NOTES
CYANOTIC DEFECTS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- *Heart defects with cyanotic presentation:* blue discoloration of skin/mucous membranes, typically seen at fingertips, lips, extremities
- Develop *in utero*
- Persistent truncus arteriosus, hypoplastic left heart syndrome, transposition of great vessels can lead to heart failure
- Persistent truncus arteriosus, tetralogy of Fallot can lead to Eisenmenger syndrome

SIGNS & SYMPTOMS

- Cyanosis
- See individual disorders

DIAGNOSIS

DIAGNOSTIC IMAGING

- Prenatal ultrasound
- Echocardiography
- Chest X-ray

OTHER DIAGNOSTICS

- ECG

TREATMENT

MEDICATIONS

- See individual disorders

SURGERY

- Definitive treatment

OTHER INTERVENTIONS

- Lifestyle changes

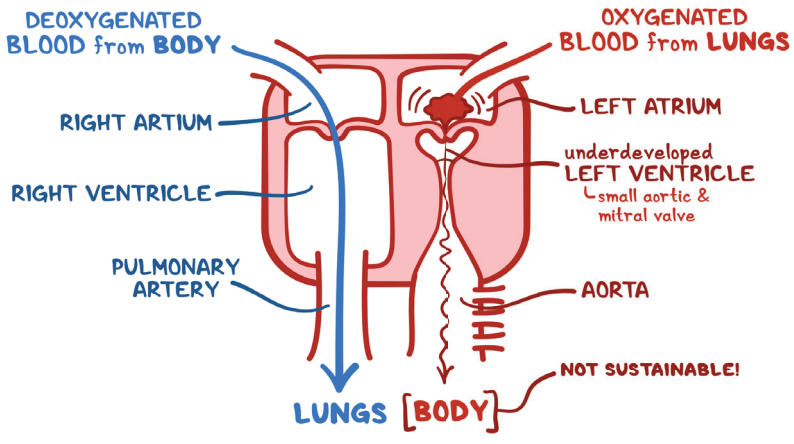

Figure 8.1 Illustration depicting blood flow in hypoplastic left heart syndrome.

HYPOPLASTIC LEFT HEART SYNDROME

osms.it/hypoplastic-left-heart-syndrome

PATHOLOGY & CAUSES

- Congenital underdevelopment of left heart

CAUSES

- *Unknown:* primary congenital heart defect may reduce flow through left ventricle/left outflow tract, affect other heart malformations
- Underdeveloped left ventricle, ascending aorta
  - Aortic/mitral valves may also be affected, narrow, or absent (atresia)
- *If untreated:* left-sided heart failure → cardiogenic shock → death

Atrial septal defect (ASD) and Patent ductus arteriosus (PDA)

- ASD/PDA required for post-natal survival in hypoplastic left heart syndrome
- *With ASD, PDA:* right heart function present but impaired; sometimes asymptomatic at birth
  - Oxygenated blood in left atrium flows into right atrium through ASD → pulmonary artery → PDA → aorta → body
  - *Within one day:* ductus arteriosus begins closing → cyanosis
- *Without ASD, PDA:* heart not capable of sustaining life outside womb
  - Right heart functions normally → oxygenated blood enters left atrium → flow backs up due to small mitral valve, small left ventricle → high pressure in left atrium, blood circulated ineffectively by left ventricle

SIGNS & SYMPTOMS

- Respiratory distress, poor feeding/failure to thrive, left-sided heart failure

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

Chest X-ray

- Cardiomegaly

OTHER DIAGNOSTICS

ECG

- Right ventricular hypertrophy
- After birth

TREATMENT

MEDICATIONS

- Prostaglandin E1 keeps ductus arteriosus open until surgery can be performed

SURGERY

- Surgical repair/heart transplant based on complexity

PERSISTENT TRUNCUS ARTERIOSUS

osms.it/truncus_arteriosus

PATHOLOGY & CAUSES

- Truncus arteriosus fails to divide into aorta/pulmonary artery
- Single giant artery branching off from right, left ventricles which splits into aorta, pulmonary artery
- Oxygenated, deoxygenated blood mix
- Deoxygenated blood mixes into systemic circulation → cyanosis

