CONGENITAL HEART DISEASE

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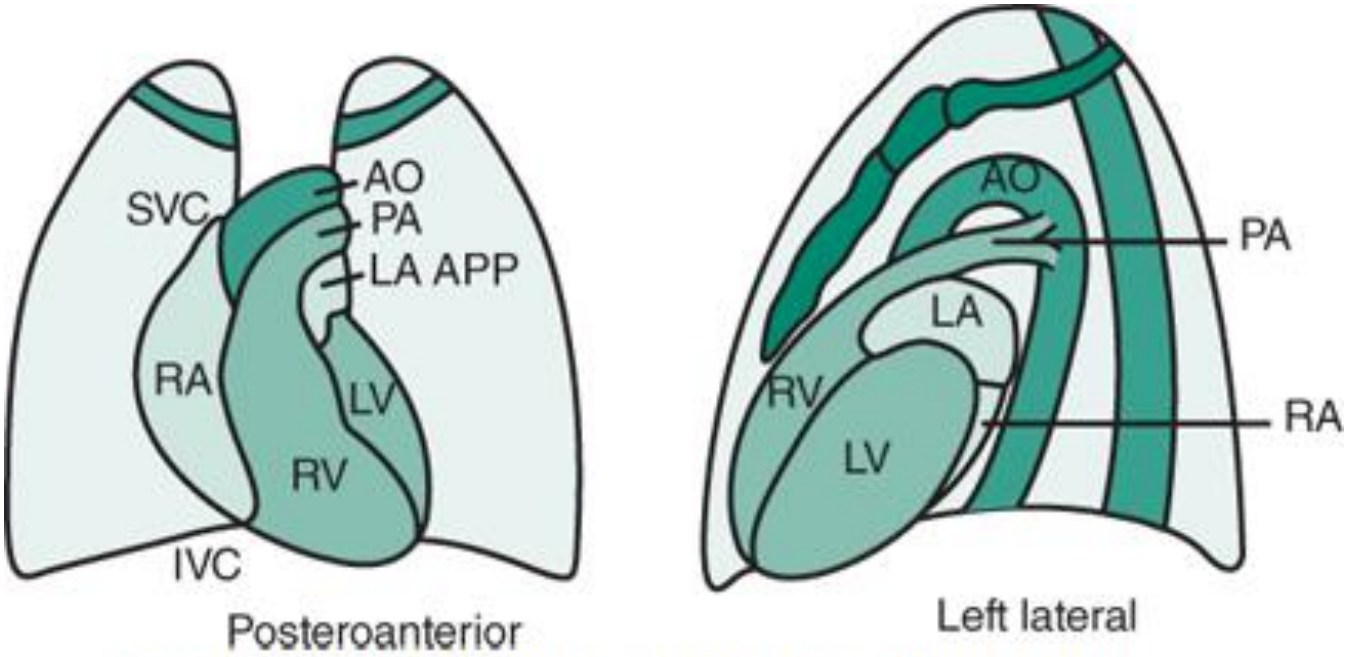
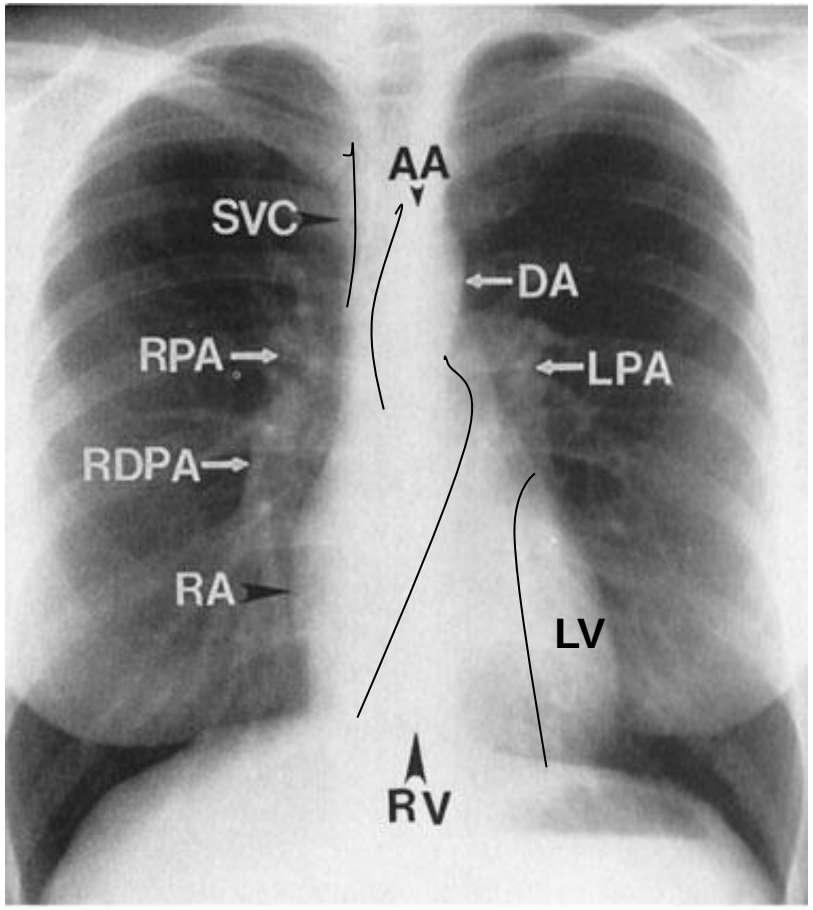


Source: W.W. Hay Jr., M.J. Levin, M.J. Abzug, Maya Bunik: Current Diagnosis & Treatment: Pediatrics, 25th Edition,

Position of cardiovascular structures in principal radiograph views. AO, aorta; IVC, inferior vena cava; LA, left atrium; LA APP, left atrial appendage; LV, left ventricle; PA, pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

Normal chest x-ray



A

Chen, MYM, Pope Jr., TL, Ott DJ: *Basic Radiology*: http://www.accessmedicine.com

A. PA view of normal chest.

RA = right atrium, RDPA = right descending pulmonary artery, RPA = right main pulmonary artery, SVC = superior vena cava, AA = aortic arch, DA = proximal descending thoracic aorta, LPA= left pulmonary artery, RV = right ventricle.

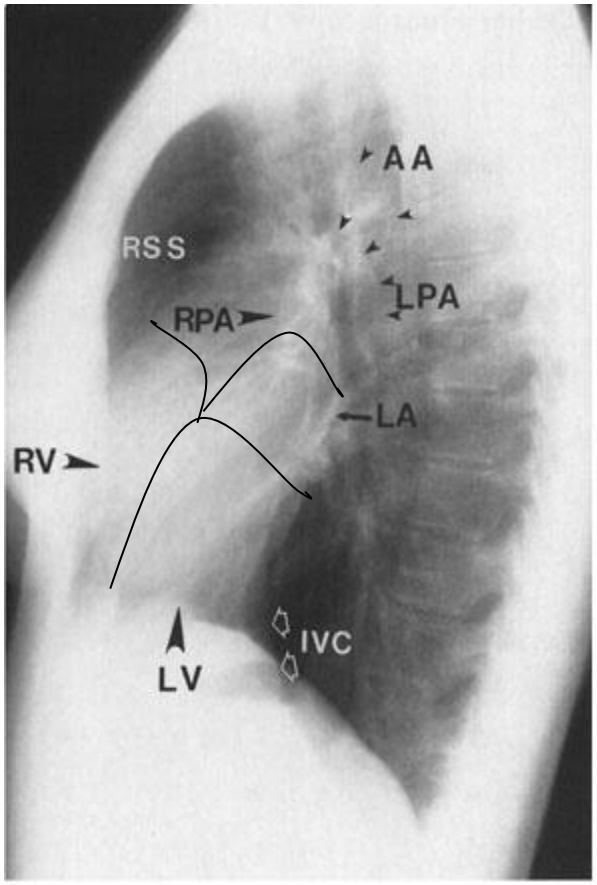
Fig. 3-1 Accessed 08/01/2010
Modified

Normal chest x-ray

B. Lateral view of normal chest.

RV = right ventricle, RSS = retrosternal clear space, AA = ascending aorta, LPA = left pulmonary artery, RPA = right pulmonary artery en face, IVC = inferior vena cava, LA = left atrium, and LV = left ventricle.

Fig. 3-1 Accessed 08/01/2010
Modified



B

Clues on the chest x-ray

- Congested pulmonary vasculature
- Active congestion
- Seen in left-to-right shunts when right ventricular output is approximately 2.5 times that of the left ventricle.
- Vessels are enlarged, are seen more peripherally than normal, and may be tortuous
- Margins remain distinct as there is little interstitial edema.
- Passive congestion
- Elevated pulmonary venous pressure that reflects left cardiac dysfunction or obstruction.

Clues on the chest x-ray

- Decreased pulmonary vasculature
- Represents decreased blood flow through the pulmonary circulation
- Usually as a result of right ventricular outflow obstruction with an associated right-to-left shunt.
- If the proximal pulmonary arteries are enlarged, with pruning of the peripheral vascular markings, then consider pulmonary artery hypertension.
- The aorta may be normal, increased or decreased in size.
- An enlarged aortic knob may be present:
- Post-stenotic dilatation

Clues on the chest x-ray

- Increased blood flow
- ASD
- VSD
- Patent ductus arteriosus (PDA)
- Truncus arteriosus
- Valvular insufficiency
- Severe Tetralogy of Fallot

Clues on the chest x-ray

- Reduced blood flow
- Systemic hypertension
- A small aortic knob usually represents reduced blood flow typically due to atrial septal defect or ventricular septal defect.
- Tetralogy of Fallot
- Pulmonic Stenosis/Pulmonic atresia
- Tricuspid stenosis/Tricuspid atresia
- Coarctation of aorta
- Hypoplastic left ventricle syndrome

Table 20–1.

Symptoms of increased and decreased pulmonary blood flow.

Decreased Pulmonary Blood Flow	Increased Pulmonary Blood Flow
Infant/toddler	
Cyanosis	Tachypnea with activity/feeds
Squatting	Diaphoresis
Loss of consciousness	Poor weight gain
Older child	
Dizziness	Exercise intolerance
Syncope	Dyspnea on exertion, diaphoresis

Current Diagnosis & Treatment: Pediatrics, 25e

Clues on the chest x-ray

- Right sided aortic arches
- Only10% are associated with congenital heart disease
- Tetralogy of Fallot

Clues on the chest x-ray

- A small or inapparent pulmonary artery
- Secondary to congenital hypoplasia or aplasia
- Decreased pulmonary flow as a result of pulmonary outflow obstruction
- As in tetralogy of Fallot
- Abnormally located
- As is in truncus arteriosus or transposition of the great arteries

Clues on the chest x-ray

- Enlarged pulmonary artery:
- Post-stenotic dilatation
- In pulmonary valve stenosis, the left pulmonary artery preferentially dilates due to the orientation of the stenotic jet
- Increased pulmonary blood flow
- Left-to-right shunts
- Pulmonary valvular insufficiency
- Pulmonary arterial hypertension
- Both the right and left pulmonary arteries enlarge
- Distinguishes this from pulmonary valve stenosis

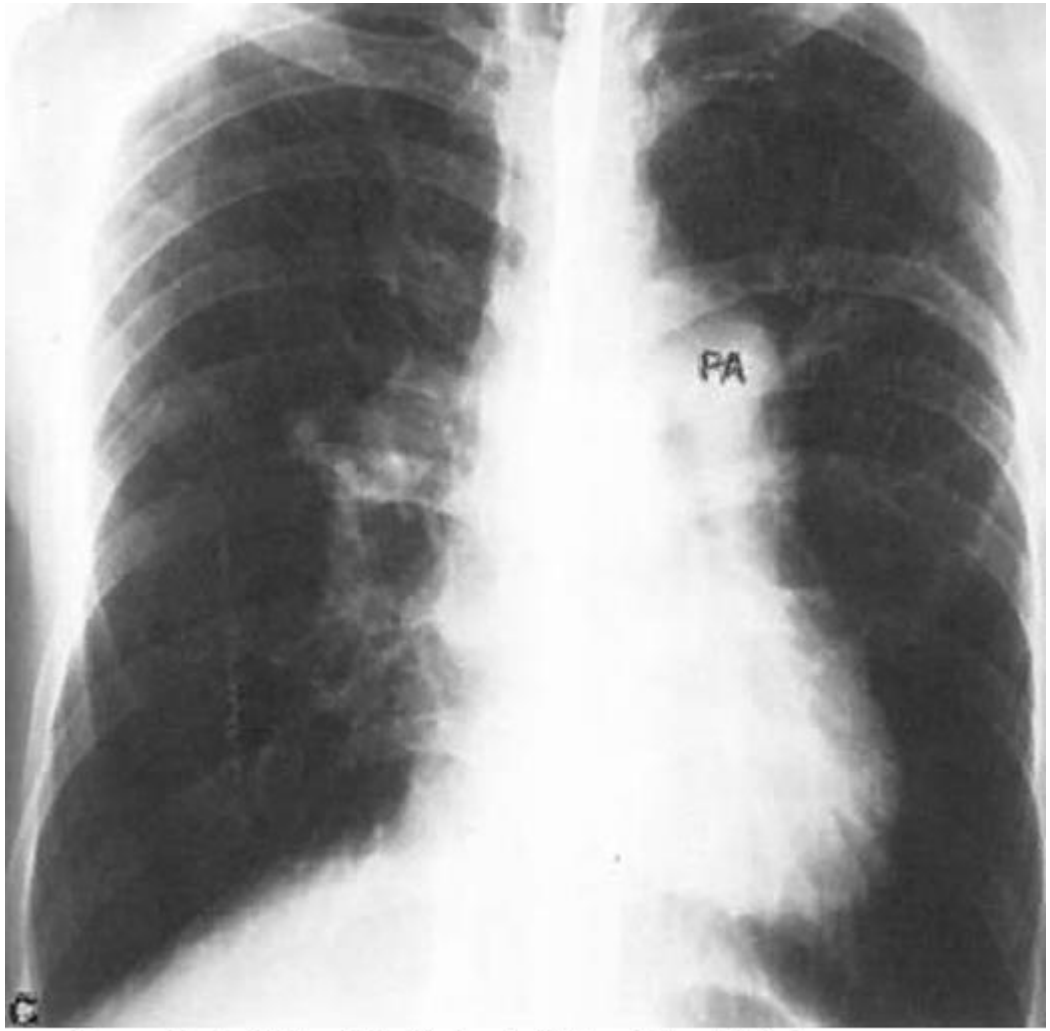


Table 20–10.

Alterations in pulmonary blood flow in cyanotic cardiac lesions.

Increased Pulmonary Blood Flow	Decreased Pulmonary Blood Flow
Total anomalous pulmonary venous return	Pulmonic stenosis
Tricuspid atresia with large ventricular septal defect	Tricuspid atresia/restrictive ventricular septal defect
Transposition of the great arteries	Tetralogy of Fallot
Truncus arteriosus	Pulmonary atresia with intact ventricular septum

Current Diagnosis & Treatment: Pediatrics, 25e

Table 20–9.

Lesion-specific chest radiographic findings.

Diagnosis	Chest Radiograph Appearance
D-transposition of the great arteries	Egg on a string
Tetralogy of Fallot	Boot-shaped heart
Unobstructed total anomalous pulmonary venous drainage	Snowman
Obstructed total anomalous pulmonary venous drainage	Small heart with congested lungs
Coarctation	Figure 3 sign + rib notching

Current Diagnosis & Treatment: Pediatrics, 25e

Cardiac tube

- Cardiac precursor cells arise from the epiblast.
- Associated with splanchnic mesoderm.
- The underlying endoderm induces these cells to form cardiac myoblasts.
- At the same time blood islands are forming nearby.
- Cardiac cells surround blood islands, forming a horseshoe shaped tube (the heart field).

Cardiac tube

- Two laterally situated tubes are present by day 19.
- The first heart field expresses transcription factors TBX5 and HAND1.
- The left ventricle will form from the first heart field.
- The second heart field lies dorsal to the first heart field
- Expresses HAND2 and FGF10.
- Most of the atria, the right ventricle, and the outflow tract arise from the second heart field.

Cardiac tube

- Initially the cardiac tube is suspended by a dorsal mesentery.
- At this point the heart consists of endothelium and splanchnic mesoderm.
- The splanchnic mesoderm develops into two layers (the myocardium and the cardiac jelly).
- Cells from the splanchnic mesoderm also migrate and give rise to epicardium.
- The result is a three layered tube.

