

Acyanotic Heart Disease:

Summary

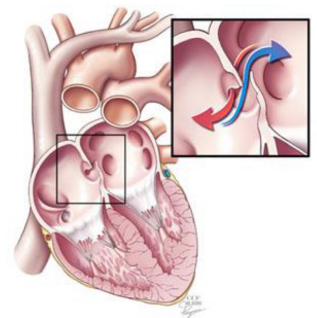
Acyanotic congenital heart defects (ACHDs) are cardiac malformations that affect the atrial or ventricular walls, heart valves, or large blood vessels. Common causes include genetic defects (e.g., trisomies), maternal infections (e.g., rubella), and maternal use of drugs or alcohol during pregnancy. ACHDs are characterized by a left-to-right shunt, which causes pulmonary hypertension and right atrial and ventricular hypertrophy. Symptoms depend on the extent of the malformation and the resulting impairment of cardiac function. Infants may be asymptomatic or present with signs of respiratory distress, failure to thrive, and/or symptoms of heart failure. Characteristic heart murmurs are important clues for establishing the diagnosis, which is typically confirmed by visualizing the defect on transthoracic echocardiography (TTE). Other studies (e.g., chest x-ray, MRI, cardiac CT, or cardiac catheterization) may be required for surgical evaluation and planning. Surgical or transcatheter repair is indicated in selected patients and pharmacological treatment is required to manage complications, e.g., heart failure, arrhythmias, and Eisenmenger syndrome.

FEEDBACK

First: Atrial septal defect:

1. Classification

- Ostium primum.** This type of ASD is a defect in the lower portion of the atrial septum. A cleft, or division, in the anterior mitral valve leaflet is almost always present and may cause mitral regurgitation. Ostium primum ASD is a common congenital heart lesion in **Down syndrome**.
- Ostium secundum.** This type of ASD is a defect in the middle portion of the atrial septum. Ostium secundum is the **most common type** of ASD.
- Sinus venosus.** This type of ASD is a defect superiorly and posteriorly in the septum near the junction of the right atrium and superior vena cava (SVC). In sinus venosus ASD, the right pulmonary veins usually drain anomalously into the right atrium or SVC instead of draining into the left atrium.



- Pathophysiology.** Blood flows across the ASD **from the left atrium to the right atrium** (i.e., **left-to-right shunt**). The direction of the blood flow is determined by the relative diastolic compliances of the right and left ventricles (which in turn are determined by systemic and pulmonary vascular resistances [SVR and PVR]). **Blood therefore flows from areas of higher resistance to areas of lower resistance.** Increased blood flow across the ASD leads to an increase in size of the right atrium and right ventricle and to increased pulmonary blood flow.

- ASD → oxygenated blood shunting from LA to RA → ↑ O₂ saturation in the RA → ↑ O₂ saturation in RV and pulmonary artery
- In more severe defects, the shunts may lead to supraventricular arrhythmias, pulmonary hypertension, and/or Eisenmenger syndrome.

3. Clinical features

- Symptoms are minimal**, if any, except in patients with an ostium primum defect who develop mitral regurgitation that results in CHF.
- Physical examination findings** include the following:
 - Increased right ventricular impulse** (manifesting as a stronger point of maximal impulse) as a result of right ventricular volume and pressure overload.
 - Systolic ejection murmur** (from excessive pulmonary blood flow) best heard at the **mid and upper left sternal borders**. A mid diastolic filling rumble representing increased blood flow through the tricuspid valve may also be heard.
 - Fixed-split second heart sound.** With blood shunting across the ASD from the left atrium to the right atrium to the right ventricle, the **second heart sound is widely split** because of the increased volume of blood that must be ejected from the right ventricle, leading to later closure of the pulmonic valve compared with the aortic valve (i.e., a wider split of the A2 and P2 components of the second heart sound). **The splitting is fixed**, and does not vary with respiration, because the blood shunting across the ASD ensures that the right ventricle receives a fixed volume of blood, by balancing the increased volume reaching the right ventricle with inspiration and the decreased volume of blood reaching the right ventricle with expiration (i.e., the normal physiologic variation in timing of aortic and pulmonic valve closure with respiration is absent).