CAUSES

- Associated with DiGeorge syndrome/22q11.2 deletion syndrome (abnormal tissue formation during development)
- Before birth, deoxygenated blood sent to mother, oxygenated blood arrives from mother
  - Fetal heart sends blood through foramen ovale
  - Oxygenated, deoxygenated blood mix in truncus arteriosus. Both circulations get same amount of oxygenated, deoxygenated blood
  - Otherwise normal fetal development
- After birth, the baby relies on own lungs → foramen ovale closes
  - Deoxygenated, oxygenated blood still mixed → cyanosis
  - Excess blood shunted to pulmonary circuit, as pressure in pulmonary circuit is less than pressure of systemic circuit

RISK FACTORS

- Combination of genes, maternal environment
- Smoking, excessive alcohol intake, teratogenic medications during pregnancy; gestational diabetes; viral illness during pregnancy (e.g. German measles);

associated with genetic disorders (DiGeorge syndrome)

COMPLICATIONS

- Cardiomegaly
- Pulmonary hypertension, can progress to permanent lung damage
- Respiratory problems
- Arrhythmia
- Valve regurgitation

SIGNS & SYMPTOMS

- Difficulty breathing, pounding heart, weak pulse, poor feeding/failure to thrive, lethargy
- With physical exertion (severity varies)
  - Dizziness, fatigue, palpitations, dyspnea
- Impaired growth
- Auscultation
  - Loud systolic murmur along left sternal border due to increased flow through mitral valve
  - Constant ejection click before S2 (closure of aortic, pulmonic valves)
  - Diastolic flow murmur at apex when pulmonary blood flow increases

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Shows heart size, lung abnormalities, possible presence of excess fluid in lungs

Echocardiogram

- Single large vessel arising from left, right ventricles
- Abnormalities of valves between large

vessel and ventricle it arises from
- May show abnormal blood movement between right, left ventricle, and volume of blood flow to lungs

OTHER DIAGNOSTICS

- Newborn pulse oximetry screening
  - Low oxygen saturation
  - Diagnose before symptoms develop

ECG

- Atrial enlargement (notching of P waves/P mitrale)
- Ventricular hypertrophy
- Abnormal T waves
- Right axis deviation

TREATMENT

- In rare cases, babies may survive into adulthood without surgical repair

MEDICATIONS

Diuretics

- Gets rid of excess fluid (e.g. chlorothiazide)

Inotropic agents

- Strengthens cardiac contractions (e.g. Digoxin, treats congestive heart failure, slows down heart rate, increases force of contractions)

Prophylaxis

- Antibiotics during dental/other surgical procedures to avoid infections

SURGERY

- *Goal:* restore normal blood flow through heart
- Procedures vary depending on individual anatomy
  - Close hole between right/left ventricles
  - Separate large vessel into pulmonary artery, aorta
  - Reconstruct single large vessel into new, complete aorta
  - Implant new tube, valve to connect right ventricle with upper part of pulmonary artery, creating new, complete pulmonary artery

OTHER INTERVENTIONS

- *Lifestyle:* possible limitation on intense physical activity
- Lifelong monitoring