Cardiac tube

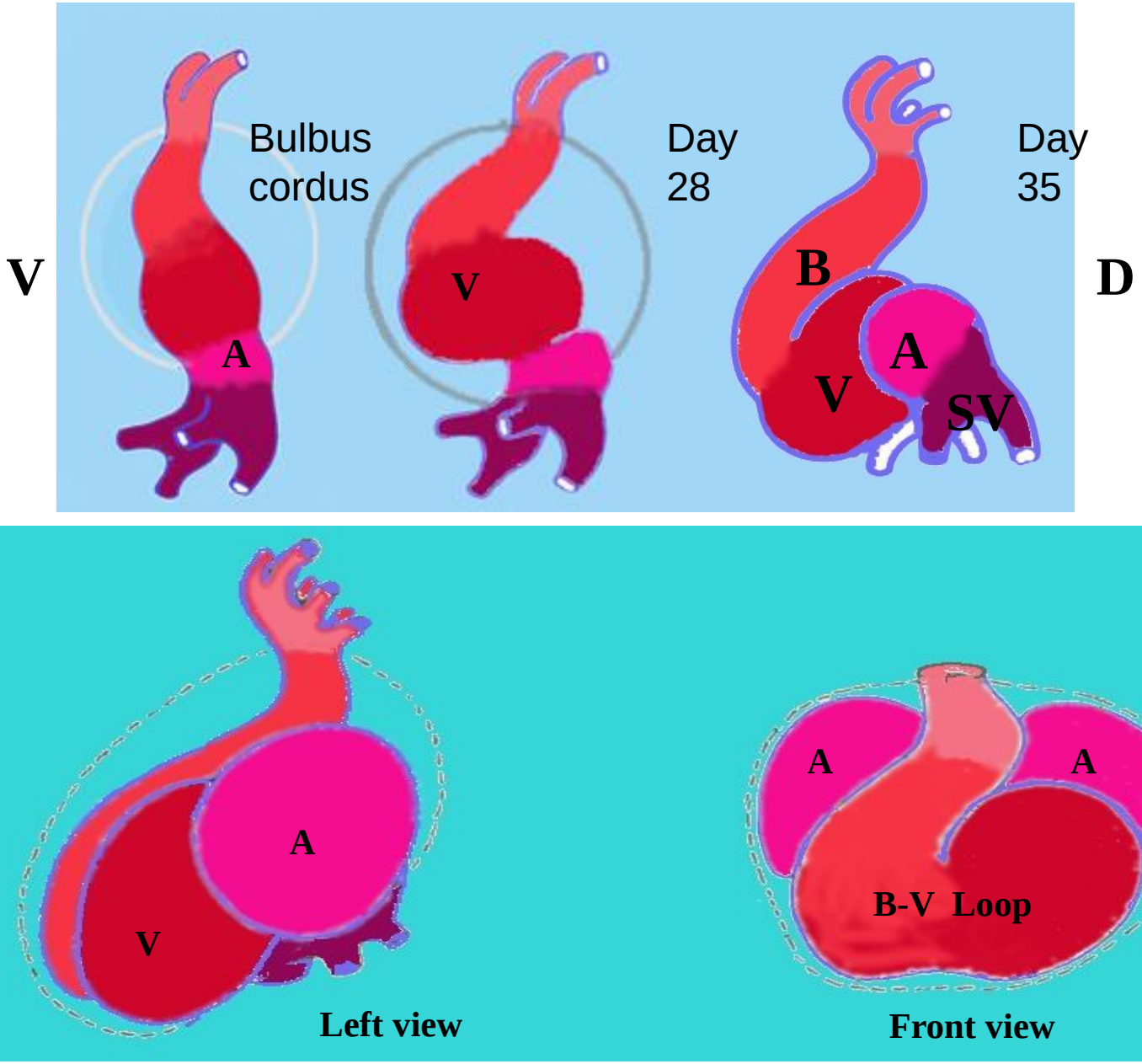
- By day 21, cells from the second heart field migrate into the anterior and posterior ends of the tube to form the right ventricle, conotruncus, and part of the atria.
- At day 28, the bulbus cordis moves anteriorly, inferiorly and to the right. The ventricle moves anteriorly, inferiorly and to the left. The atrium shifts posteriorly and superiorly.
- Cardiac neural crest cells migrate into the outflow tract from the neural folds to septate the outflow tract and pattern the bilaterally symmetric aortic arch arteries III, IV, VI.

Cardiac tube

- Also at day 28, a subset of endocardial cells delaminates and invades the extracellular matrix
- Underlies the future atrioventricular canal and outflow tract
- Proliferates and differentiates into mesenchymal cells that are responsible for valve development (endocardial cushions).

Cardiac tube

- Rotation at the 5th week permits the right sinus horn to be incorporated into the right atrium (sinus venarum).
- Valves form for the inferior vena cava and the coronary sinus.
- The crista terminalis divides the trabeculated part of the right atrium from the sinus venarum.
- The moderator band (septomarginal trabecula) is located in the right ventricle.


V
Bulbus cordus
A
Day 28
V
Day 35
B
A
V
SV
D
A
A
V
Left view
B-V Loop
Front view
Day 50

Endocardial cushions

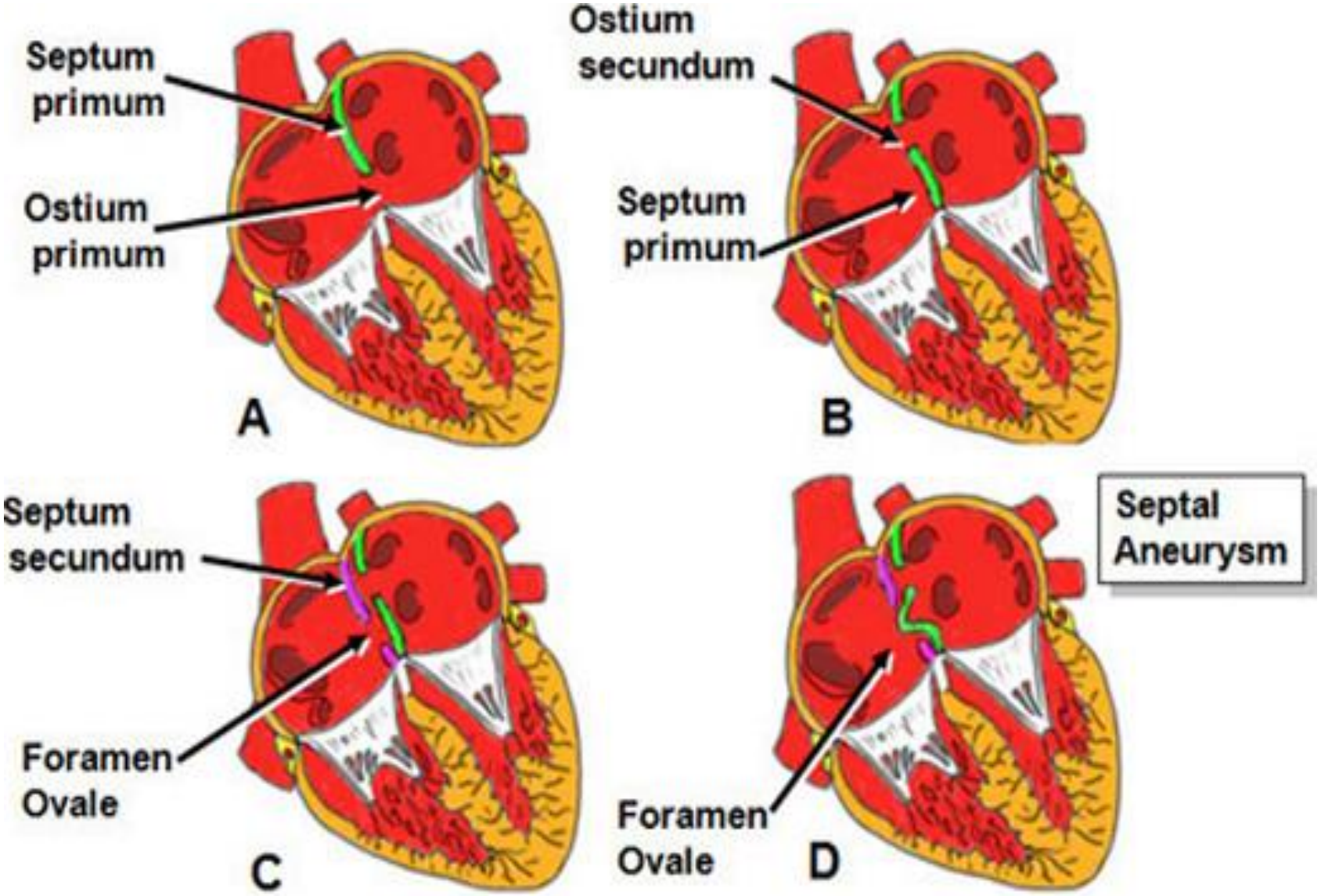
- At day 50, septation of the ventricles, atria, and atrioventricular valves results in the appropriately configured four chambered heart.
- That endocardial cushion between the bulbous cordis and truncus arises from the neural crest and serves to partition the truncus.
- That endocardial cushion between the atria and ventricle arises from splanchnic mesoderm and partitions the atria, ventricles and atrio-ventricular canals.

Endocardial cushions

- The septum primum grows down from the roof of the common atrium towards the fused endocardial cushions.
- It is a movable septum.
- During fetal period septum primum acts like a valve of foramen ovale letting blood to flow from right atrium (high pressure side) to left atrium (low pressure side)
- The septum secundum appears in the roof of common atrium on the right side of septum primum.
- It is a rigid septum.

Endocardial cushions

- The ostium primum is closed by the fusion of septum primum with the AV septum.
- The ostium secundum is formed by the rupture of upper part of septum primum.



Source: Maxine A. Papadakis, Stephen J. McPhee, Michael W. Rabow: Current Medical Diagnosis and Treatment 2020

Development of the vessels

- All arteries, veins and lymphatic channels form from mesoderm and all vessels begin as groups of mesodermal cell clusters.
- Extra-embryonic vessels form initially from the yolk sac.
- Embryonic vessels form shortly after the extra-embryonic vessels.
- Blood cells form from the yolk sac.
- At 9 weeks, in the liver; at 12 weeks, in the spleen; at 28 weeks, in the bone marrow.

Development of the vessels

- During the 4th and 5th weeks of development, pharyngeal arches form at the cranial end of the embryo and each of the arches receives a blood vessel.
- During the 5th week of development, three systems of veins can be observed:
- Vitelline (omphalomesenteric); umbilical; cardinal.
- The ductus venosus bypasses the sinusoids of the liver.
- After birth the left umbilical vein obliterates to form the ligamentum teres hepatis.
- After birth the ductus venosum obliterates to form the ligamentum venosum.

Fetal circulation

- Two parallel circulations with 3 shunts.
- Eustachian valve directs oxygenated blood from inferior vena cava across foramen ovale.
- Ductus arteriosus directs blood from pulmonary arteries to aorta.
- Ductus venosus directs oxygenated blood from umbilical vein to inferior vena cava.

Fetal circulation

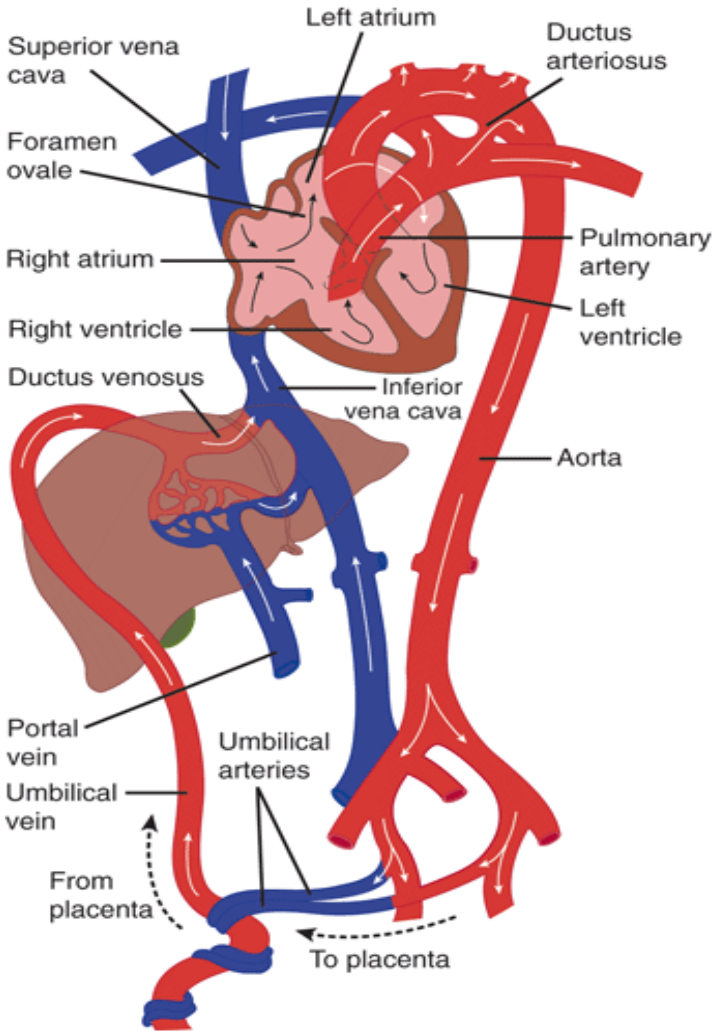


Fig. 34-18 Accessed 02/01/2010

Important transcription factors

- GATA4, TBX5, and NKX2-5 are three transcription factors that bind to one another and co-regulate target genes required for proper development of the heart.
- Mutations in any one are associated with septal and conduction defects.
- Endoderm secretes NKX2-5
- BMP2 and 4 with blockade of WNT (secreted by neural tube) leads to expression of NKX2-5 and creates heart forming region.

Important transcription factors

- GATA4 and TBX20 mutations are found in cardiomyopathy.
- Notch 1 mutation found in bicuspid aortic valve.
- Jagged1 and Notch2 mutations are found in Tetralogy of Fallot.
- BMP also upregulates FGF8
- Retinoic acid gradient leads to venous development

Innocent murmurs

- Still's murmur
- Inferior aspect of left lower sternal border
- Vibratory/musical quality; soft. Often positional (heard upright/sitting; may disappear when supine.)
- Early systolic
- Pulmonary ejection murmur
- Ejection sound heard at superior aspect of left lower sternal border
- Peripheral pulmonary stenosis
- Low-pitch murmur with radiation to back and axillae

Table 20–4.

Pathologic murmurs.

Systolic Ejection	Pansystolic	Diastolic	Continuous
Semilunar valve stenosis (AS/PS/truncal stenosis)	VSD	Semilunar valve regurgitation	Runoff lesions
ASD	AVVR (MR/TR)	AI/PI/truncal insufficiency	PDA/AVM/aortopulmonary collaterals
Coarctation		AV valve stenosis (MS/TS)	

Table 20–2.

Cardiac defects in common syndromes.

Genetic Syndrome	Commonly Associated Cardiac Defect
Down syndrome	AVSD
Turner syndrome	Bicuspid aortic valve, coarctation, dilated aortic root, hypertension
Noonan syndrome	Dysplastic pulmonic valve, HCM
Williams-Beuren syndrome	Supravalvular aortic stenosis, PPS, coronary ostial stenosis
Marfan syndrome	MVP, MR, dilated aortic root
Fetal alcohol syndrome	VSD, ASD
Maternal rubella	PDA, PPS
Loeys-Dietz syndrome	Aneurysmal PDA, dilated aortic root, tortuous arteries throughout the body

Diagnostic clues

- Heart failure within the first 12 to 18 hours of life is usually a result of malformations that involve pressure or volume overload independent of pulmonary flow
- Severe valvular regurgitation
- Systemic arterio-venous fistula

Diagnostic clues

- A majority of full-term infants presenting with severe heart failure during the remainder of the first week have critical obstruction to systemic arterial flow
- Unmasked by narrowing or closure of the ductus arteriosus.
- Aortic atresia
- Coarctation of the aorta
- Interruption of the aortic arch
- Critical aortic stenosis.

Diagnostic clues

- During the second week of life
- Aortic atresia and coarctation remain the most common causes of heart failure
- Left ventricular volume overload from ventricular septal defect
- Transposition of the great arteries with a ventricular septal defect
- Truncus arteriosus make their appearance.
- These malformations present as the pulmonary vascular resistance falls, increasing the left-to-right shunt.

Diagnostic clues

- Ventricular septal defect is the most frequent cause of congestive failure
- Others (in descending order):
- Transposition of the great arteries
- Coarctation of the aorta
- Complete atrio-ventricular canal
- Patent ductus arteriosus.

Table 12.2 Frequencies of Congenital Cardiac Malformations[a]

Malformation	Incidence per Million Live Births	%
Ventricular septal defect	4482	42
Atrial septal defect	1043	10
Pulmonary stenosis	836	8
Patent ductus arteriosus	781	7
Tetralogy of Fallot	577	5
Coarctation of the aorta	492	5
Atrioventricular septal defect	396	4
Aortic stenosis	388	4
Transposition of the great arteries	388	4
Truncus arteriosus	136	1
Total anomalous pulmonary venous connection	120	1
Tricuspid atresia	118	1
Total	9757	

[a]Presented as upper quartile of 44 published studies. Percentages do not add up to 100% because of rounding. Does not include bicuspid aortic valves.

Data from: Hoffman JI, Kaplan S: The incidence of congenital heart disease, *J Am Coll Cardiol* 39(12):1890–1900, 2002.