So in auscultation:

Auscultation [3]

- **Mid-systolic ejection murmur** over the second left ICS, parasternal
- **Widely split second heart sound** (S_2) over the second left ICS, which is **fixed** (does not change with respiration)
- Soft mid-diastolic murmur over the lower left sternal border

Diagnostics [10][23][24]

- **Echocardiography** (confirmatory test)
 - TTE with Doppler
 - Confirms interatrial communication
 - Best visualized in the apical four-chamber and subcostal views
 - Agitated saline study: if TTE findings are equivocal [23]
- **ECG**
 - Signs of RV hypertrophy, e.g.:
 - Vertical or right axis deviation
 - P pulmonale, and/or PR prolongation
 - Complete or incomplete right bundle branch block
 - Atrial tachyarrhythmias, e.g., atrial fibrillation, atrial flutter, are common in adults with ASDs. [23]
- **Chest x-ray**
 - Rounding of the left heart contour due to an enlarged right atrium, right ventricle, and pulmonary artery
 - Enlarged lung markings due to enhanced pulmonary vasculature
- **Pulse oximetry**

4. Management. Treatment of primum and sinus venosus ASDs is closure by open heart

surgery to prevent right-sided heart failure, pulmonary hypertension, atrial dysrhythmias, and paradoxical embolism (a stroke caused by a blood clot traveling from the right atrium to the left atrium via the ASD). Most **secundum ASDs** can be **successfully closed using interventional catheterization procedures** with implantation of atrial septal occlusion devices.

ⓘ Up to 40% of ASDs spontaneously close by 5 years of age. [26]

⚠ Surgical repair is contraindicated in patients with right-to-left shunts (e.g., in Eisenmenger syndrome). [10]

Complications [27]

- Paradoxical embolism (↑ risk of ischemic stroke): an embolus from the venous circulation passes from the right atrium through the ASD into the left atrium, enters the arterial circulation into the brain, and causes a stroke
- Heart failure

FEEDBACK

Second: Ventricular Septal Defect:

1. **muscular septum** : largest component.
2. **endocardial cushion**: The inlet or posterior septum.
3. The **subarterial or supracristal septum** comprises conotruncal tissue.
4. The **membranous septum** : below the aortic valve and is relatively small.

1. **Classification.** VSDs are classified by location as **inlet, trabecular (muscular), membranous, and outlet (supracristal)** (Figure 8-3).
2. **Pathophysiology.** After birth, as pulmonary vascular pressure decreases, blood flows across the VSD *from the left ventricle to the right ventricle*, owing to the lower resistance within the pulmonary circulation compared to the resistance within the systemic circulation. However, with time, the pulmonary vessels hypertrophy in response to this increased pulmonary flow. This hypertrophy may lead to increased PVR (pulmonary hypertension). If treated early, the increased PVR is usually reversible. If increased pulmonary blood flow persists, the pulmonary hypertension may become irreversible and even progress to *Eisenmenger syndrome* [see section III.D.3.d.(2)].

- Most commonly located in the **membranous part** of the ventricular septum (*pars membranacea*)
- Defect in ventricular septum → left-to-right shunt with the following consequences:

- RV volume overload → **RV eccentric hypertrophy**
- Excessive pulmonary blood flow → ↑ pulmonary artery pressure → **pulmonary hypertension**
- ↓ Cardiac output
- LV volume overload → **LV eccentric hypertrophy**
- ↑ O₂ saturation in right ventricle and pulmonary artery

- the **most common congenital heart defect**, 25% of all congenital heart disease.
- **Perimembranous VSDs** are the most common of all VSDs (67%).
- prognostically and in approach to repair the location of the VSD is important.
- physiologically, the amount of flow crossing a VSD depends on the size of the defect and the pulmonary vascular resistance.