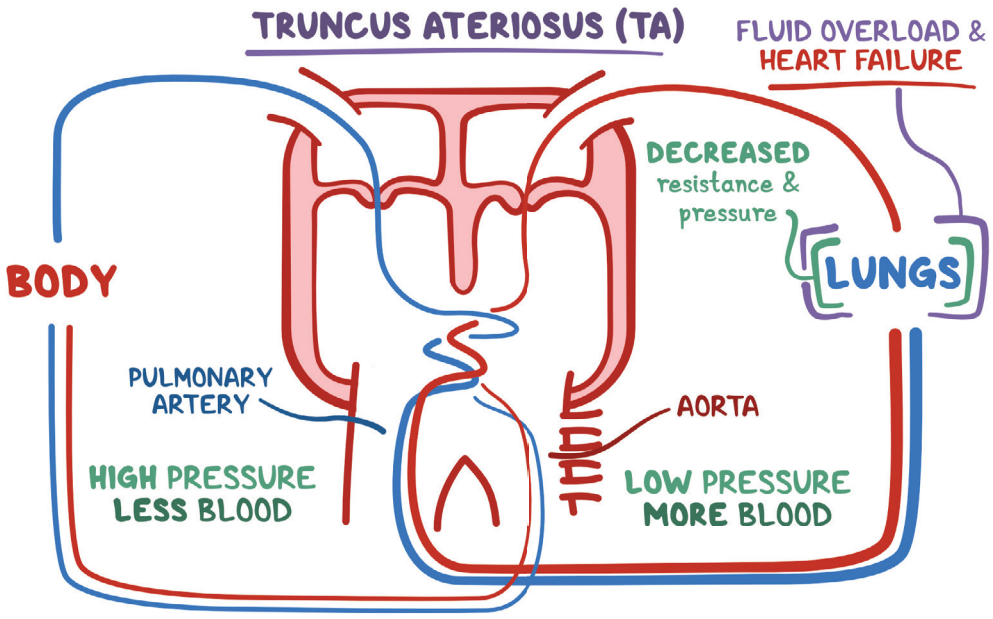

Figure 8.2 Illustration depicting blood flow through the heart in persistent truncus arteriosus.

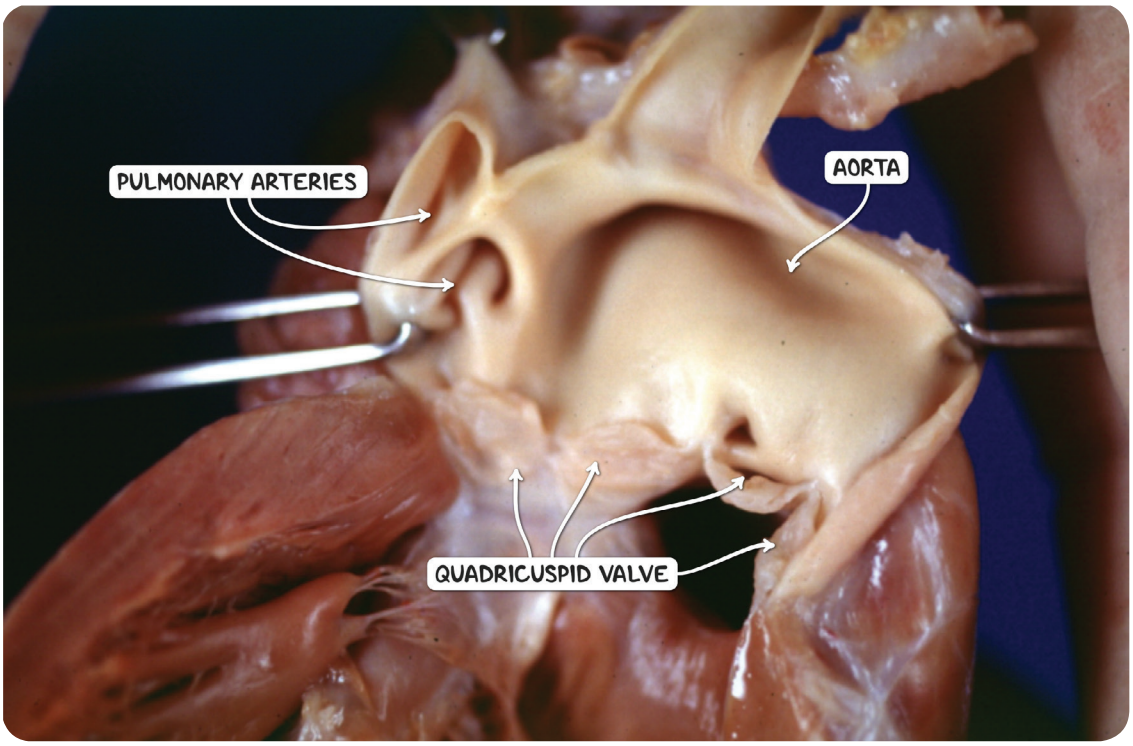

Figure 8.3 Gross pathology of a persistent truncus arteriosus. Both the left and right ventricles pump blood to both the aorta and pulmonary artery through a quadricuspid truncus valve.