Table 12.3 Selected Examples of Gene Defects Associated With Congenital Heart Disease[a]

Disorder	Gene(s)	Gene Product Function
Nonsyndromic		
ASD or conduction defects	*NKX2.5*	Transcription factor
ASD or VSD	*GATA4*	Transcription factor
Tetralogy of Fallot	*ZFPM2* or *NKX2.5*	Transcription factors
Syndromic[b]		
Alagille syndrome—pulmonary artery stenosis or tetralogy of Fallot	*JAG1* or *NOTCH2*	Signaling proteins or receptors
Char syndrome—PDA	*TFAP2B*	Transcription factor
CHARGE syndrome—ASD, VSD, PDA, or hypoplastic right side of the heart	*CHD7*	Helicase-binding protein
DiGeorge syndrome—ASD, VSD, or outflow tract obstruction	*TBX1*	Transcription factor
Holt-Oram syndrome—ASD, VSD, or conduction defect	*TBX5*	Transcription factor
Noonan syndrome—pulmonary valve stenosis, VSD, or hypertrophic cardiomyopathy	*PTPN11*, *KRAS*, *SOS1*	Signaling proteins

ASD, Atrial septal defect; *CHARGE*, posterior coloboma, heart defect, choanal atresia, retardation, genital and ear anomalies; *PDA*, patent ductus arteriosus; *VSD*, ventricular septal defect.

[a]Different mutations can cause the same phenotype, and mutations in some genes can cause multiple phenotypes (e.g., *NKX2.5*). Many of these congenital lesions also can occur sporadically, without specific genetic mutation.

[b]Only the cardiac manifestations of the syndrome are listed; the other skeletal, facial, neurologic, and visceral changes are not.

Atrioventricular septal defects

- In the asplenia syndrome, the complete variety is almost universal;
- With polysplenia, it occurs in about one-quarter of cases.
- An atrial septal defect of the secundum type is present in about half of these cases.
- A double orifice of the mitral valve may be associated with the incomplete type.
- Tetralogy of Fallot may be associated with the complete type.

Atrioventricular septal defect

- Murmur often inaudible in neonates.
- Loud pulmonary component of S_2.
- Common in infants with Down syndrome.
- ECG with extreme left-axis deviation.

Atrioventricular septal defect

- Results from incomplete fusion of the embryonic endocardial cushions.
- The endocardial cushions help to form the “crux” of the heart
- Includes the lower portion of the atrial septum, the membranous portion of the ventricular septum, and the septal leaflets of the tricuspid and mitral valves.

Atrioventricular septal defect

- Complete AVSD
- The valves are located in the mid-portion of the defect.
- Both atrial and ventricular components of the septal defect are present and the left- and right-sided AV valves share a common ring or orifice.
- Bi-directional shunting if pulmonary vascular resistance increased
- Common in Down's syndrome

Atrioventricular septal defect

- Partial AVSD
- There is a low insertion of the AV valves, resulting in a primum ASD without a ventricular defect component.
- In partial AVSD, there are two separate AV valve orifices and usually a cleft in the left-sided valve.
- Partial AVSD behaves as an ASD

Clinical diagnosis

- Neonates may have tachypnea, diaphoresis with feeding, recurrent pneumonia
- Murmur may be inaudible in neonate due to relatively equal systemic and pulmonary resistance
- At 4-6 weeks with drop in pulmonary vascular resistance, a soft VSD-like murmur develops
- S_2 is loud
- A diastolic flow murmur is present at the apex and the left lower sternal border

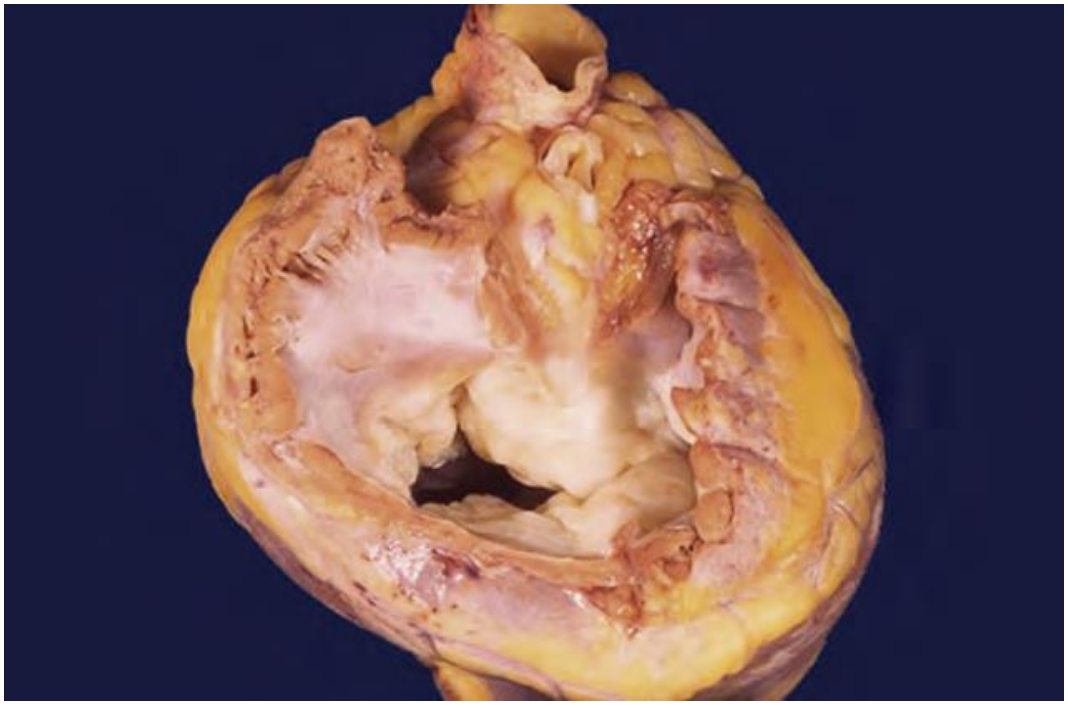
Clinical diagnosis

- If severe pulmonary vascular obstructive disease is present, there is usually dominant RV enlargement.
- S_2 is palpable at the pulmonary area
- No thrill is felt.
- No diastolic flow murmurs are heard.
- If a right-to-left shunt is present, cyanosis will be evident.

Testing and Treatment

- Cardiac enlargement with increased pulmonary vascular markings on chest x-ray.
- Echocardiography is diagnostic
- Surgical closure of the partial form is of low risk.
- May develop LV outflow obstruction and mitral valve dysfunction as late post-surgical complications
- Complete form must be closed before age 1, before the onset of irreversible pulmonary hypertension

Atrioventricular septal defect



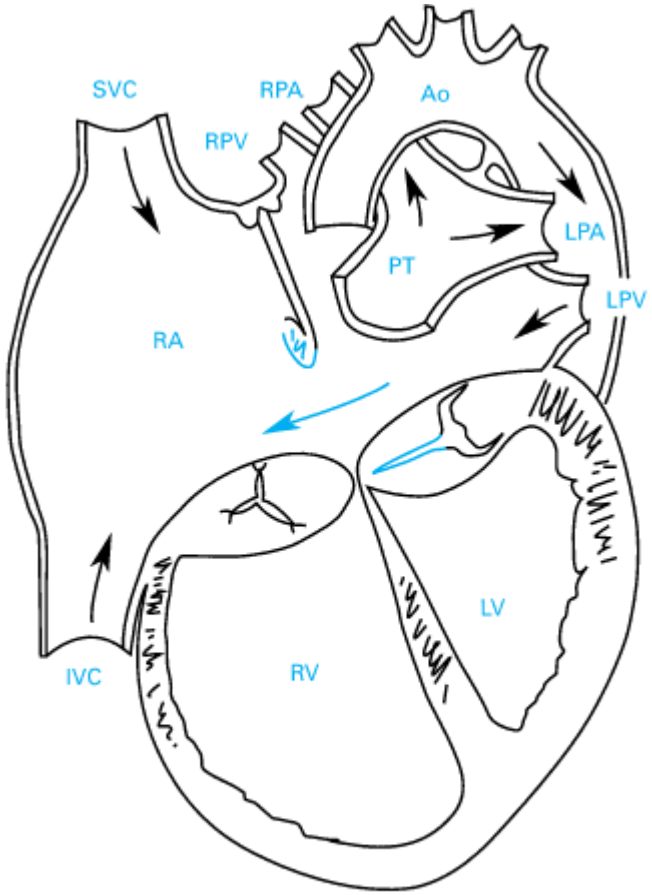
Sheppard, MN, Herrington, CS, “The Cardiovascular System,” in Herrington, CS (ed), Muir’s Textbook of Pathology, 15th edition. 2014. CRC Press. Boca Raton, Florida. Fig 2-21

A severe defect is shown in which there is only a single large atrioventricular valve, as visible superiorly, that separates a single ventricle from a single atrium.

This patient was able to survive with this two-chambered heart because a small amount of residual interventricular septum provided some direction to flow of oxygenated and unoxygenated blood, and because of pulmonic stenosis, which protected the lungs from the shunting.

Explanted heart (atria not seen)

Common atrioventricular canal



Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

The mitral valve shows a cleft in its anterior leaflet, while the tricuspid valve is undisturbed.

Ao, aorta; IVC, inferior vena cava; LPA, left pulmonary artery; LPV, left pulmonary vein; LV, left ventricle; RPA, right pulmonary artery; RPV, right pulmonary vein; SVC, superior vena cava.

Source: From Edwards JE. Classification of congenital heart disease in the adult. In: Roberts WC, ed. *Congenital Heart Disease in Adults*. Philadelphia: Davis; 1979:1. Reproduced with permission from the publisher and author.

Fig. 82-13 Accessed 04/01/2010

CYANOTIC CONGENITAL HEART DISEASE

Truncus arteriosus

- Early heart failure with or without cyanosis.
- Systolic ejection click.
- Develops embryologically as a result of failure of the division of the common truncus arteriosus into the aorta and the pulmonary artery.
- A VSD is almost always present.
- The number of truncal valve leaflets varies from one to six
- The valve may be insufficient or stenotic.

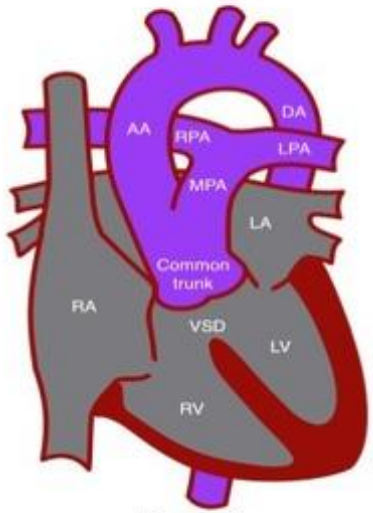
Truncus arteriosus

- A single main pulmonary artery may arise from the base of the trunk and gives rise to branch pulmonary arteries (type 1).
- Alternatively, the pulmonary arteries may arise separately from the common trunk, either in close association with one another (type 2)
- Or widely separated (type 3).
- This lesion can occur in association with an interrupted aortic arch.
- Oxygen saturation in the pulmonary artery is equal to that in the systemic arteries

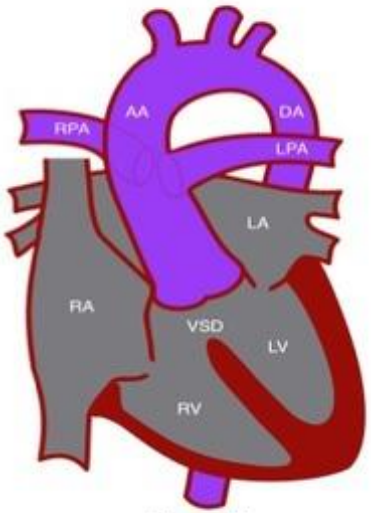
Classification

- Collett and Edwards system
- type I: (most common) both aorta and main pulmonary artery arise from a common trunk
- type II: pulmonary arteries arise separately from the posterior aspect of trunk, close to each other just above the truncal valve (negligible main pulmonary artery segment)
- type III: (least common) pulmonary arteries arise independently from either side of the trunk
- type IV: neither pulmonary arterial branch arising from the common trunk (pseudotruncus), currently considered a form of pulmonary atresia with a ventricular septal defect.

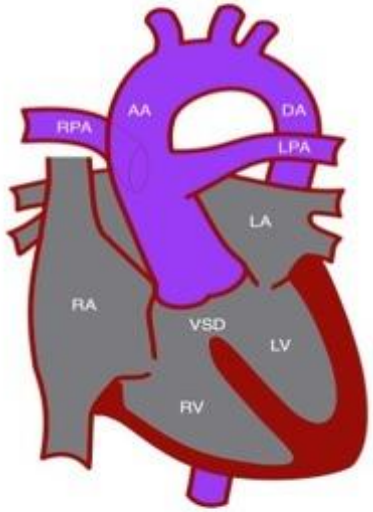
Collett & Edwards classification of truncus arteriosus



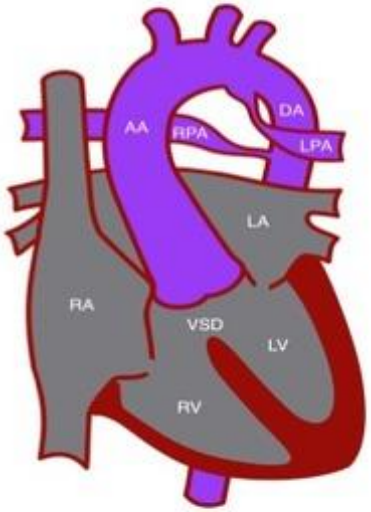
Type I



Type II



Type III

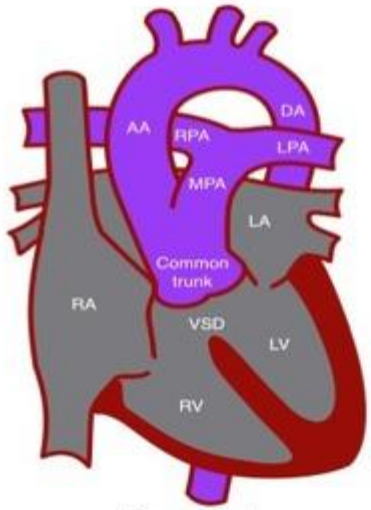


Type IV

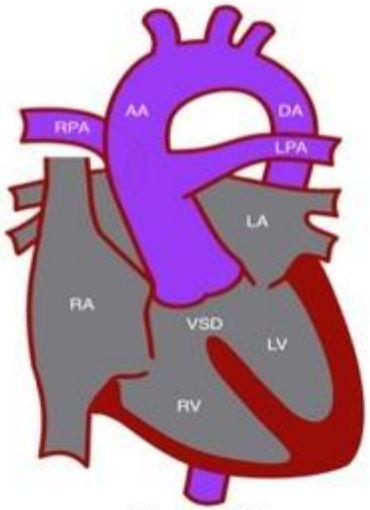
Classification

- Van Praagh system
- type A1**:** identical to the type I of Collett and Edwards
- type A2**:** separate origins of the branch pulmonary arteries from the common trunk
- type A3**:** origin of one branch pulmonary artery (usually the right) from the common trunk, with other lung supplied either by collaterals or a pulmonary artery arising from the aortic arch
- type A4**:** presence of an associated interrupted aortic arch

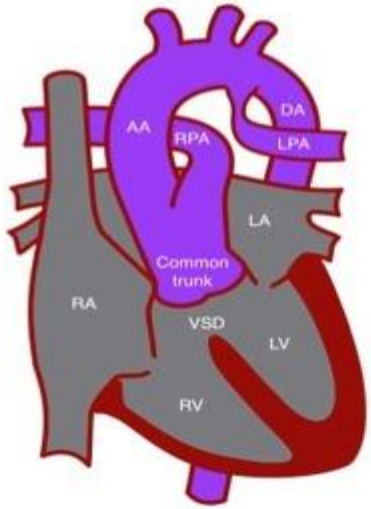
Van Praagh classification of truncus arteriosus



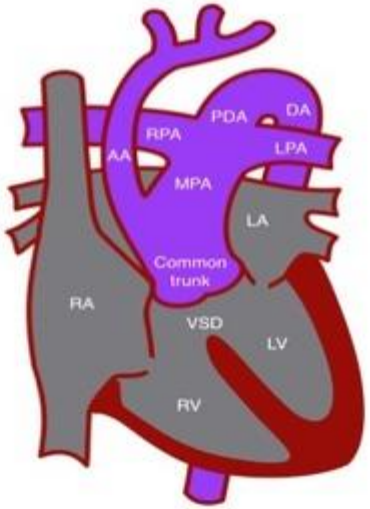
Type A1



Type A2



Type A3



Type A4

V. Tatco MD

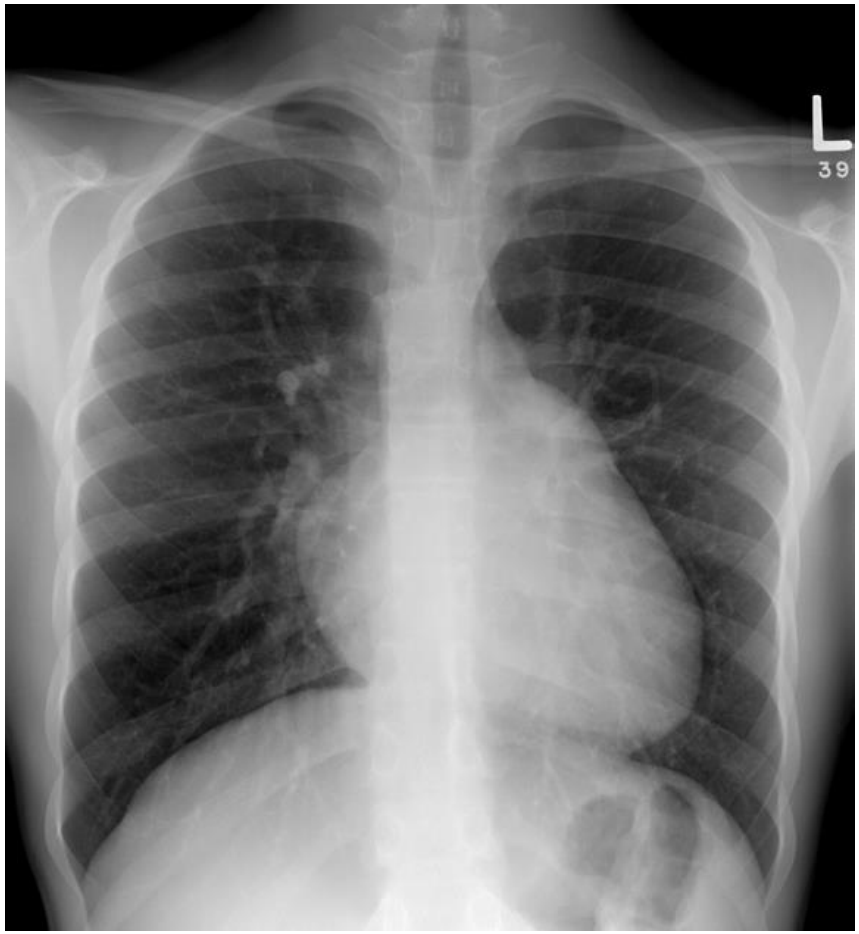
Truncus arteriosus

- A systolic thrill is common at the lower left sternal border.
- A loud early systolic ejection click is commonly heard. S_2 is single and accentuated.
- A loud holosystolic murmur is audible at the left lower sternal border.
- A diastolic flow murmur can often be heard at the apex
- Chest x-ray discloses cardiomegaly, absence of the main pulmonary artery segment, and a large aorta that has a right arch 30% of the time

Truncus arteriosus

- Echocardiography is diagnostic
- Surgery is usually performed in the neonatal period or early infancy to prevent pulmonary hypertension.
- The VSD is closed to allow LV egress to the truncal valve.
- The pulmonary artery (type 1) or arteries (types 2–3) are separated from the truncus as a block, and a valved conduit is fashioned from the RV to the pulmonary circulation.