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- Even large VSDs are not symptomatic at birth because the pulmonary vascular resistance is normally elevated at this time (increase in the pulmonary arterioles media + relative polycythemia).
- As the pulmonary vascular resistance decreases over the **first 6 to 8 weeks of life** → amount of shunt ↑ → symptoms develop
- A left-to-right shunt at the ventricular level has 3 hemodynamic consequences:
 1. increased LV volume load → LV dilatation and then hypertrophy → increase pulmonary venous pressure → **raises** pulmonary capillary pressure → increase pulmonary interstitial fluid → pulmonary edema
 2. excessive pulmonary blood flow
 3. reduced systemic cardiac output
- The natural history has a wide spectrum, ranging from spontaneous closure to congestive heart failure (CHF) to death in early infancy.
- Spontaneous closure frequently occurs in children, usually occurs by the age of 2 years. Closure is uncommon after 4 years of age. Closure is most frequently observed in muscular defects (80%).
- Patients are at risk for infective endocarditis, but small muscular VSDs pose no other adverse possibilities.

NOTE:

The increase in pulmonary venous pressure is not seen with an ASD because LA pressures are low, as blood can readily exit it through the atrial communication

and even progress to Eisenmenger syndrome [see section 11.2.3.3.(2)].

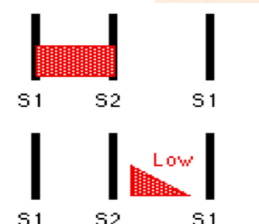
3. **Clinical features and course** vary greatly depending on the magnitude of the left-to-right shunting across the VSD. **The amount of blood flow directed from one side of the heart to the other side (i.e., the shunt) is determined by both the size of the VSD and the degree of PVR.** For example, the larger the VSD and the lower the PVR, the greater the blood flow across the VSD and into the pulmonary vessels. *The greater the pulmonary blood flow, the more symptomatic the patient.* Typical clinical presentations include the following:

- Small VSDs** have little to no shunt across the VSD and **may close spontaneously.** On examination, small VSDs typically have a *high-pitched holosystolic murmur* ranging from a grade 3 to grade 4 murmur. A thrill at the lower left sternal border and a grade 4 high-pitched holosystolic murmur is indicative of a very restrictive defect with a high flow velocity across the VSD. Key point: **As the size of the VSD decreases, the intensity of the murmur increases.**
- Moderate VSDs** may have a large shunt across the VSD that may result in signs and symptoms of CHF. A **holosystolic murmur** is usually present, and its intensity depends on the size of the shunt. If the left-to-right shunt across the VSD is large (2:1 pulmonary-to-systemic flow—i.e., twice as much blood flows to the lungs as to the systemic circulation), then a diastolic murmur of mitral turbulence may also be heard at the apex (mitral filling rumble representing the excess blood from the lungs now passing through the mitral valve).
- Large VSDs** often cause signs and symptoms of CHF. They have less turbulence across the VSD, so the *systolic murmur is shorter and lower in pitch.* A mitral filling rumble may be heard at the apex. The S_2 tends to be loud and more narrowly split than usual, with P_2 (pulmonary closure sound) being accentuated due to high pulmonary artery pressure.
- PVR may eventually become elevated in moderate or large VSDs in response to chronically high pulmonary flow.** When this occurs, clinical features change.
 - When PVR becomes elevated,** the right ventricular impulse is noticeably increased and the second heart sound may be single and loud. The mitral filling rumble disappears because of diminished pulmonary blood flow as a result of decreased left-to-right shunting. Symptoms of CHF also diminish as PVR increases, because of the decrease in pulmonary blood flow.
 - If PVR remains elevated, pulmonary hypertension may become irreversible, even if the VSD is surgically closed.** In the extreme situation in which PVR

increases and exceeds SVR, shunting changes from left-to-right to right-to-left (a condition termed **Eisenmenger syndrome**). The **right-to-left shunt is manifested by cyanosis.**

- ♥ Small VSDs, with little shunt, are often
 1. Asymptomatic
 2. a loud murmur.
- ♥ Moderate - large VSDs result in pulmonary over circulation and CHF, presenting as
 1. fatigue,
 2. diaphoresis with feedings
 3. poor growth.

- The typical physical finding with a VSD is:
 1. **pansystolic murmur** heard best at the lower left sternal border.
 2. **mid-diastolic murmur** at the apex. As larger shunts result in increased flow across the mitral valve.
 3. splitting of S_2 & intensity of P_2 depend on pulmonary artery pressure.