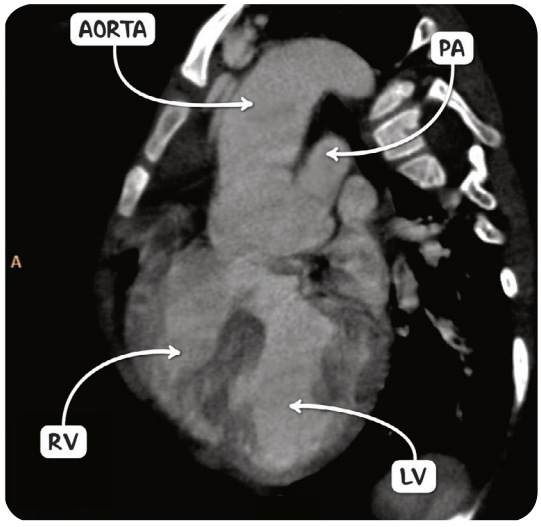

Figure 8.4 Gross pathology of a persistent truncus arteriosus. Both the left and right ventricles pump blood to both the aorta and pulmonary artery through a quadricuspid truncus valve.

TETRALOGY OF FALLOT

osms.it/tetralogy-of-fallot

PATHOLOGY & CAUSES

- Combination of four congenital heart abnormalities
- *Right ventricular outflow tract stenosis (pulmonic stenosis):* obstructs pulmonary circulation
- *Right ventricular hypertrophy:* compensates for right ventricular outflow tract stenosis
- *Ventricular septal defect (VSD):* hole in wall between ventricles. High pressure in right ventricle → blood shunts from right to left → deoxygenated blood to body
- *Aorta overrides ventricular septal defect:* aorta in abnormal position. Variable presentations

CAUSES

- Arises during cardiovascular development
- Most common cause of cyanotic congenital heart defects
- Four abnormalities together cause
  - Mixing oxygenated, deoxygenated blood
  - Narrowed vessels/valves that increase cardiac workload
- Severity of stenosis affects blood flow, changing pressure differentials
  - *Mild stenosis:* left-right shunt → oxygenated blood simply goes through pulmonary circulation again
  - *Severe stenosis:* right-left shunt → deoxygenated blood enters body circulation → less oxygen to tissues
- *Leads to pulmonic regurgitation:* blood flows backwards into RV, right heart overloads, can cause right-sided heart failure
- Associated with alcohol exposure in utero, maternal age 40+ years, poor nutrition or viral illness during pregnancy (e.g. rubella), Down syndrome or DiGeorge syndrome, positive family history of tetralogy of Fallot

SIGNS & SYMPTOMS

- Depend upon severity of stenosis
- Less severe right ventricular outflow obstruction often asymptomatic
- Cyanosis around lips, fingernails ("blue baby syndrome")
- Poor feeding/failure to thrive
- Harsh holosystolic murmur at left upper sternal border → sounds like pulmonary stenosis
  - Right ventricular heave
- Older infants, children
  - Clubbed fingers, toes after a few months
  - Exertional dyspnea
  - *Hypercyanotic episode (tet spell):* on exertion, infant's oxygen demands increase → sudden decrease in oxygen saturation → cyanosis

DIAGNOSIS

DIAGNOSTIC IMAGING

Echocardiography

- Pre/postnatal

Chest X-ray

- "Boot-shaped" heart

MNEMONIC: PROVe

Components of tetralogy of Fallot

Pulmonary infundibular stenosis

Right ventricle hypertrophy

Overriding aorta

Ventricular septal defect

OTHER DIAGNOSTICS

ECG

- Right ventricular hypertrophy, right atrial enlargement

TREATMENT

MEDICATIONS

Prostaglandin E1 analogs (alprostadil)

- Severe cases
- Keep ductus arteriosus open → improve cyanosis until surgery

OTHER INTERVENTIONS

- Treat tet spell
  - *Infants squat to reduce cyanosis:* kinks femoral arteries → increases vascular resistance → increases systemic pressure → increases pressure in left ventricle to greater than pressure in right ventricle → reverse shunt to left-right → resolve cyanosis