Truncus arteriosus



https://radiopaedia.org/articles/truncus-arteriosus?lang=us

Right-to-left shunts

- Deoxygenated blood from the right side of the heart goes to the left side and is delivered to the body.
- This type of shunt usually results in cyanosis at the time of birth.
- “Blue” baby
- Tetralogy of Fallot is the most common cause of right-to-left shunts in newborns.
- Aorta overrides the ventricular septal defect in the right ventricle
- Pulmonic stenosis as protective (develop right ventricular hypertrophy)
- Secondary polycythemia as erythropoietin production stimulated by hypoxemia

Right-to-left shunts

- Tricuspid atresia (with ventricular septal defect or atrial septal defect).
- Ebstein's anomaly is tricuspid insufficiency; half have associated atrial septal defect.
- Truncus Arteriosus. Common arterial trunk receives a mixture of blood from the right and left ventricles. Initially presents as left-to-right shunt but reverses over months as pulmonary vascular resistance increases.
- Total Anomalous Pulmonary Venous Return. Pulmonary veins drain into a left inominate vein or coronary sinus and return to the left atrium by an atrial septal defect or patent foramen ovale.

Tetralogy of Fallot

- Five features are characteristic:
- (1) VSD.
- (2) Concentric RVH.
- (3) RV outflow obstruction due to infundibular stenosis.
- May have pulmonary valve stenosis as well, usually due to a bicuspid pulmonary valve or RV outflow hypoplasia.
- The degree of right ventricular outlet tract obstruction determines the amount of shunting across the VSD, pulmonary flow, and cyanosis.

Tetralogy of Fallot

- Mild pulmonic stenosis or: left to right shunting and no cyanosis
- Severe pulmonic stenosis: right to left shunting and cyanosis
- (4) Dilated aorta
- 50%, overrides the septum.
- (5) A right-sided aortic arch in 25%
- If there is an associated ASD, the complex is referred to as pentalogy of Fallot.

Tetralogy of Fallot

- Echocardiography/Doppler may underestimate significant pulmonary valve regurgitation.
- Cardiac catheterization if hypoplastic pulmonary arteries
- Arrhythmias are common
- Periodic ambulatory monitoring is recommended.
- Serious arrhythmias and sudden death may occur if the QRS is wide or the RV becomes quite large, or both.

Tetralogy of Fallot

- The aorta can be quite enlarged and aortic regurgitation may occur.
- If more than 50% of the aorta overrides into the RV outflow tract, the anatomy is referred to as a “double outlet RV.”
- 25%, right-sided aortic arch with anomalous left arch
- 7-9%, anterior descending coronary artery from the right cusp

Clinical diagnosis

- Few children are asymptomatic.
- Those with severe obstruction are deeply cyanotic from birth.
- In those with significant RV outflow obstruction, many have cyanosis at birth, and nearly all have cyanosis by age 4 months.
- The cyanosis usually is progressive, as subvalvular obstruction increases.
- Children with mild obstruction are minimally cyanotic or acyanotic.

Clinical diagnosis

- Systolic ejection murmur at the upper left sternal border in the 3rd left intercostal space
- Radiates to the back
- Most adult patients with tetralogy of Fallot have been operated on, usually with an RV outflow patch and VSD closure.
- The insertion site of a prior Blalock or other shunt may create a stenotic area in the branch PA and a continuous murmur occurs as a result.
- These pulmonary arterial stenotic bruits may best be heard on the lateral chest wall.

Clinical diagnosis

- Easy fatigability and dyspnea on exertion are common
- Hypoxemic spells, also called cyanotic or "Tet spells," are one of the hallmarks of severe disease
- In infants occur most commonly with crying or feeding
- In older children they can occur with exercise
- Most commonly start at age 4–6 months

Clinical diagnosis

- Tet spells are characterized by:
- (1) sudden onset of cyanosis or deepening of cyanosis
- (2) dyspnea
- (3) alterations in consciousness
- From irritability to syncope
- (4) decrease in or disappearance of the systolic murmur

Clinical diagnosis

- Increased right ventricular outlet tract obstruction or decreased systemic resistance leads to right to left shunting across VSD, resulting in decreased pulmonary blood flow, hypoxemia, and cyanosis.
- Knee chest position ameliorates
- Squatting to increase aortic valve reflection, increasing left heart pressures, reducing flow through the shunt
- Morphine (to reduce ventilatory drive).
- May require vasopressor and Oxygen.

Clinical diagnosis

- The jugular venous pulsations (JVP) may reveal an increased a wave from poor RV compliance or rarely a c-v wave due to tricuspid regurgitation.
- The right-sided arch has no consequence.
- Often a persistent pulmonary outflow murmur.
- P_2 may or may not be audible.
- A right-sided gallop may be heard.
- A residual VSD or an aortic regurgitation murmur may be present.

Testing

- The ECG reveals RVH and right axis deviation
- In repaired tetralogy, there is often a right bundle branch block pattern.
- The chest x-ray shows a classic boot-shaped heart with prominence of the RV and a concavity in the RV outflow tract.
- The aorta may be enlarged and right-sided.
- Echocardiography/Doppler usually establishes the diagnosis by noting the unrestricted (large) VSD, the RV infundibular stenosis, and the enlarged aorta

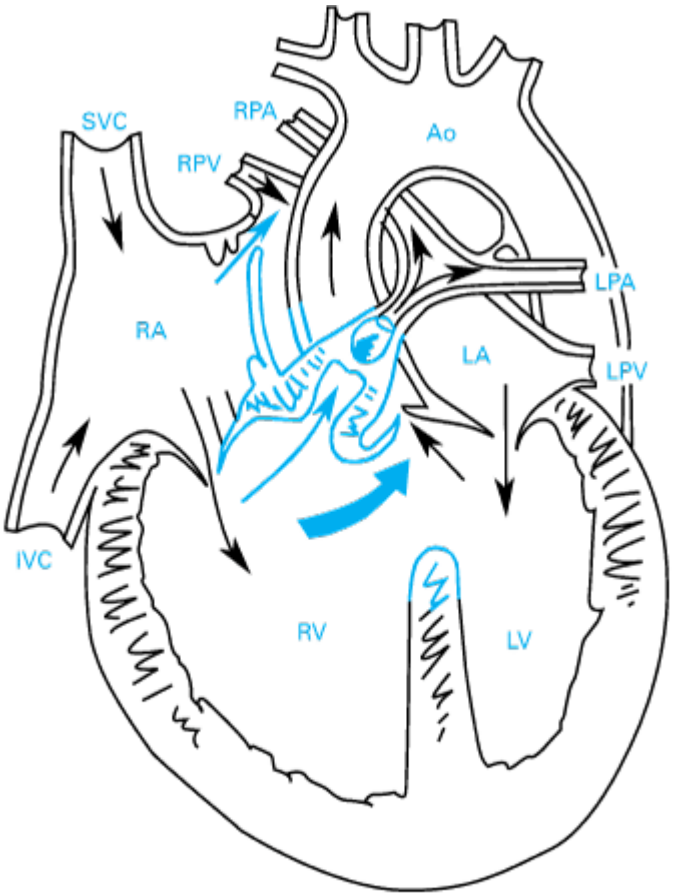
Treatment

- If significant RV outflow obstruction is present in the neonatal period, a systemic arterial to pulmonary artery shunt may be the initial surgical procedure to improve pulmonary blood flow
- The palliative procedure enables blood to reach the underperfused lung:
- Directly attaching one of the subclavian arteries to a main PA branch (classic Blalock shunt)

Treatment

- Shunt from the subclavian artery to the ipsilateral pulmonary artery to replace the ductus arteriosus which is ligated and divided or stented (modified Blalock shunt)
- The major limiting anatomic feature of total correction is the size of the pulmonary arteries.
- Those with complete repair may survive into adulthood.

Tetralogy of Fallot



Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

Fig. 82-29 Accessed 04/01/2010

1. There are infundibular and pulmonary valvular stenoses.
2. There is also right-to-left shunting at the atrial level.

Ao, aorta; IVC, inferior vena cava; LA, left atrium; LPA, left pulmonary artery; LPV, left pulmonary vein; LV, left ventricle; RA, right atrium; RPA, right pulmonary artery; RPV, right pulmonary vein; RV, right ventricle; SVC, superior vena cava.

Source: From Edwards JE. Classification of congenital heart disease in the adult. In: Roberts WC, ed. *Congenital Heart Disease in Adults*. Philadelphia: Davis; 1979:1. Reproduced with permission from the publisher and author

Tetralogy of Fallot

- Anterior malalignment of ventricular septum
- Ventricular septal defect (VSD)
- Right ventricular outlet tract obstruction
- Right ventricular hypertrophy
- Overriding aorta
- The degree of right ventricular outlet tract obstruction determines the amount of shunting across the VSD, pulmonary flow, and cyanosis.
- Mild pulmonic stenosis or: left to right shunting and no cyanosis
- Severe pulmonic stenosis: right to left shunting and cyanosis

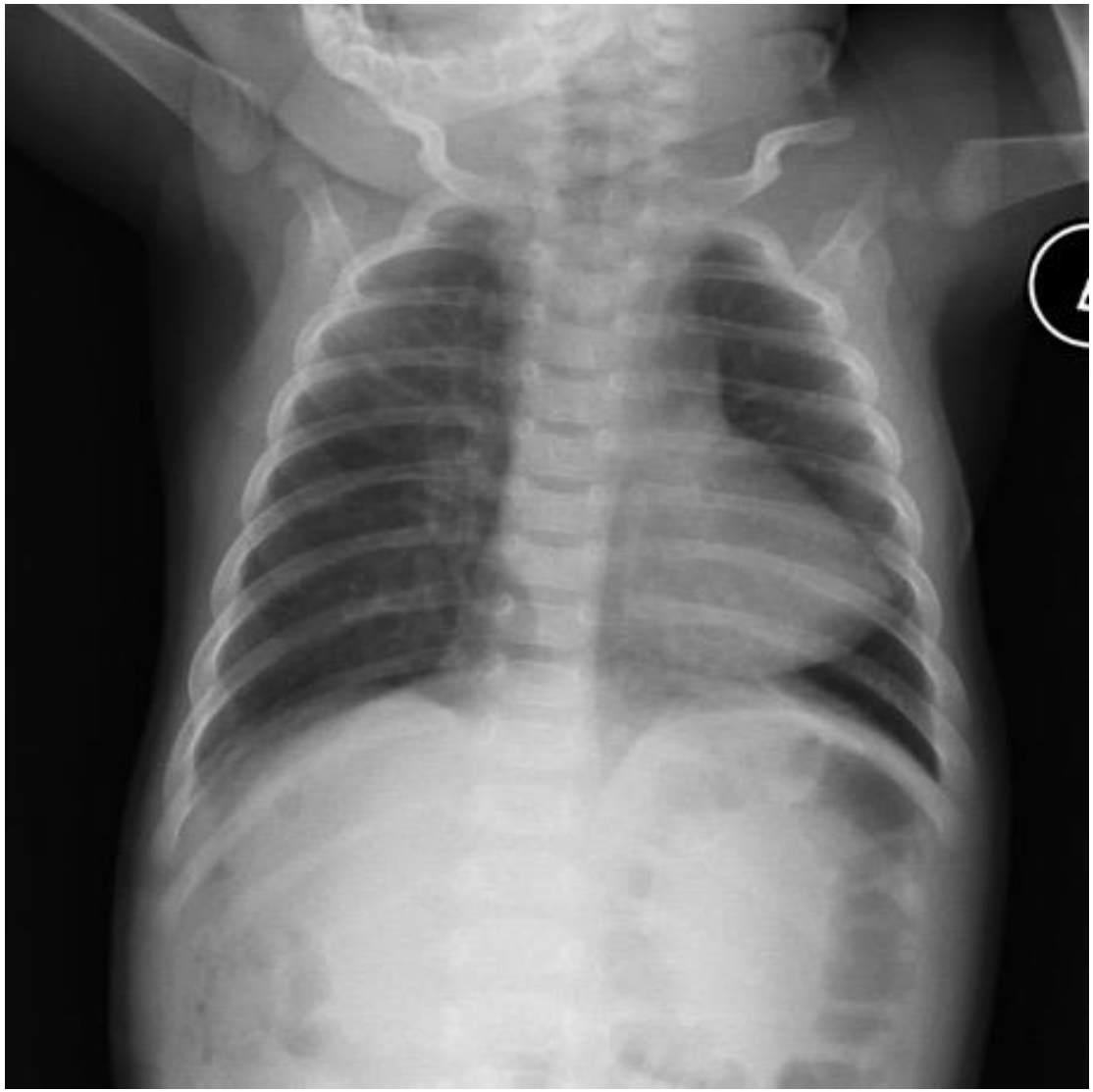
Hyperoxia test

- Obtain an arterial blood gas specimen after patient breathes 100% O_2 for 10 minutes.
- P_aO_2 < 100 mmHg; cyanotic congenital heart disease likely
- P_aO_2 > 250 mmHg; cyanotic congenital heart disease unlikely
- A “failed” hyperoxia test is a neonatal emergency.
- Urgent intervention required.

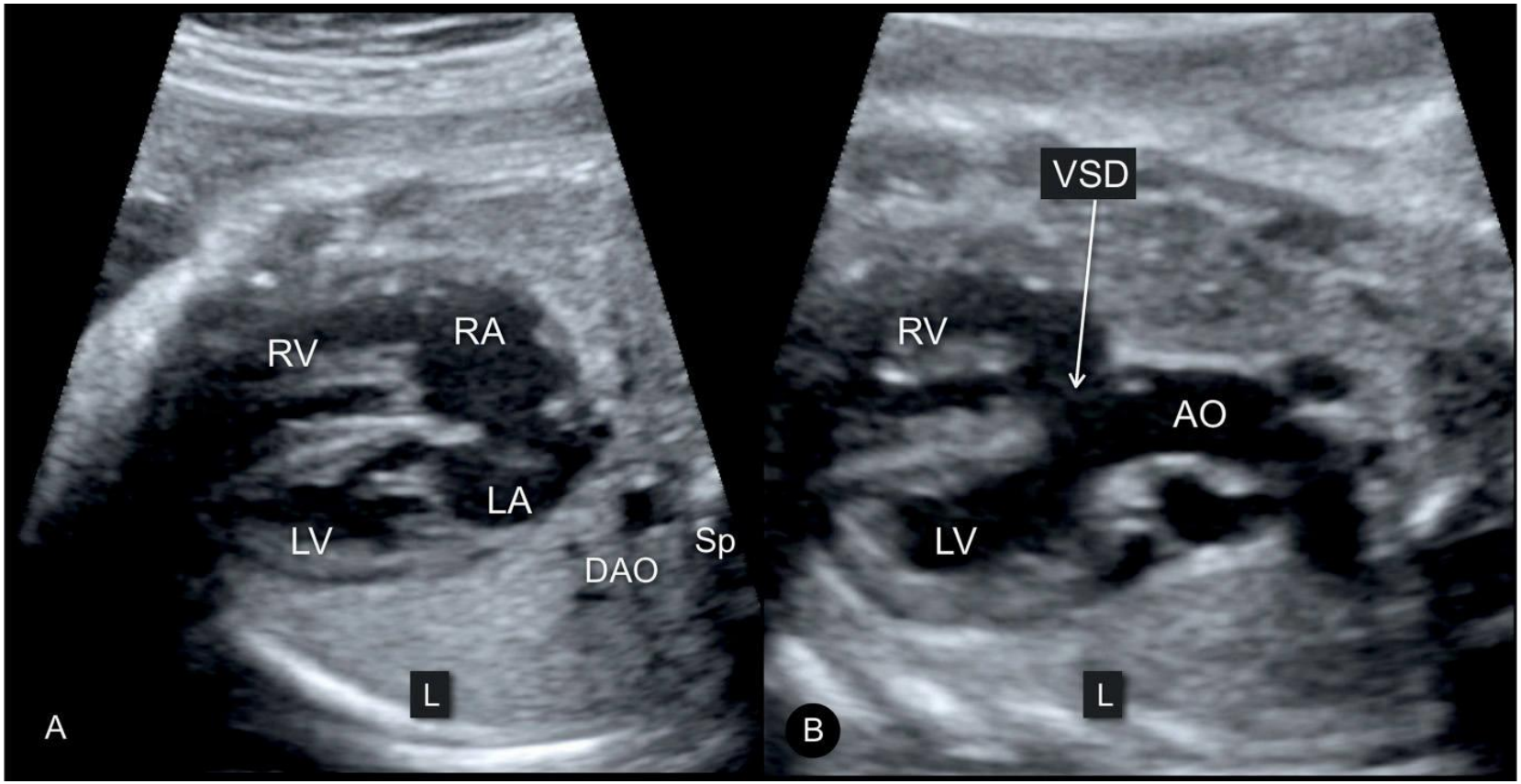
Tetralogy of Fallot

- There is a loud systolic murmur along the left sternal border, a loud single S2, and possible thrill.
- Right axis deviation and right ventricular hypertrophy on EKG.
- The chest x-ray demonstrates a “boot shaped heart” of normal size.
- There may be increased pulmonary vascular markings depending on pulmonary blood flow.
- Associated with mutations of Jagged 1 and Notch 2 as well as with deletions on chromosome 22.
- May be seen with di George syndrome (TBX1 mutation).