IMAGING STUDIES

- ECG and chest x-ray findings depend on the size of the VSD.
 - ♥ Small VSDs may have normal studies.
 - ♥ Larger VSDs cause volume overload to the left side of the heart resulting in ECG findings of left atrial and ventricular enlargement and hypertrophy.
- A chest radiograph may reveal:
 - ♥ cardiomegaly, enlargement of the left ventricle
 - ♥ Increase in the pulmonary artery silhouette and increased pulmonary blood flow.
 - ♥ **Increased flow** or **increased pulmonary vascular resistance** → Pulmonary hypertension → right ventricular enlargement and hypertrophy.

S1 S2

TREATMENT

- 35% of all VSDs close spontaneously.
- Small VSDs usually close spontaneously.
- if they do not close:
 - ♥ surgical closure may not be required
 - ♥ **prophylactic antibiotics** are needed to prevent subacute bacterial endocarditis.
- moderate to large VSDs includes :
 - ♥ Initial treatment of **diuretics** and **digoxin**
 - ♥ closure of the defect if poor growth or pulmonary hypertension despite therapy
- Most VSDs are closed in **surgery**, but some VSDs, especially muscular defects, can be closed with **devices** placed at cardiac catheterization.

3rd: Patent Ductus Arteriosus

1. **Definition.** In the fetus, the ductus arteriosus connects the pulmonary artery to the aorta. After birth, as the PaO_2 rises, the ductus arteriosus normally fibroses. If it remains open, it is termed a **PDA**. The incidence of PDA is especially high in preterm infants.
2. **Postnatal pathophysiology.** In healthy infants the SVR exceeds PVR, and blood therefore flows through the PDA from the aorta to the pulmonary artery (left-to-right shunt). This leads to increased pulmonary blood flow (Figure 8-4).

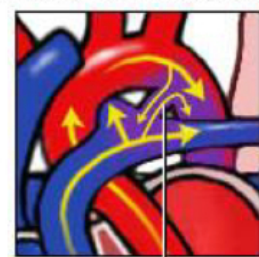
ETIOLOGY AND EPIDEMIOLOGY

- The ductus arteriosus allows blood to flow from the pulmonary artery to the aorta during fetal life.
- Failure of the normal closure of this vessel results in a PDA .
- With a falling pulmonary vascular resistance after birth, left-to-right shunting of blood and increased pulmonary blood flow occur.
- Excluding premature infants, PDAs = 5%-10% of congenital heart disease.

CLINICAL MANIFESTATIONS

- Symptoms depend on the amount of extra blood flow to the lungs.
- The magnitude of the shunt, which can be similar to a VSD, depends on:
 1. the size of the PDA (including diameter, length, and tortuosity)
 2. pulmonary vascular resistance
- Patients with small PDAs are asymptomatic.
- Moderate to larger shunts produce the symptoms of CHF as the pulmonary vascular resistance decreases over the first 6 to 8 weeks of life.
- The physical examination depends on the size of the shunt.
 1. **widened pulse pressure** is often present due to the runoff of blood into the pulmonary circulation during diastole.
 2. **continuous machine-like murmur** can be heard at the left infraclavicular area, and a thrill may be palpable. radiates along the pulmonary arteries and is often well heard over the left back.
 3. Larger shunts with increased flow across the mitral valve may result in a mid-diastolic murmur at the apex and a **hyperdynamic precordium**.
 4. Splitting of S_2 and the intensity of the P_2 depend on the pulmonary artery pressure. ↑ pulmonary pressures → ↑ intensity of P_2 .

Abnormal circulation



Patent ductus arteriosus

TREATMENT

- Spontaneous closure of a PDA after a few weeks of age is **uncommon** in full-term infants.
- Moderate and large PDAs may be managed initially with **diuretics** and **digoxin** but eventually requires closure.
- Closure of small PDAs also is recommended because of the **risk of subacute bacterial endocarditis**.
- Most PDAs can be closed in the catheterization laboratory by either **coil embolization** or a **PDA closure device**.