SURGERY

- Cardiac repair surgery (first year of life)
  - VSD patch closure (only oxygenated blood flows from left ventricle into aorta)
  - Right ventricular outflow tract enlarged

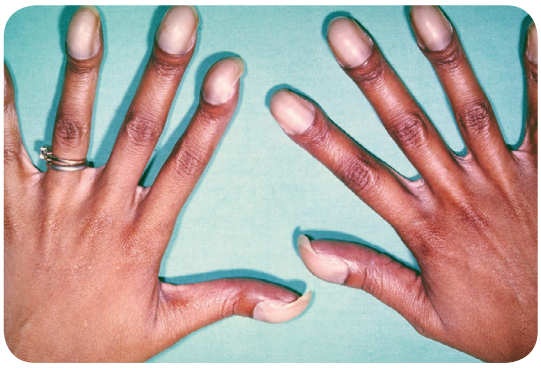

Figure 8.5 Digital clubbing in an adult with tetralogy of Fallot.

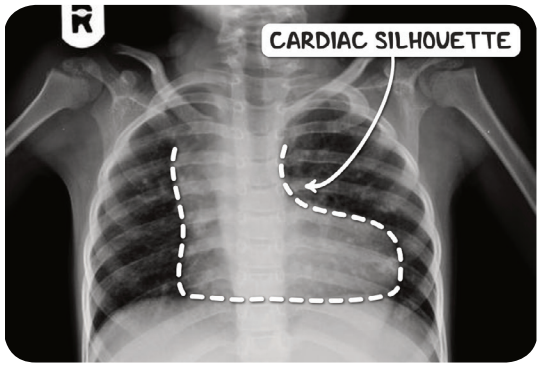

Figure 8.6 A chest radiograph of an infant demonstrating the classic boot-shaped heart seen in tetralogy of Fallot.

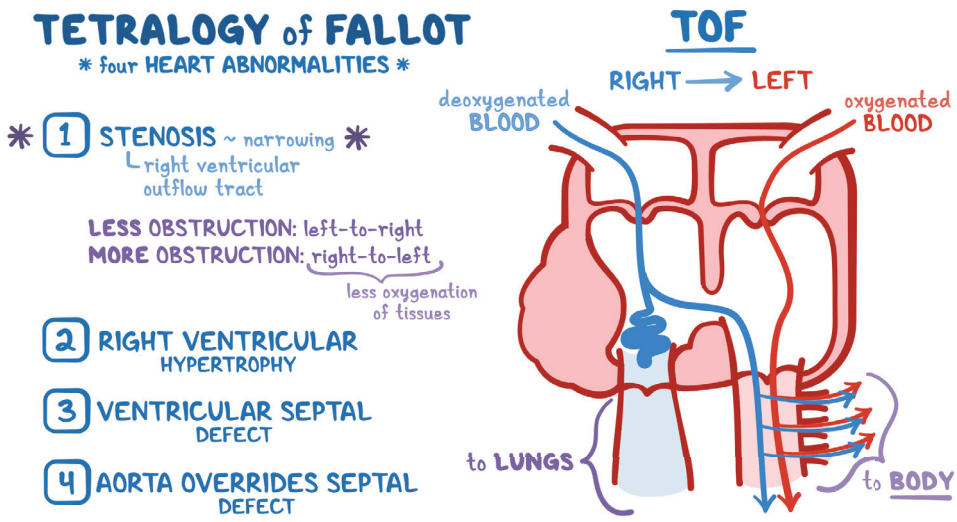

Figure 8.7 Illustration depicting blood flow through a heart with Tetralogy of Fallot.