Tetralogy of Fallot



Tetralogy of Fallot



https://obgynkey.com/wp-content/uploads/2016/08/00461.jpeg

Tetralogy of Fallot

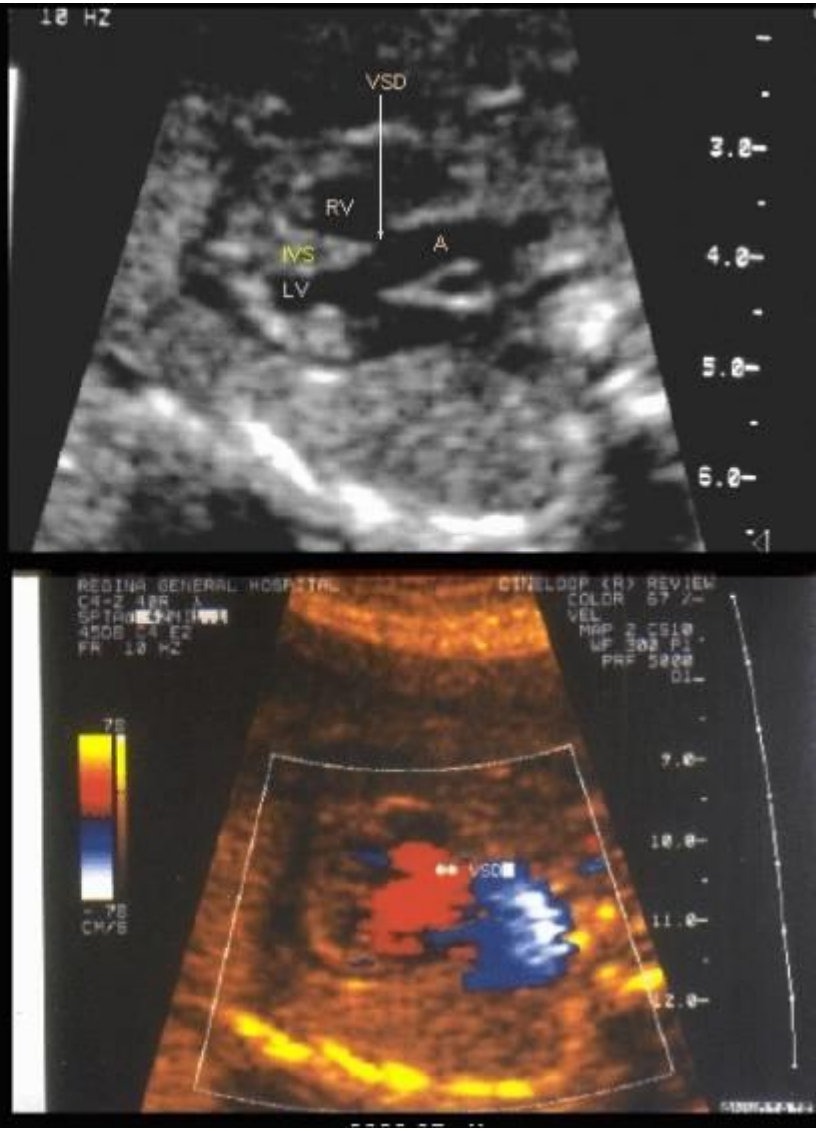
LV: Left Ventricle

RV: Right Ventricle

IVS: Interventricular septum

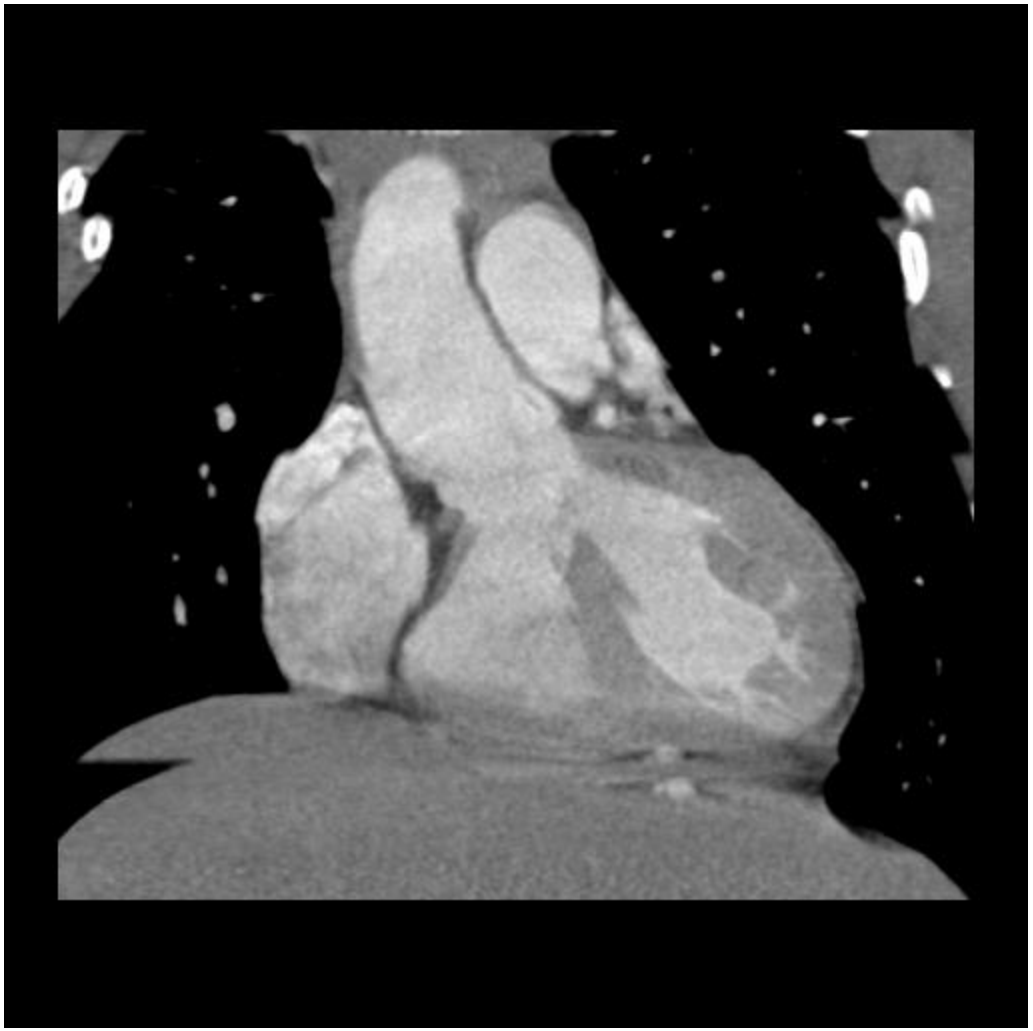
VSD: Interventricular Septal Defect

A: Aorta (Overriding Aorta)



http://www.fetalultrasound.com/online/text/7-049_files/image002.jpg

Tetralogy of Fallot



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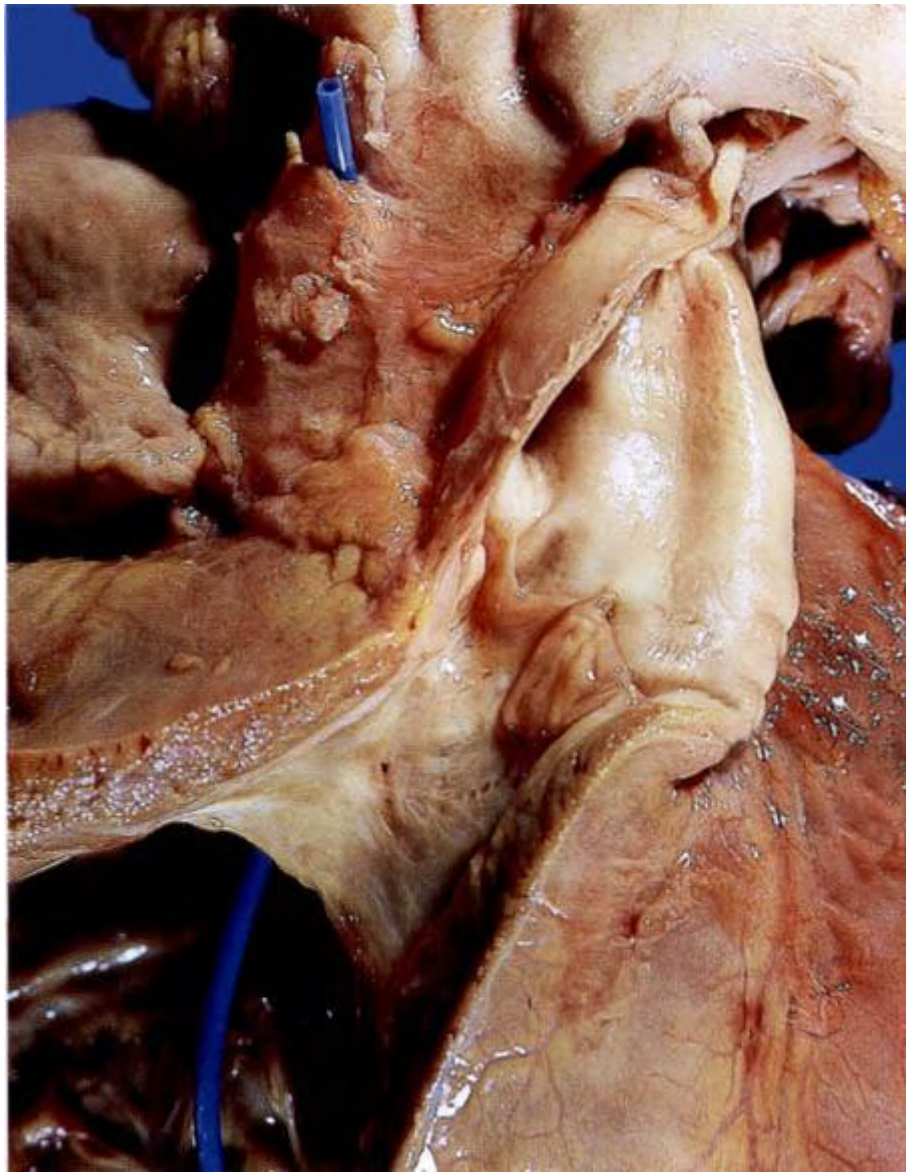


Tetralogy of Fallot

The outflow tract of the right ventricle is viewed in this specimen. There is pulmonary valve stenosis with dilatation of the pulmonary artery beyond this. The right ventricle is hypertrophied and the blue pointer passes from the cavity of the right ventricle into the aorta. This demonstrates the presence of a ventricular septal defect and an aorta overriding both ventricles.

Sheppard, MN, Herrington, CS, “The Cardiovascular System,” in Herrington, CS (ed), Muir’s Textbook of Pathology, 15th edition. 2014. CRC Press. Boca Raton, Florida. Fig. 1-62

Tetralogy of Fallot

- Total surgical repair as infant. Pulmonic valve abnormalities worsen later in life as heart grows. Rhythm abnormalities place at risk for sudden death.
- 75% of repaired patients survive past 1 year of age.
- 60% survive 4 years
- 30% survive 10 years
- 5% reach 40 years of age

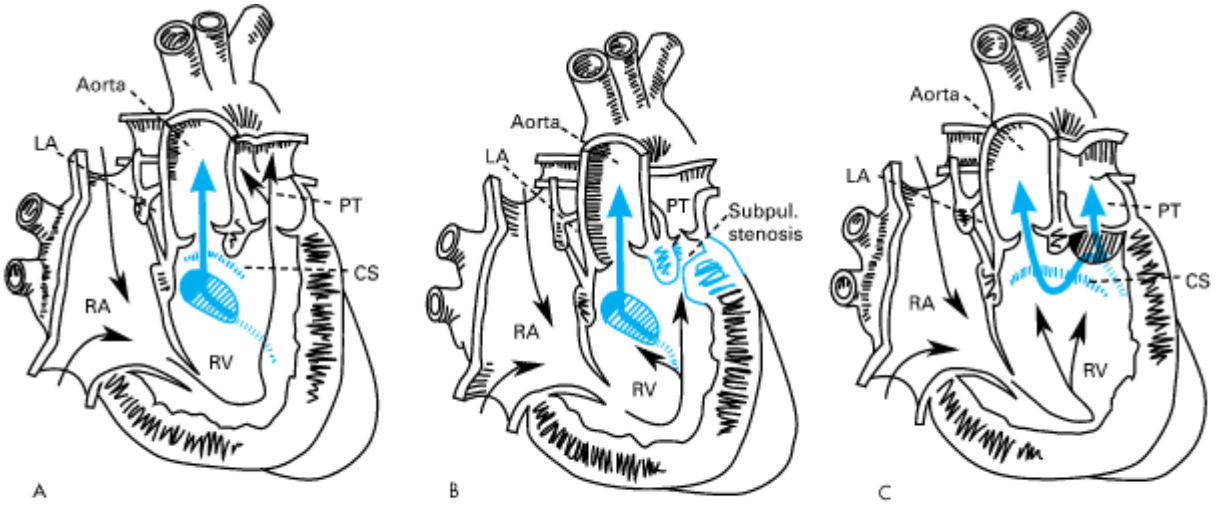
Teratology of Fallot
Other flows

- Tetralogy of Fallot with ASD associated with left to right shunt and no cyanosis.
- Flow is from left atrium to right atrium to right ventricle to left ventricle via VSD
- Tetralogy of Fallot with patent ductus arteriosus (PDA)
- Flow is from aorta (desaturated) via PDA into pulmonary artery through lungs into left atrium

Pulmonary atresia with ventricular septal defect

- Symptoms depend on degree of pulmonary blood flow.
- Pulmonary blood flow via PDA and/or aortopulmonary collaterals.
- Extreme form of tetralogy of Fallot
- Echocardiography is diagnostic

Double-outlet right ventricle

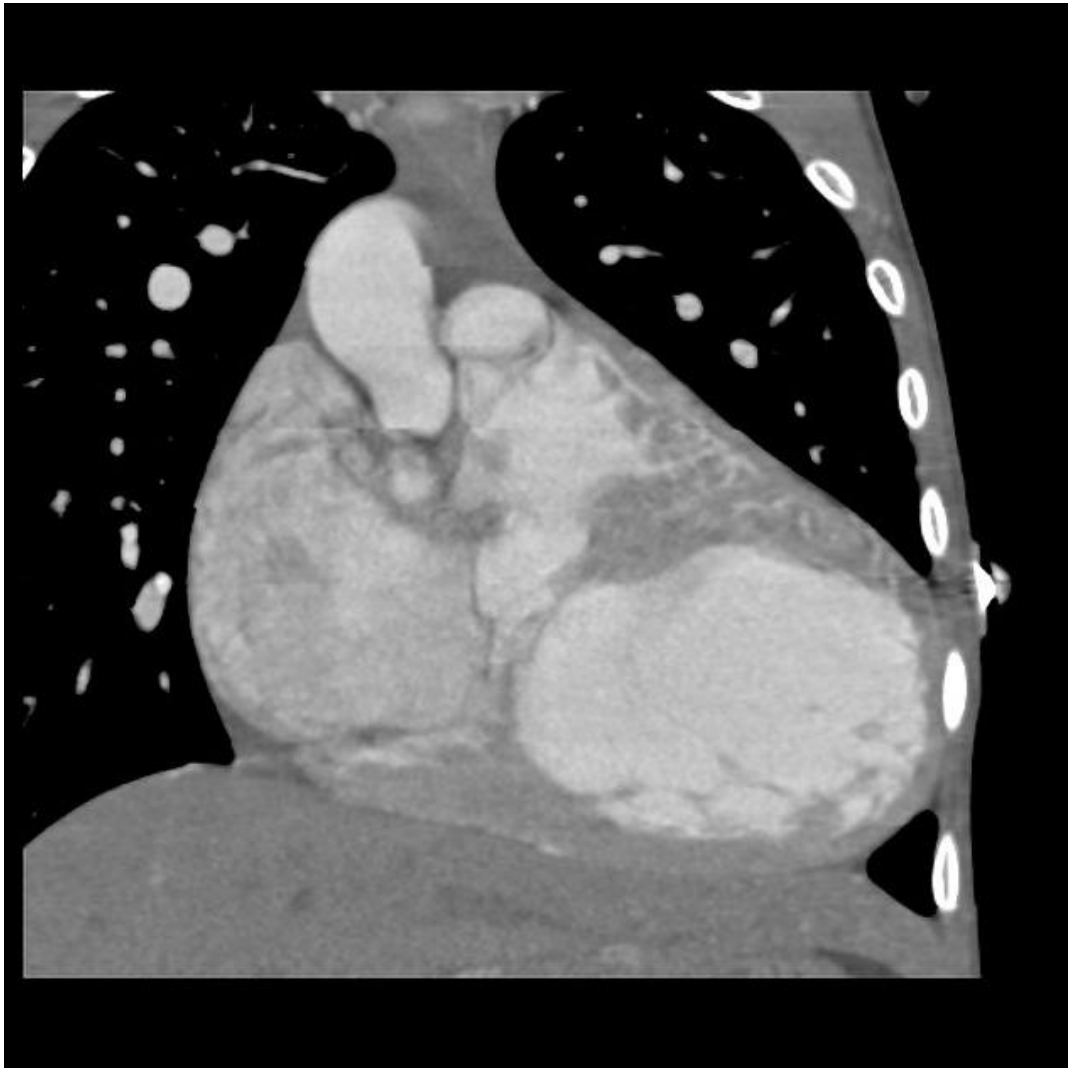


A. With subaortic ventricular septal defect without pulmonary stenosis. **B.** With subaortic ventricular septal defect and subpulmonary stenosis (Subpulmonary stenosis). **C.** With subpulmonary, supracristal ventricular septal defect. The so-called Taussig-Bing complex.