Indomethacin is often used in premature infants to close a PDA medically.

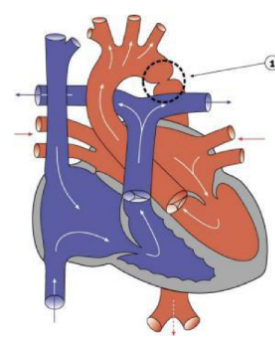
PDAs in the nonneonate are usually closed surgically by transcatheter insertion of a device.

4th: Coarctation of the aorta

1. **Definition.** Coarctation of the aorta is narrowing of the aortic arch, just below the origin of the left subclavian artery and typically at, or just proximal to, the origin of the ductus arteriosus. It may be a discrete narrowing (hourglass) or a long-segment obstruction.
2. **Pathophysiology.** The narrowed segment obstructs or diminishes flow from the proximal to the distal aorta (**Figure 8-5**).

ETIOLOGY AND EPIDEMIOLOGY

- Coarctation of the aorta occurs in 10% of all congenital heart defects.
- It is almost always **juxtaductal** in position.
- During development of the aortic arch, the area near the insertion of the ductus arteriosus fails to develop correctly, resulting in a narrowing of the aortic lumen.



Clinical features. Signs and symptoms depend on the severity of the obstruction and on the presence of any other associated cardiac abnormalities.

- a. **Neonates or infants with severe coarctation** may **depend on a right-to-left shunt through the PDA** for perfusion of the lower thoracic and descending aorta. Such

infants may be **minimally symptomatic initially**, but **as the PDA closes, symptoms of CHF develop and progress**.

1. Blood pressure may be elevated in the upper extremities and low in the lower extremities before the onset of CHF (before the PDA becomes narrower or closes).
 2. Once the neonate or infant develops CHF, pulses in all four extremities are poor due to low cardiac output.
- b. **Older children or adolescents** may have no symptoms and may have only hypertension or a heart murmur. **Hypertension** is typically noted in the **right arm**, and commonly in the left arm as well, with *reduced blood pressure in the lower extremities*.
 1. **Femoral pulse**, which normally precedes the radial pulse, is **dampened and delayed** until after the radial pulse (**radiofemoral delay**).
 2. **Blood pressure and pulse findings** may be less prominent if collateral vessels (intercostal arteries) develop, which allow the ascending aortic pressure and flow to circumvent the coarctation.
 3. **Bicuspid aortic valve or aortic stenosis** is present in 50% of patients with coarctation of the aorta. If either of these conditions is present, a systolic murmur of aortic stenosis may be heard [see [section III.G.3](#)].
 4. **Bruit of turbulence** through the coarctation may be audible at the left upper back near the left scapula.



IMAGING STUDIES

- The ECG and chest x-ray show evidence of right ventricular hypertrophy in infantile coarctation with marked **cardiomegaly** and **pulmonary edema**.
- In older children, the ECG and chest x-ray usually show left ventricular hypertrophy and a mildly enlarged heart.
- **Rib notching** may also be seen in older children (>8 years old) with large collaterals.
- **Echocardiography** shows:
 1. the site and degree of coarctation
 2. presence of left ventricular hypertrophy
 3. aortic valve morphology and function
 4. associated lesion

Management

- a. **Initial management** in the **symptomatic neonate** is directed at improving circulation to the lower body.
 1. **Intravenous prostaglandin E (PGE)** is given urgently to open the ductus arteriosus.
 2. **Inotropic medications** are given to overcome myocardial depression, and low-dose dopamine is used to maximize renal perfusion and function.
- b. **Corrective repair**
 1. **Surgery** is the usual approach in newborns and infants and involves excision of the narrowed segment followed by end-to-end anastomosis. Late recurrence of narrowing may occur in up to 50% of patients.
 2. **Balloon angioplasty with or without endovascular stent placement** is the treatment of choice for short segment coarctations in school-age children and teens who have not undergone surgical correction. It is also the primary intervention for **recurrent coarctation after surgical correction**.

Prognosis is excellent, but long-term concerns include recurrence of coarctation and upper extremity hypertension with exercise.

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