TOTAL ANOMALOUS PULMONARY VENOUS RETURN

osms.it/anomalous-pulmonary-venous

PATHOLOGY & CAUSES

- Congenital heart defect characterized by anomalous connection of the pulmonary veins and the heart
- Occurs during first eight weeks of fetal development; cause unknown

TYPES

Supracardiac variant

- Most common
- Pulmonary veins open into brachiocephalic veins/superior vena cava (SVC)

Cardiac variant

- Pulmonary veins open into coronary sinus/right atrium

Infradiaphragmatic variant

- Pulmonary veins open into portal/hepatic veins

Mixed variant

- Oxygenated blood travels through pulmonary veins to right atrium/veins → blood does not leave pulmonary circulation → no systemic circulation
- Incompatible with life unless foramen ovale/patent ductus arteriosus present → oxygenated and deoxygenated blood mix → established connection between pulmonary and systemic circulation
- Anomalous connections often accompanied by pulmonary vein obstruction → pulmonary venous hypertension, severe cyanosis

COMPLICATIONS

- Recurrent pulmonary vein stenosis

SIGNS & SYMPTOMS

- Severity of symptoms depend upon presence/degree of obstruction
- Cyanosis, tachypnea, tachycardia, dyspnea, failure to thrive, recurrent respiratory infections
- *In case of infradiaphragmatic variant:* liver enlargement

DIAGNOSIS

DIAGNOSTIC IMAGING

Chest X-ray

- *Snowman sign (figure of 8):* dilated SVC, pulmonary vein, brachiocephalic artery formshead; dilated right atrium forms snowman's body

Echocardiography

- Right ventricular and pulmonary artery volume loading
- Might show left atrium with no connecting veins

LAB RESULTS

- *Assess oxygenation and acid-base status:* decreased values

OTHER DIAGNOSTICS

ECG

- Right ventricular hypertrophy

Auscultation

- Systolic ejection murmur
- Increased pulmonary component of S2
- Split S2
- S3 gallop

TREATMENT

SURGERY

- Surgery to establish blood flow from the right atrium to left atrium
    - If present, pulmonary venous obstruction must be identified and treated promptly

OTHER INTERVENTIONS

- Cardiac catheterization

TRANSPOSITION OF THE GREAT VESSELS (TGA)

osms.it/transposition_of_great_vessels

PATHOLOGY & CAUSES

- Abnormal development causes aorta to arise from right ventricle, pulmonary artery to arise from left ventricle
- Transposition creates two small circuits of blood flow rather than one large
    - *Right side:* right ventricle → aorta → body → right atrium → right ventricle (blood never oxygenated)
    - *Left side:* left ventricle → pulmonary artery → lungs → pulmonary veins → left atrium → left ventricle (blood never deoxygenated)
- After birth → lungs used for oxygen → foramen ovale, ductus arteriosus close → no exchange between two circuits → cyanosis, death
- Sometimes, foramen ovale or ductus arteriosus stay open, or baby has ventricular septal defect (VSD); allows blood to circulate
- Different levels of severity of transposition of the great arteries (TGA)
    - *d-TGA:* dextro-TGA/complete TGA (*dextro* = aorta on right)
    - *l-TGA:* levo-TGA/congenitally corrected TGA (*levo* = aorta on left). Ventricles, valves switched. Great vessels in normal orientation, but connected to wrong ventricle. Normal blood flow circuits preserved

RISK FACTORS

- *During pregnancy:* diabetes, rubella, poor nutrition, consumption of alcohol, > 40 years old

SIGNS & SYMPTOMS

- *In utero:* asymptomatic
- *d-TGA:*
    - Cyanosis, unchanged with supplemental oxygen (less severe if VSD present)
    - Tachypnea
    - Acidosis
- *l-TGA:*
    - Asymptomatic

DIAGNOSIS

DIAGNOSTIC IMAGING

Echocardiogram

- Evaluate heart function, structure

Chest X-ray

- Classic triad
    - Heart appears as egg on its side/"egg on a string" appearance
    - Lung congestion
    - Cardiomegaly

Angiogram

- Pre-surgery

TREATMENT

MEDICATIONS

- *Prostaglandin E:* short-term solution. Keeps ductus arteriosus open

SURGERY

- *Balloon atrial septostomy:* short-term solution. Hole created in atrial septum
- Surgically switch great vessels
  - Five year survival rate > 80%
  - *No treatment:* one year survival rate 10%