CS, crista supraventricularis; LA, left atrium; PT, main pulmonary arterial trunk; RA, right atrium; RV, right ventricle.

Fig. 82-42 Accessed 04/01/2010

Double outlet right ventricle



https://radiopaedia.org/articles/double-outlet-right-ventricle?lang=us

Complete transposition of the great arteries

- May present with extreme cyanosis (isolated circuits) and a single, loud S_2.
- Second most common cyanotic lesion
- Two distinct circulations:
- Aorta arises from right ventricle
- Pulmonary artery arises from left ventricle
- Must be accompanied by a ventricular septal defect, patent ductus arteriosus, or foramen ovale if pulmonary and systemic circuits are to interconnect to permit survival.
- A murmur is heard if a VSD or pulmonic stenosis is present.

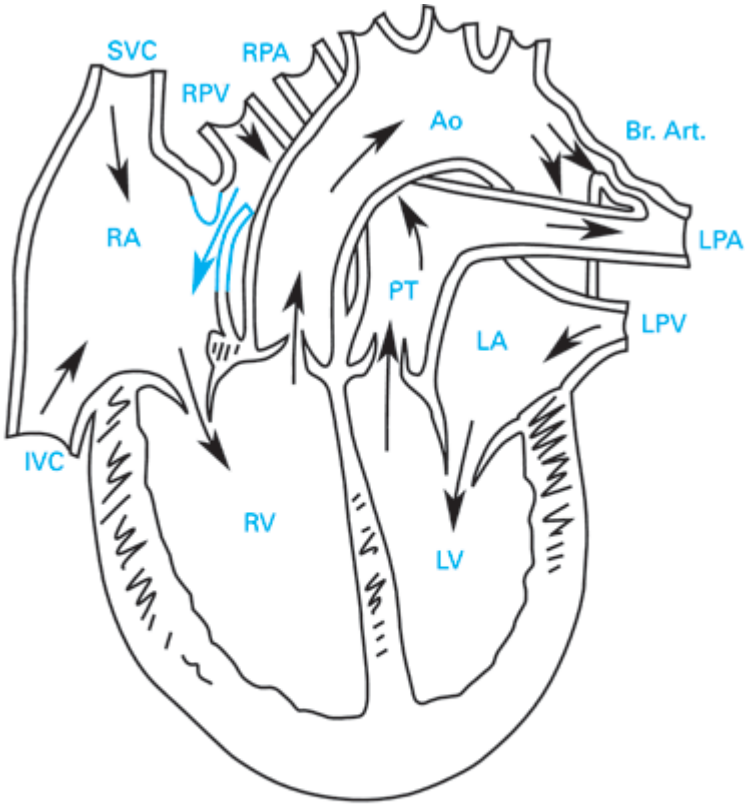
Complete transposition of the great arteries

- Associated with polyhydramnios such as seen in diabetic mothers
- Chest x-ray demonstrates cardiomegaly and a prominent aortic arch with unimpressive pulmonary arteries ("egg on a string").
- Vascular markings are increased.
- The EKG shows right ventricular hypertrophy as the right ventricle is acting as the systemic ventricle. The T wave is upright in V_1 after 3 days of age.
- Acute treatment is arterial balloon septostomy.

Complete transposition of the great arteries

- More than 50 percent of the semilunar valve orifices of both great arteries arise from the morphologic right ventricle.
- In most cases, the ventricles display a D loop, and the pulmonary arterial origin is normally positioned, arising from a conus above the right ventricle.
- The aorta also arises from the right ventricle above conal tissue.
- The two semilunar valves are at about the same level
- There is no fibrous continuity between the semilunar and mitral valves.

Complete transposition of the great arteries



A

Fig. 82-36 Accessed 04/01/2010

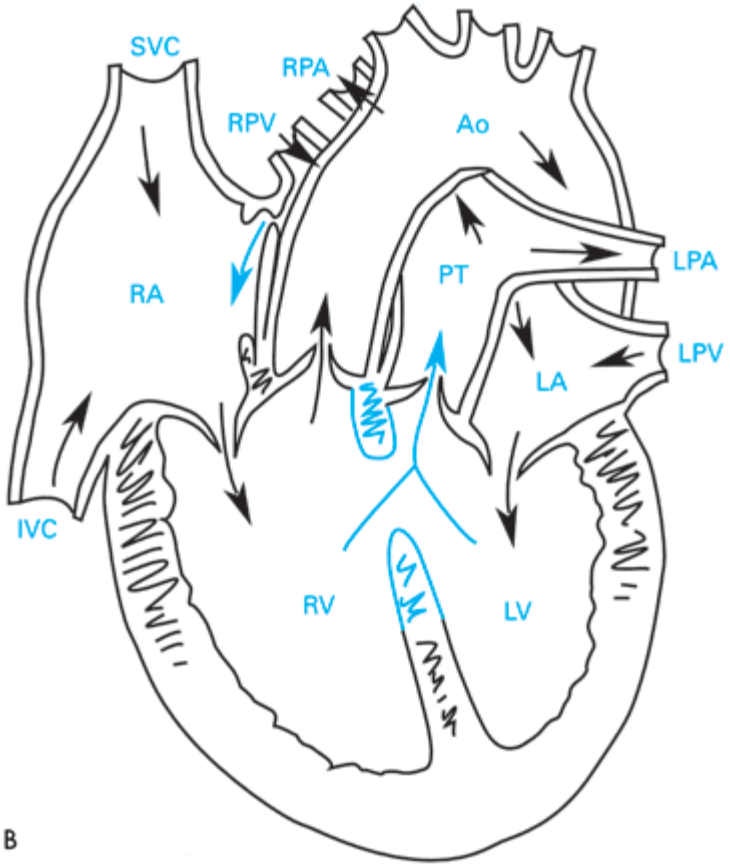
A. With intact ventricular septum. A patent foramen ovale and enlarged bronchial arteries (Br. Art.) are present.
Ao, aorta; LA, left atrium; LPA, left pulmonary artery; LPV, left pulmonary vein; LV, left ventricle; PT, main pulmonary arterial trunk; RA, right atrium; RPA, right pulmonary artery; RPV, right pulmonary vein; RV, right ventricle; SVC, superior vena cava.

Complete transposition of the great arteries

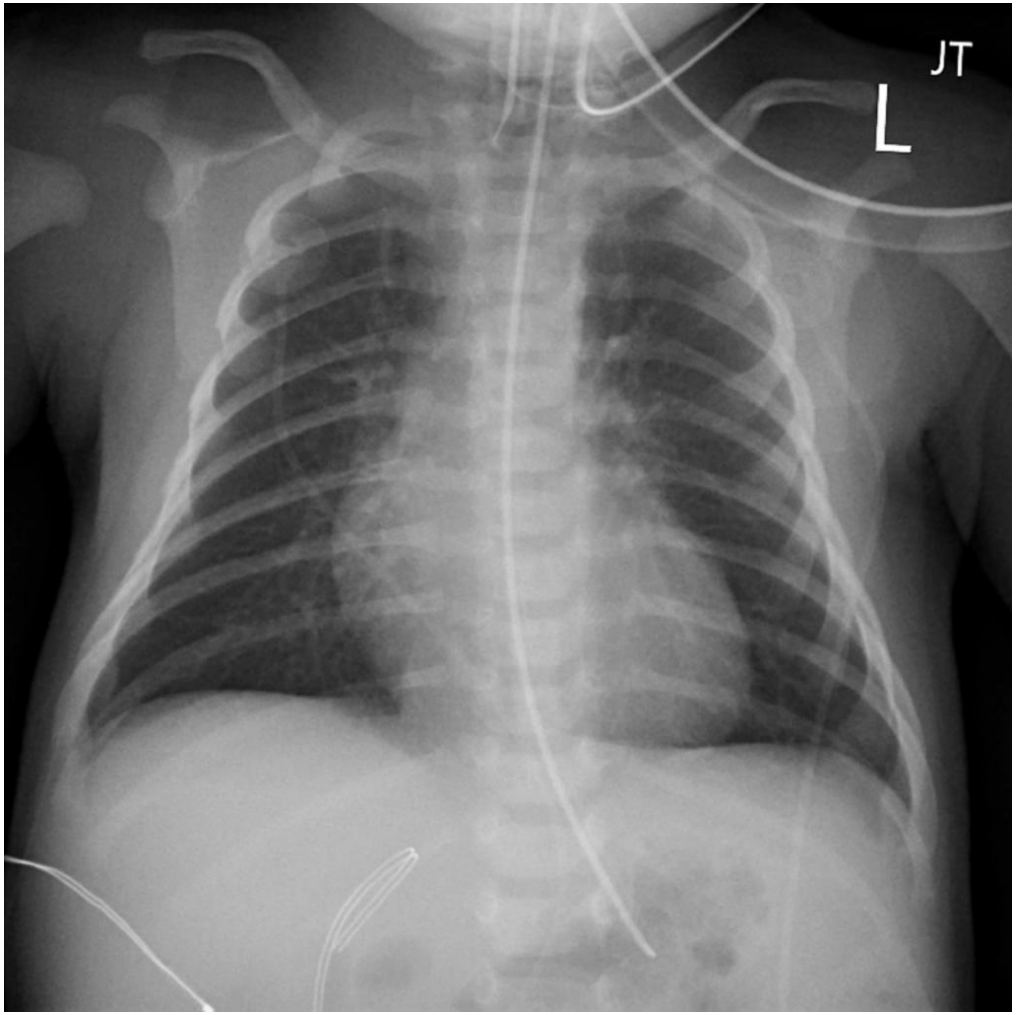
B. With ventricular septal defect and without pulmonary stenosis.

Ao, aorta; LA, left atrium; LPA, left pulmonary artery; LPV, left pulmonary vein; LV, left ventricle; PT, main pulmonary arterial trunk; RA, right atrium; RPA, right pulmonary artery; RPV, right pulmonary vein; RV, right ventricle; SVC, superior vena cava.

