nd yr of life. If pulmonary blood flow is restricted by the development of pulmonary vascular disease, the patient may survive into early adulthood. Surgical results have been very good, and many patients with repaired truncus are entering young adulthood.

TREATMENT
In the 1st few weeks of life, many of these infants can be managed with anticongestive medications; as pulmonary vascular resistance falls, heart failure symptoms worsen and surgery is indicated, usually in the next few weeks. Delay of surgery much beyond 4–8 wk may increase the likelihood of pulmonary vascular disease; some centers advocate neonatal repair at the time of diagnosis.

Tetralogy of Fallot

Tetralogy of Fallot (1) obstruction to right ventricular outflow (pulmonary stenosis), (2) ventricular septal defect (VSD), (3) dextroposition of the aorta with override of the ventricular septum, and (4) right ventricular hypertrophy . Obstruction to pulmonary arterial blood flow is usually at both the right ventricular infundibulum (subpulmonic area) and the pulmonary valve. Complete obstruction of right ventricular outflow (pulmonary atresia with VSD) is classified as an extreme form of tetralogy of Fallot. The degree of pulmonary outflow obstruction varies, with the severity of the obstruction determining the degree of the patient's cyanosis.

PATHOPHYSIOLOGY
More commonly, the subpulmonic or infundibular muscle, the crista supraventricularis, is hypertrophic, which contributes to the subvalvar stenosis. Pulmonary blood flow may be supplied by a patent ductus arteriosus (PDA) and by multiple major aortopulmonary collateral arteries (MAPCAs) arising from the ascending and descending aorta.

The VSD is usually nonrestrictive and large, is located just below the aortic valve. The aortic arch is right sided in 20% of cases, and the aortic root is usually large and overrides the VSD to a varying degree. When obstruction to right ventricular outflow is mild to moderate and a balanced shunt is present across the VSD, the patient may not be visibly cyanotic (acyanotic or “pink” tetralogy of Fallot). When obstruction is severe, cyanosis will be present from birth and worsen when the ductus begins to close.

CLINICAL MANIFESTATIONS

Often, cyanosis is not present at birth, but with increasing hypertrophy of the right ventricular infundibulum and patient growth, cyanosis occurs later in the 1st yr of life. Older children with long-standing cyanosis who have not undergone surgery may have dusky blue skin, gray sclerae with engorged blood vessels, and marked clubbing of the fingers and toes.

Paroxysmal hypercyanotic attacks (hypoxic,"blue," or "tet" spells) are a particular problem during the 1st 2 yr of life. The spells occur most frequently in the morning on initially awakening or after episodes of vigorous crying. The spells may last from a few minutes to a few hours but are rarely fatal. Short episodes are followed by generalized weakness and sleep. Severe spells may progress to unconsciousness and, occasionally, to convulsions or hemiparesis. Infants who are only mildly cyanotic at rest are often more prone to the development of hypoxic spells because they have not acquired the homeostatic mechanisms to tolerate rapid lowering of arterial oxygen saturation, such as polycythemia.

Depending on the frequency and severity of hyper cyanotic attacks, one or more of the following procedures should be instituted in sequence:-

(1) Placement of the infant on the abdomen in the knee-chest position while making certain that the infant's clothing is not constrictive.(2) Administration of oxygen (although increasing inspired oxygen will not reverse cyanosis caused by intracardiac shunting).(3) Injection of morphine subcutaneously in a dose not in excess of 0.2 mg/kg. Calming and holding the infant in a knee-chest position may abort progression of an early spell. β-Adrenergic blockade by the intravenous administration of propranolol is also useful.

DIAGNOSIS

The cardiac silhouette has been likened to that of a boot or wooden shoe ("coeur en sabot"). The electrocardiogram demonstrates right axis deviation and evidence of right ventricular hypertrophy. Cardiac catheterization demonstrates a systolic pressure in the right ventricle equal to systemic pressure.

COMPLICATIONS

Cerebral thromboses, are common in the presence of extreme Polycythemia and dehydration. Thromboses occur most often in patients younger than 2 yr. These patients may have iron deficiency anemia. Therapy consists of adequate hydration and supportive measures. Brain abscess is less common than cerebral vascular events and extremely rare when most patients are repaired at young ages. Patients with a brain abscess are usually older than 2 yr. Seizures may occur; localized neurologic signs depend on the site and size of the abscess and the presence of increased intracranial pressure. CT or MRI confirms the diagnosis. Antibiotic therapy may help keep the infection localized, but surgical drainage of the abscess is usually necessary.

Bacterial endocarditis may occur in the right ventricular infundibulum or on the pulmonic, aortic, or, rarely, tricuspid valves. Antibiotic prophylaxis is essential before and after dental and certain surgical procedures. Heart failure is not a usual feature in patients with the tetralogy of Fallot. It may occur in a young infant with "pink" or acyanotic tetralogy of Fallot. As the degree of pulmonary obstruction worsens with age, the symptoms of heart failure resolve and eventually the patient experiences cyanosis, often by 6–12 mo of age.