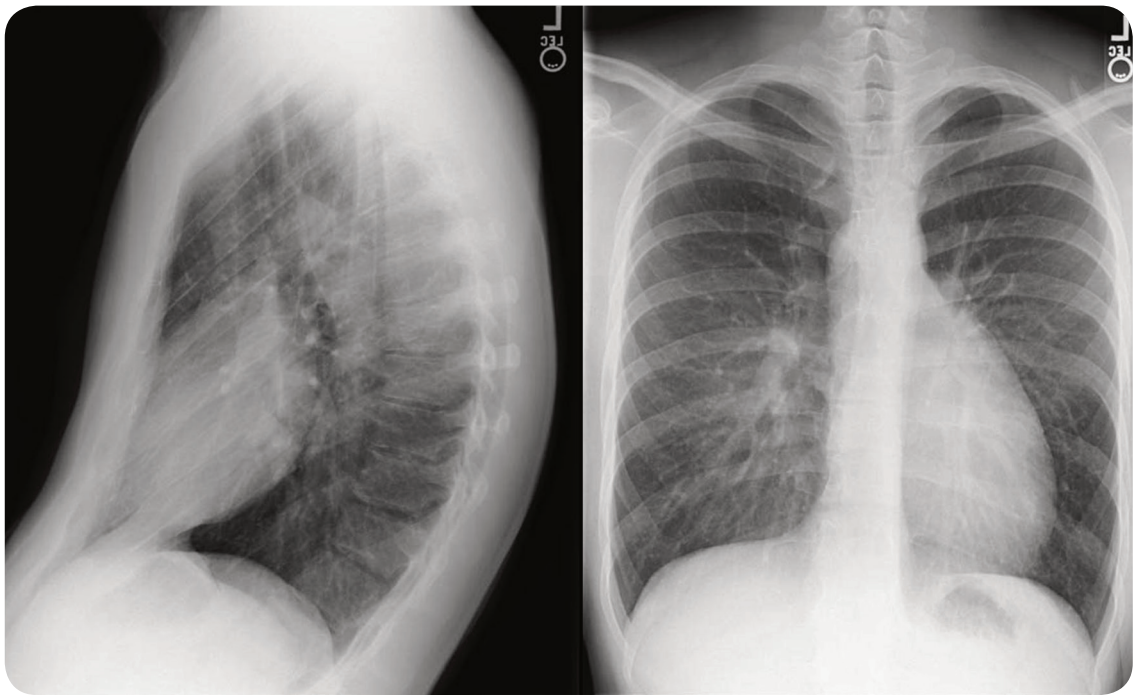

Figure 8.8 Chest radiograph in both a lateral (L) and frontal (R) view, demonstrating the "egg on a string" sign of transposition of the great vessels.

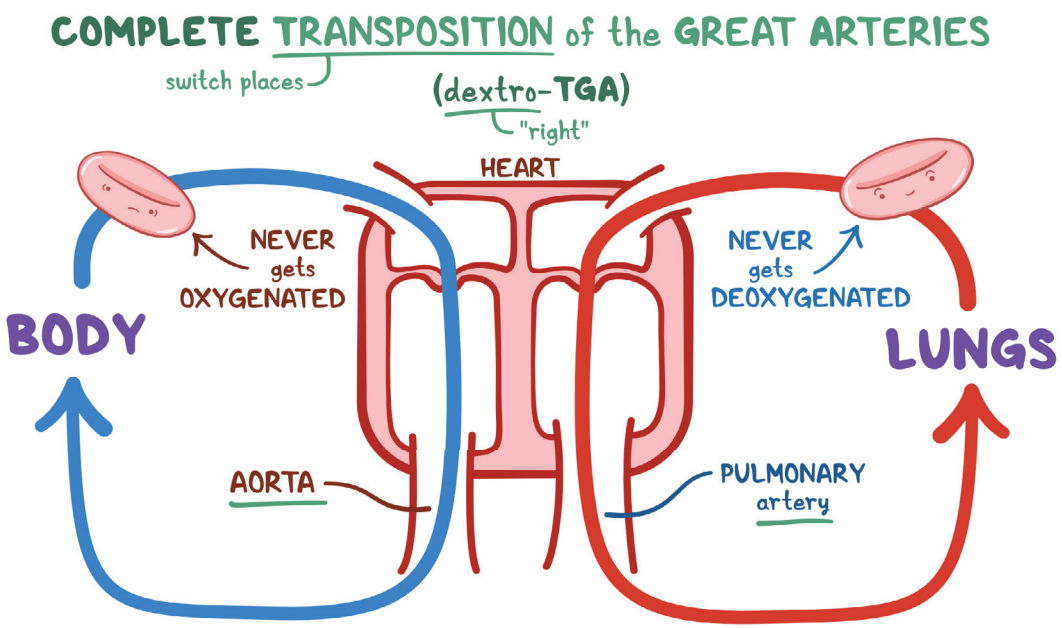

Figure 8.9 Illustration depicting blood flow through a heart with dextro transposition of the great arteries.