Fig. 82-36 Accessed 04/01/2010



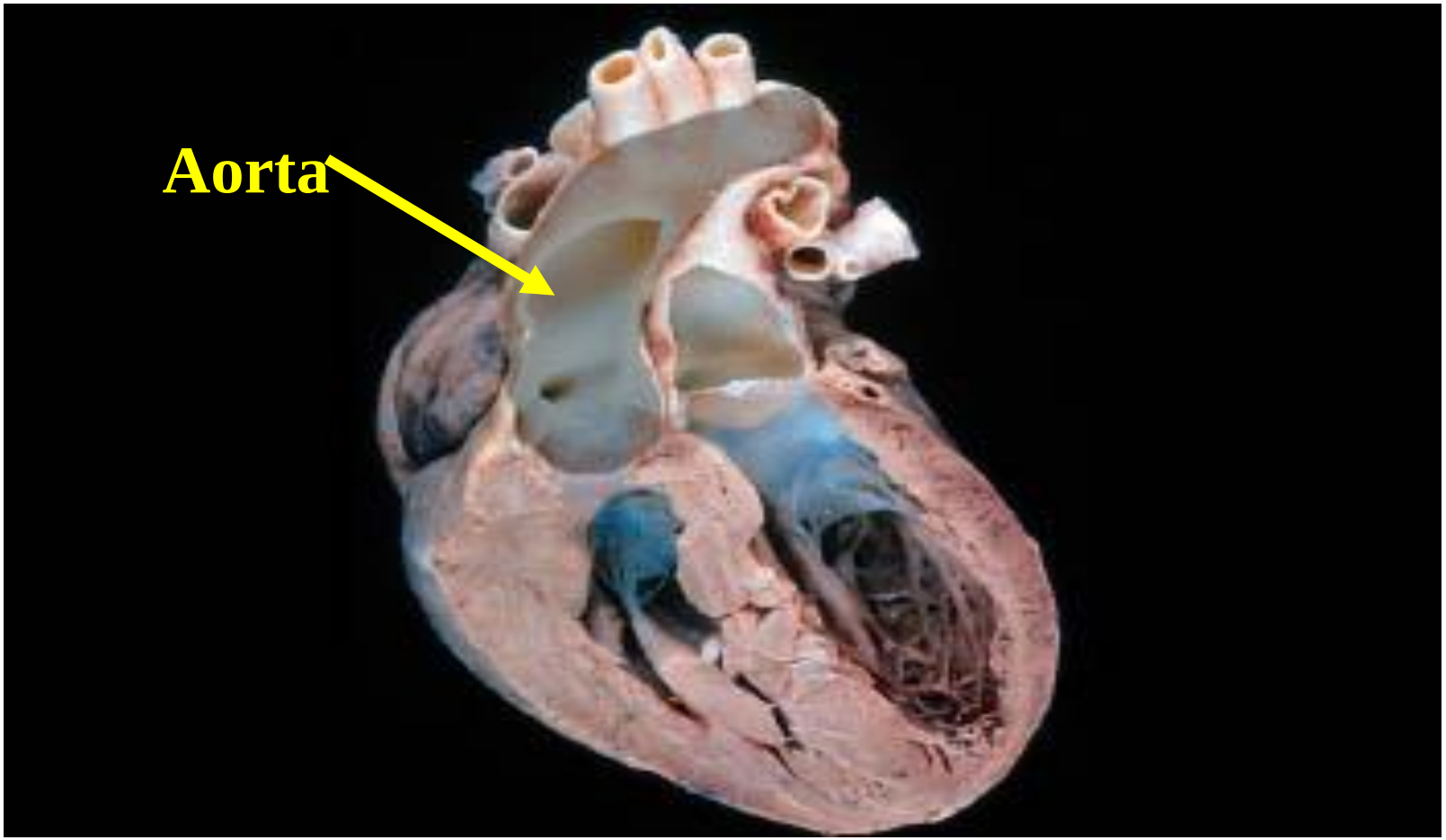
Transposition of the great arteries



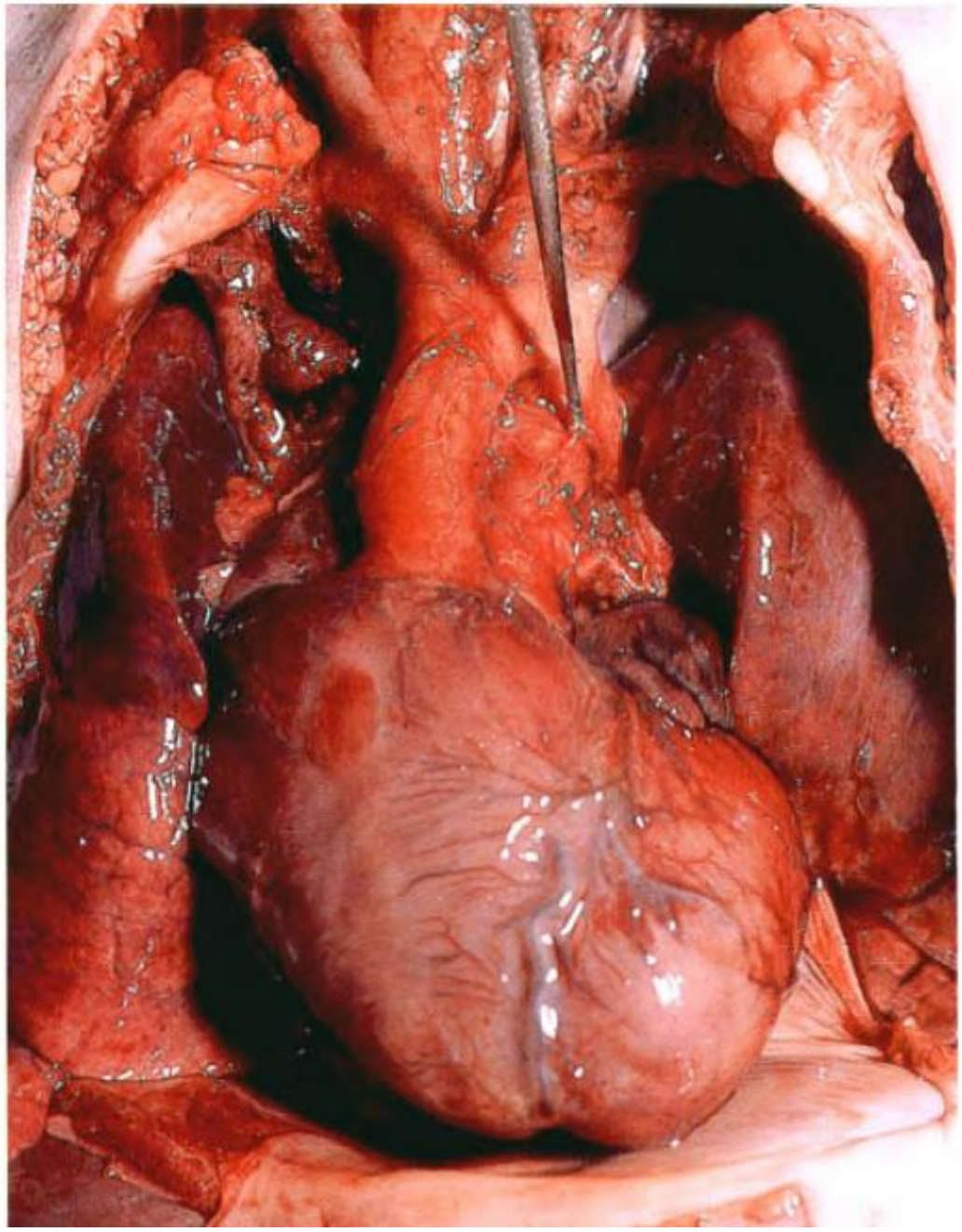
Also known as transposition of the great vessels.
No cardiomegaly is present. No edema in lung fields.

https://radiopaedia.org/cases/transposition-of-great-arteries-1?lang=us

Transposition of the great arteries



Schoen, F and Mitchell, RN, “The Heart,” in Kumar, V, Abbas, AK, Aster, JC (eds), Robbins and Cotran The Pathologic Basis of Disease (9th ed.), 2015. Elsevier. Philadelphia. Fig. 12-7 (Courtesy William D. Edwards, MD, Mayo Clinic, Rochester, Minn.)



Transposition of the great arteries

Neonatal death. The aorta arises from the right ventricle and the pulmonary artery from the left ventricle. The pointer is on the ductus arteriosus.

Sheppard, MN, Herrington, CS, “The Cardiovascular System,” in Herrington, CS (ed), Muir’s Textbook of Pathology, 15th edition. 2014. CRC Press. Boca Raton, Florida. Fig. 1.61

Pulmonary atresia with intact ventricular septum

- Completely different lesion from pulmonary atresia with VSD.
- Symptoms depend on degree of pulmonary blood flow.
- Cyanosis at birth.
- Pulmonary blood flow is always ductal dependent with rare aortopulmonary collateral arteries being present.

Pulmonary atresia with intact ventricular septum

- RV-dependent coronary arteries sometimes are present.
- A blowing systolic murmur resulting from the associated PDA may be heard at the pulmonary area.
- A holosystolic murmur is often heard at the lower left sternal border, as many children develop tricuspid insufficiency
- Echocardiography is diagnostic

Tricuspid atresia

- Marked cyanosis present from birth.
- The entire systemic venous return must flow through the atrial septum (either ASD or PFO) to reach the left atrium.
- The left atrium thus receives both the systemic venous return and the pulmonary venous return.
- Complete mixing occurs in the left atrium
- EKG with left-axis deviation, right atrial enlargement, and LVH
- Echocardiography is diagnostic

Tricuspid atresia

- In infants with diminished pulmonary blood flow, PGE_1 is given until an aortopulmonary or Blalock shunt can be performed.
- A Glenn procedure (SVC to pulmonary artery anastomosis) is done with takedown of the aortopulmonary/Blalock shunt at 4–6 months when saturations begin to fall
- Completion of the Fontan procedure (redirection of IVC and SVC to pulmonary artery) is performed when the child reaches around 15 kg.
- May live into the 20s

Hypoplastic left heart syndrome

- Mild cyanosis at birth.
- Minimal auscultatory findings.
- Stenosis or atresia of the mitral and aortic valves is the rule.
- In the neonate, survival depends on a PDA as it is the only source of blood to the aorta and the coronary arteries
- Rapid onset of shock with ductal closure.
- Echocardiography is diagnostic

Hypoplastic left heart syndrome

- In the Norwood procedure, the relatively normal main pulmonary artery is transected and connected to the small ascending aorta.
- The entire aortic arch must be reconstructed due to its small size.
- Then, either a Blalock shunt from the subclavian artery to the pulmonary artery or a Sano shunt from the RV to the pulmonary artery must be created to restore pulmonary blood flow.

Hypoplastic left heart syndrome

- Children who have a Norwood procedure will later require a Glenn anastomosis of the SVC to pulmonary artery with takedown of the systemic-pulmonary shunt (at age 6 months)
- Later, a Fontan procedure that connects the IVC to pulmonary artery, completing the systemic venous bypass of the heart (at age 2–3 years).
- 1 year survival without transplant is 70%

Total anomalous pulmonary venous return

- Abnormal pulmonary venous connection leading to cyanosis.
- Occurs with or without a murmur and may have accentuated P_2.
- Right atrial enlargement and RVH.
- The veins empty into a confluence that usually is located behind the left atrium. That confluence drains into the systemic venous circulation.
- Complete mixing of systemic and pulmonary venous blood occurs at the level of the right atrium.

Total anomalous pulmonary venous return

- Pulmonary veins drain into right atrium or other location instead of left atrium.
- Must have ASD or patent foramen ovale for survival.
- Types
- Supracardiac
- Most common
- Pulmonary vein into superior vena cava or innominate
- Intracardiac:
- Pulmonary vein into coronary sinus or right atrium.

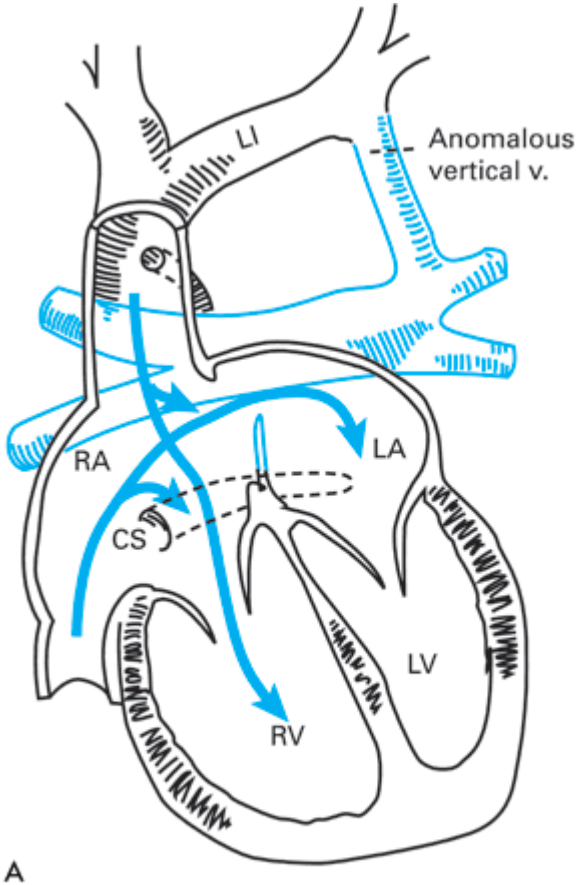
Total anomalous pulmonary venous return

- Subdiaphragmatic:
- Pulmonary vein into inferior vena cava, portal vein, ductus venosus, or hepatic vein.
- If obstructed, a surgical emergency

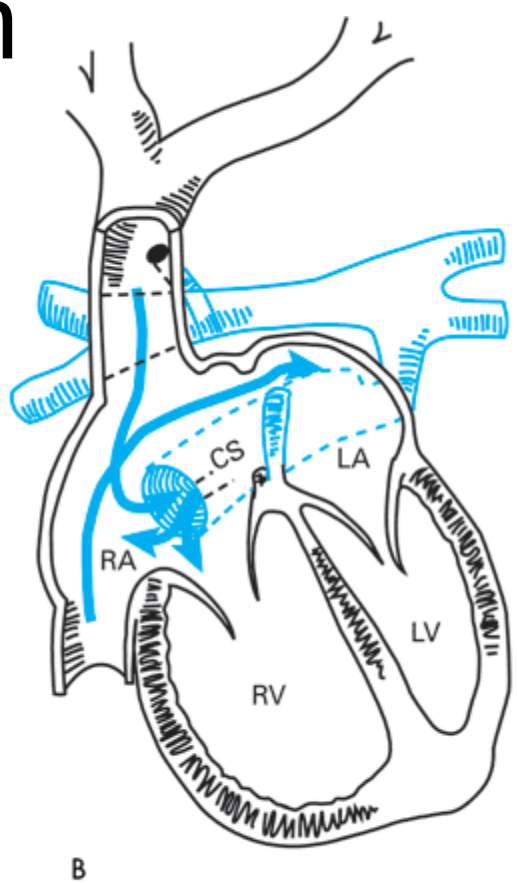
Total anomalous pulmonary venous return

- There is a hyperdynamic right ventricular impulse.
- S2 is fixed and widely split.
- Systolic ejection murmur at the left upper sternal border and a mid-diastolic rumble at the left lower sternal border
- Right axis deviation and right ventricular hypertrophy are noted on EKG.
- The chest x-ray shows cardiomegaly and increased pulmonary vascular markings.
- Definitive diagnosis is made by echocardiography.

Total anomalous pulmonary venous return

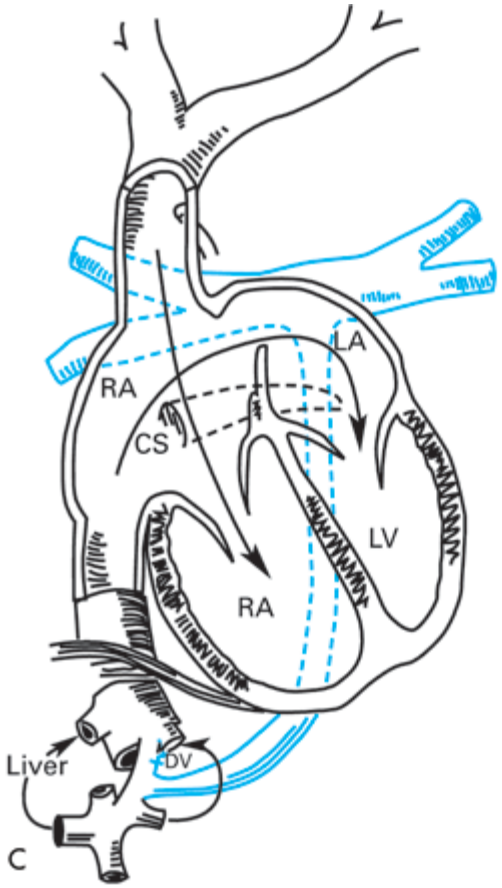


A



B

Total anomalous pulmonary venous return



Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

A. Total anomalous pulmonary venous connection to the left brachiocephalic (innominate) vein (LI).
B. Total anomalous pulmonary venous connection to the coronary sinus (CS).
C. Total anomalous pulmonary venous connection of the infradiaphragmatic type to the ductus venosus (DV).

LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle

Fig. 82-34 Accessed 04/01/2010

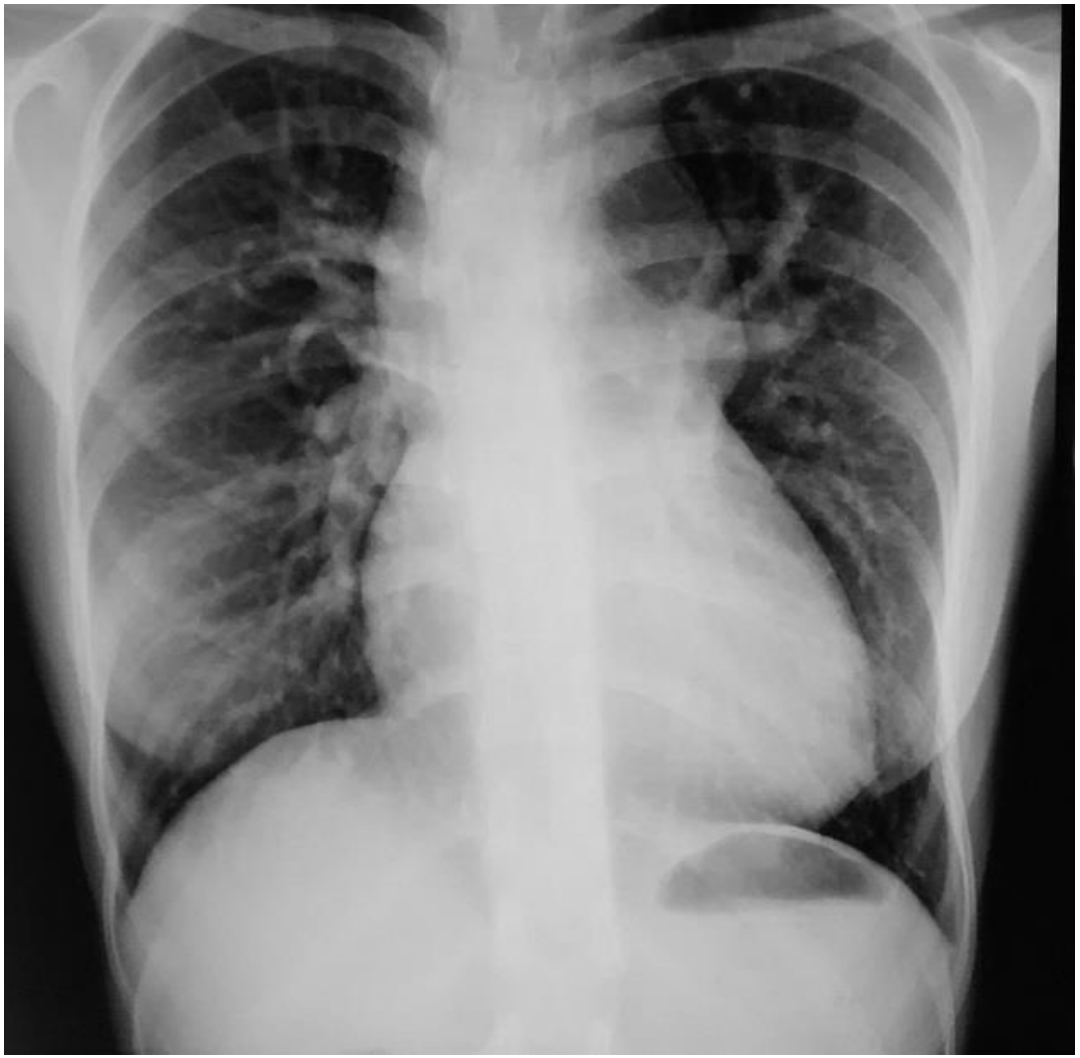
Total anomalous pulmonary venous return

- A right-to-left shunt must be present at the atrial level, either as an ASD or a PFO.
- If the atrial septum is restrictive, balloon septostomy is needed at birth to allow filling of the left heart.
- RV heave is palpable
- P_2 is increased.
- Systolic and diastolic murmur may be heard as a result of increased flow across the pulmonary and tricuspid valves

Total anomalous pulmonary venous return

- Chest x-ray reveals cardiomegaly involving the right heart and pulmonary artery.
- Pulmonary vascular markings are increased.
- EKG shows right axis deviation
- Echocardiography/Doppler is diagnostic
- May require heart-lung transplant

Total anomalous pulmonary venous return (supracardiac)



The paratracheal shadow on the right is the prominent superior vena cava, and on the left is the vertical vein. The innominate vessel lies in the midline above base of heart. These three prominent vessels together form the head of the 'snowman'. The body is formed by the rest of the heart. Also described as a figure eight.

ACYANOTIC CONGENITAL HEART DISEASE

Left-to-right shunts

- A left-to-right shunt increases the amount of blood delivered to the right side of the heart and will result in hypertrophy and dilation of the right atrium or right ventricle (or both), depending upon the type of shunt.
- Eventually, the pressure in the right side of the heart increases and surpasses that in the left side of the heart, resulting in a reversal of the shunt from left-to-right to a right-to-left shunt.
- This change is called Eisenmenger syndrome (and results in right ventricle enlargement).