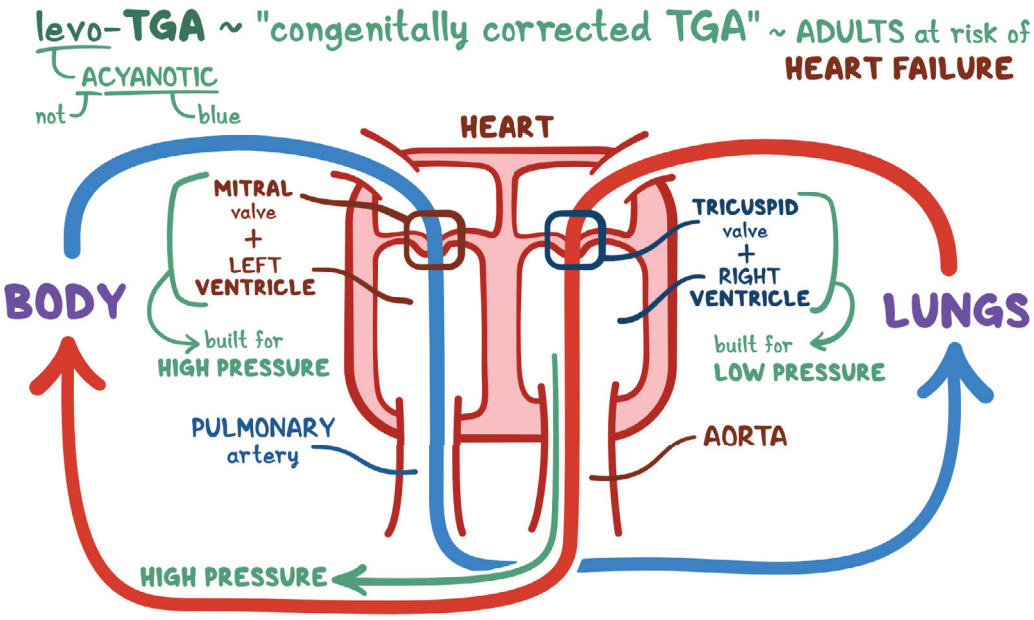

Figure 8.10 Illustration depicting blood flow through a heart with levo transposition of the great arteries.

CYANOTIC HEART DEFECT OVERVIEW

	MALFORMATION	KEY DIAGNOSTIC CLUES
HYPOPLASTIC LEFT HEART SYNDROME	Underdeveloped left side of the heart; associated with ASD and PDA for survival	CXR: Cardiomegaly ECG: Right ventricular hypertrophy Prostaglandin E required until surgery
PERSISTENT TRUNCUS ARTERIOSUS	No division causes one giant artery; associated with DiGeorge syndrome	Auscultate: Systolic murmur, ejection click Pulse Oximetry: Low ECG: Atrial enlargement, ventricular hypertrophy, right axis deviation
TETRALOGY OF FALLOT	Pulmonic stenosis, right ventricular hypertrophy, overriding aorta, VSD	Auscultate: Harsh holosystolic murmur LUSB CXR: "Boot-shaped" heart Exertional cyanosis
TRANSPOSITION OF THE GREAT VESSELS	Aorta arises from right ventricle; pulmonary artery from left ventricle	Cyanosis persists with oxygen supplementation CXR: "Egg on a string"Prostaglandin E required until surgery
TOTAL ANOMALOUS PULMONARY VENOUS RETURN	Pulmonary veins do not connect properly to heart; associated with patent foramen ovale or PDA for survival	Present