Ventricular Septal Defect

- Most common abnormality found with congenital heart disease.
- 90% occur in membranous septum.
- 85% close spontaneously, particularly those in muscular septum.
- Wide splitting of S_2.
- Holosystolic murmur.
- Secondary pulmonary hypertension because of high-pressure of shunted blood.
- Left ventricular hypertrophy (to biventricular hypertrophy) and left atrial enlargement on EKG.

Ventricular septal defect

- A restrictive VSD is small and makes a louder murmur than an unrestricted one, often with an accompanying thrill.
- The higher the gradient across the septum, the smaller the left-to-right shunt.
- Small defects may be asymptomatic.
- Larger defects result in pulmonary hypertension (Eisenmenger physiology) if the pulmonary circuit is not protected by RV outflow tract obstruction.
- Echocardiography/Doppler is diagnostic.

Ventricular septal defect

- Most common congenital heart defect
- Four types:
- Type A, the outflow tract VSD lies underneath the semilunar valves
- Type B, the membranous VSD has three variations
- Type C, the inlet VSD is present below the tricuspid valve and often part of the AV septal defect
- Type D is the muscular VSD

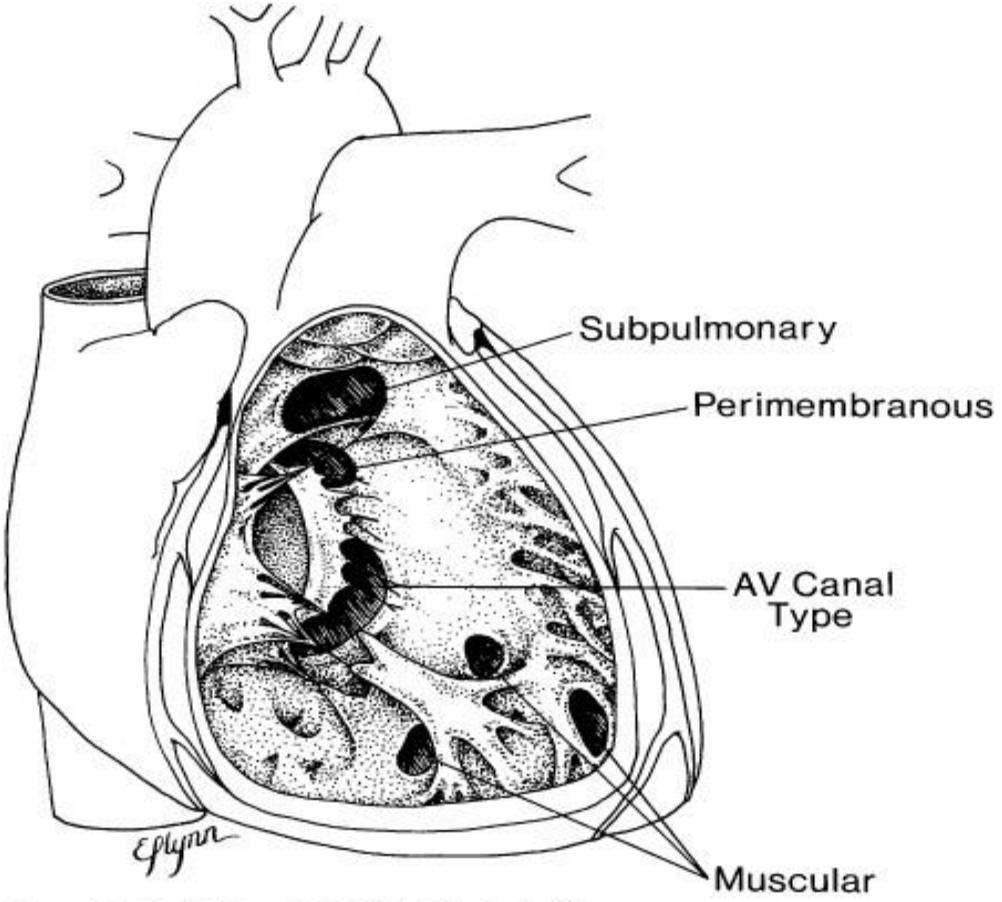
Ventricular septal defect

- A small or restrictive VSD diameter is less than 25% of the aortic root diameter
- A moderately restrictive VSD diameter is 25–75% of the aorta
- An unrestricted VSD size is greater than 75% of the aortic diameter.

Ventricular septal defect

- Endocarditis and paradoxical embolus as complications.
- Eisenminger's syndrome noted with reversal of shunt and late development of cyanosis
- Prompt surgical closure of defects is recommended in all individuals older than the age of 2 years if the pulmonary arterial systolic pressure is greater than half the systemic arterial systolic pressure, the mean pulmonary pressure exceeds 25 mmHg, or the R_p/R_sratio is higher than 0.3:1.

Ventricular septal defect

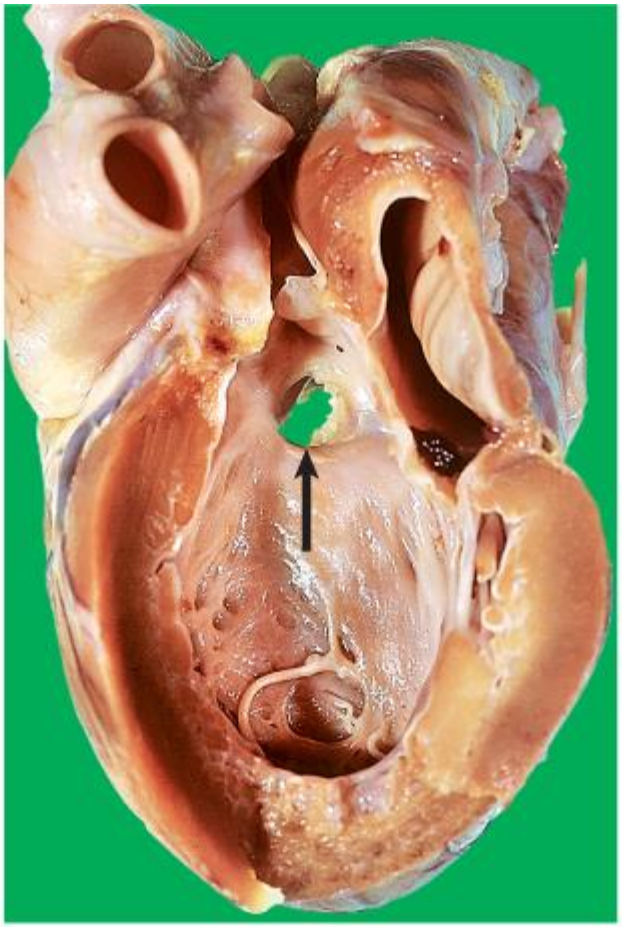


Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

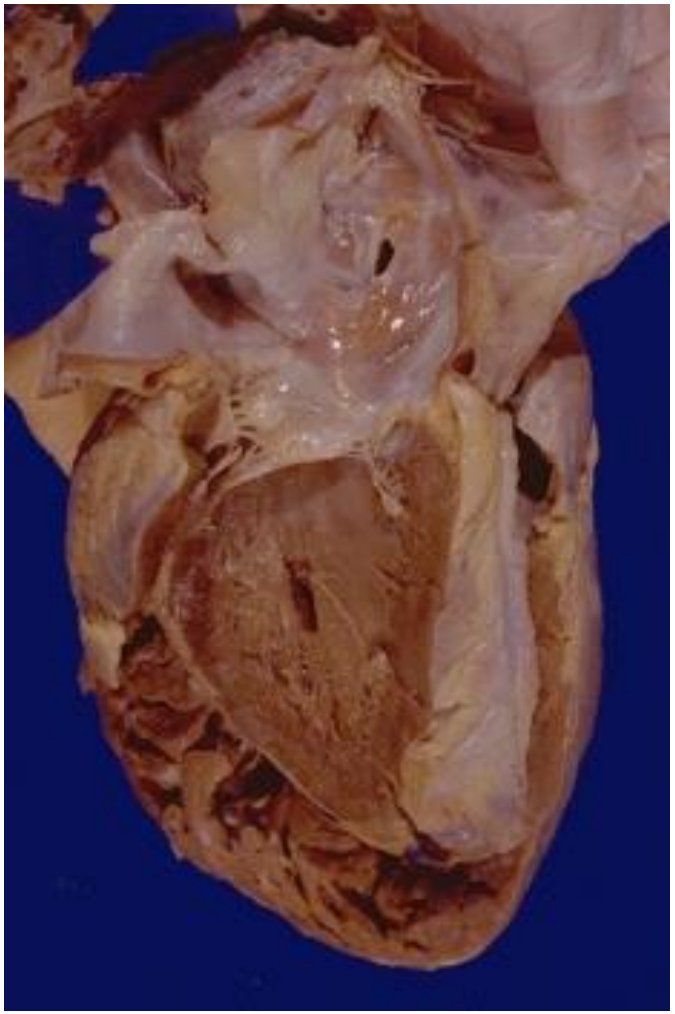
Fig. 82-5 Accessed 04/01/2010

Source: From Freed MD. Fetal and transitional circulation. In: Fyler DC, ed. Reproduced with permission from the publisher and author.

Ventricular septal defect (membranous)



Ventricular septal defect (muscular)



Here is a heart with both an atrial septal defect (ASD) and a muscular ventricular septal defect (VSD). The heart is opened on the left side.

Minimal shunting present.

Significant risk for infective endocarditis.

https://webpath.med.utah.edu/CVHTML/CV085.html

Ventricular septal defect

- Membranous and muscular septal defects may spontaneously close in childhood as the septum grows and hypertrophies.
- 80-85% of cases
- A left-to-right shunt is present to the degree of associated RV pressure.
- The smaller the defect, the greater is the gradient from the LV to the RV and the louder the murmur.
- Holosystolic murmur in the left 3^{rd}-4^{th} interspaces along the sternum
- Thrill is common.

Testing

- The EKG may be normal or may show right, left, or biventricular hypertrophy depending on the size of the defect and the PVR.
- With large shunts, the LV, the LA, and the pulmonary arteries are enlarged and pulmonary vascularity is increased on chest x-ray.
- Echocardiography can demonstrate the size of the overloaded chambers and can usually define the defect anatomy.
- Doppler can qualitatively assess the magnitude of shunting

Outcomes

- Patients with a small VSD have a normal life expectancy except for the small risk of infective endocarditis.
- With large VSD shunts, heart failure may develop early in life, and survival beyond age 40 years is unusual without intervention
- Larger defects result in pulmonary hypertension (Eisenmenger physiology) if the pulmonary circuit is not protected by RV outflow tract obstruction.

Treatment

- Surgical repair is low risk
- Pulmonary hypertension may be treated medically
- All patients who have a right-to-left shunt present should have filters placed on any intravenous lines to avoid any contamination or air bubbles from becoming systemic.

Atrial septal defect

- Often asymptomatic and discovered on routine physical examination.
- With an atrial septal defect (ASD) and left to right shunt:
- RV lift
- S_2 widely split and fixed.
- Systolic ejection murmur over the pulmonary area
- Echocardiography demonstrates evidence of RA and RV volume overload.
- Associated with fetal alcohol syndrome

Atrial septal defect

- Second most common cause of left-to-right shunt.
- Most commonly occur at the fossa ovalis (secundum type) 90%
- May also occur below the fossa ovalis (primum type).
- It is rare to see the lesion near the superior vena cava (sinus venosus type).
- Widely split and fixed S_2 because of increased venous pressure and delayed closure of pulmonic valve.
- Systolic ejection murmur at upper left sternal border.

Atrial septal defect

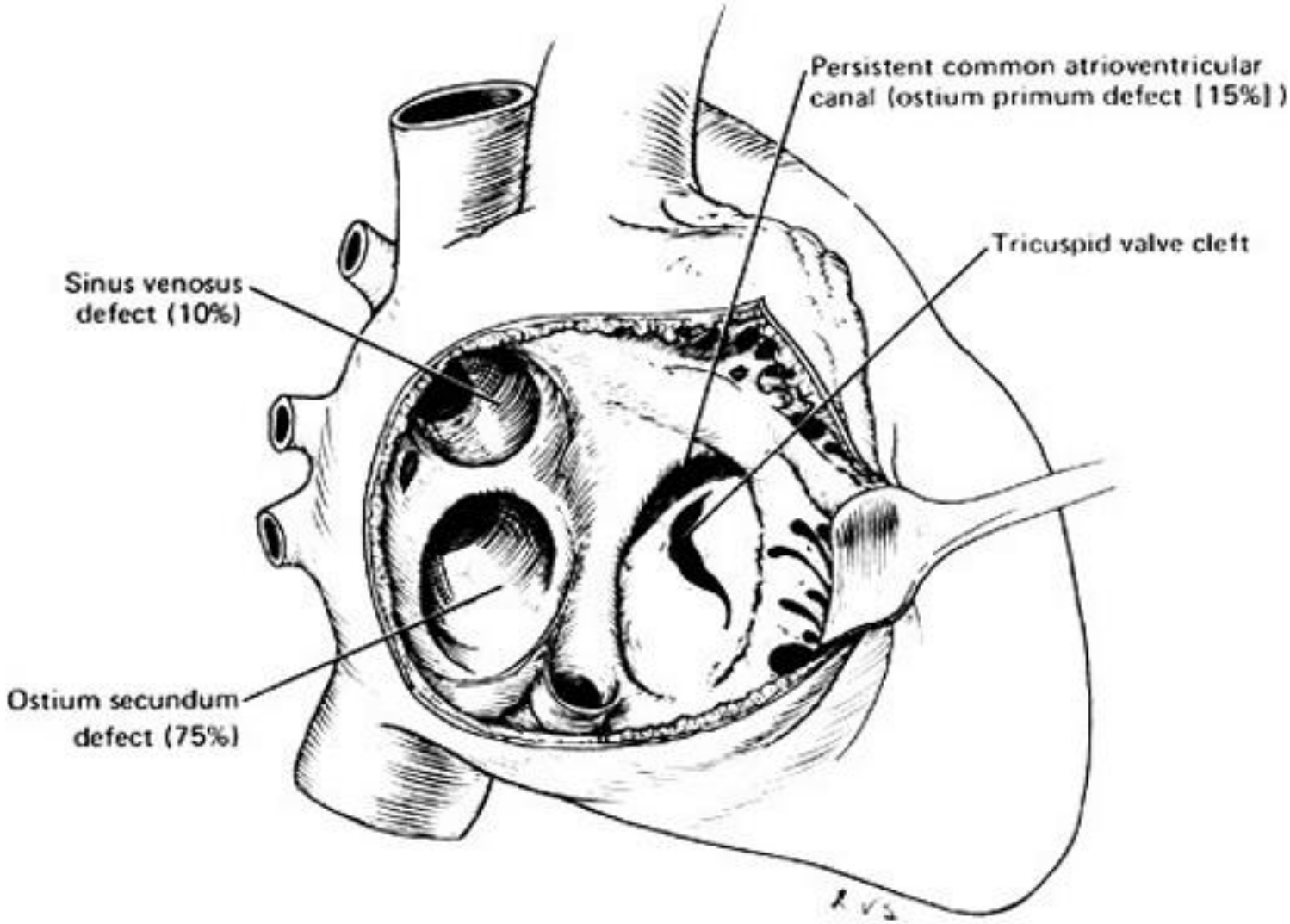
- ASD is found in approximately 6 percent of children who survive beyond the first year of life with congenital heart disease.
- If one excludes mitral valve prolapse and a congenitally bicuspid aortic valve, it is the most common form of congenital heart disease among adults.
- May be asymptomatic before adulthood
- Right atrial enlargement but little pulmonary hypertension noted as shunt is low pressure.
- Right axis deviation on EKG.

Atrial septal defect

- A septum primum defect involves both the atrial septum and the atrioventricular valve ring, so the membranous portion of the interventricular septum is involved as well.
- A mitral valve defect is often associated with this type of ASD because of the associated defect in the AV ring.

Embryology

- Ostium primum defect associated with atrioventricular septal defects
- Ostium secundum defect usual in ASD
- Females 2:1
- May be associated with Holt-Oran syndrome
- Sinus venosus defect frequently associated with abnormal pulmonary venous return
- The location of the sinus venosus is intimately related to the right upper pulmonary vein.



Source: Maxine A. Papadakis, Stephen J. McPhee, Michael W. Rabow: Current Medical Diagnosis and Treatment 2020

Physiology

- Normally oxygenated blood from the higher-pressure LA shunts into the RA
- RV output and pulmonary blood flow increase.
- Pulmonary pressure is elevated.
- As the RV compliance worsens from the chronic volume overload, the RA pressure may rise and the degree of left-to-right shunting may decrease

Physiology

- If the shunt reverses, cyanosis appears.
- The major factor in the direction of shunt flow is the compliance of the respective atrial chambers.
- 15%, severe pulmonary hypertension with cyanosis (Eisenmenger physiology)

Complications

- 25% of population have a PFO
- Paradoxical emboli more common with a PFO than with and ASD
- Suspected if cryptogenic stroke before 55 years of age
- Increased shunting in patients with a PFO and sleep apnea as the RA compliance may worsen during apneic spells when pulmonary pressures increase.

Clinical diagnosis

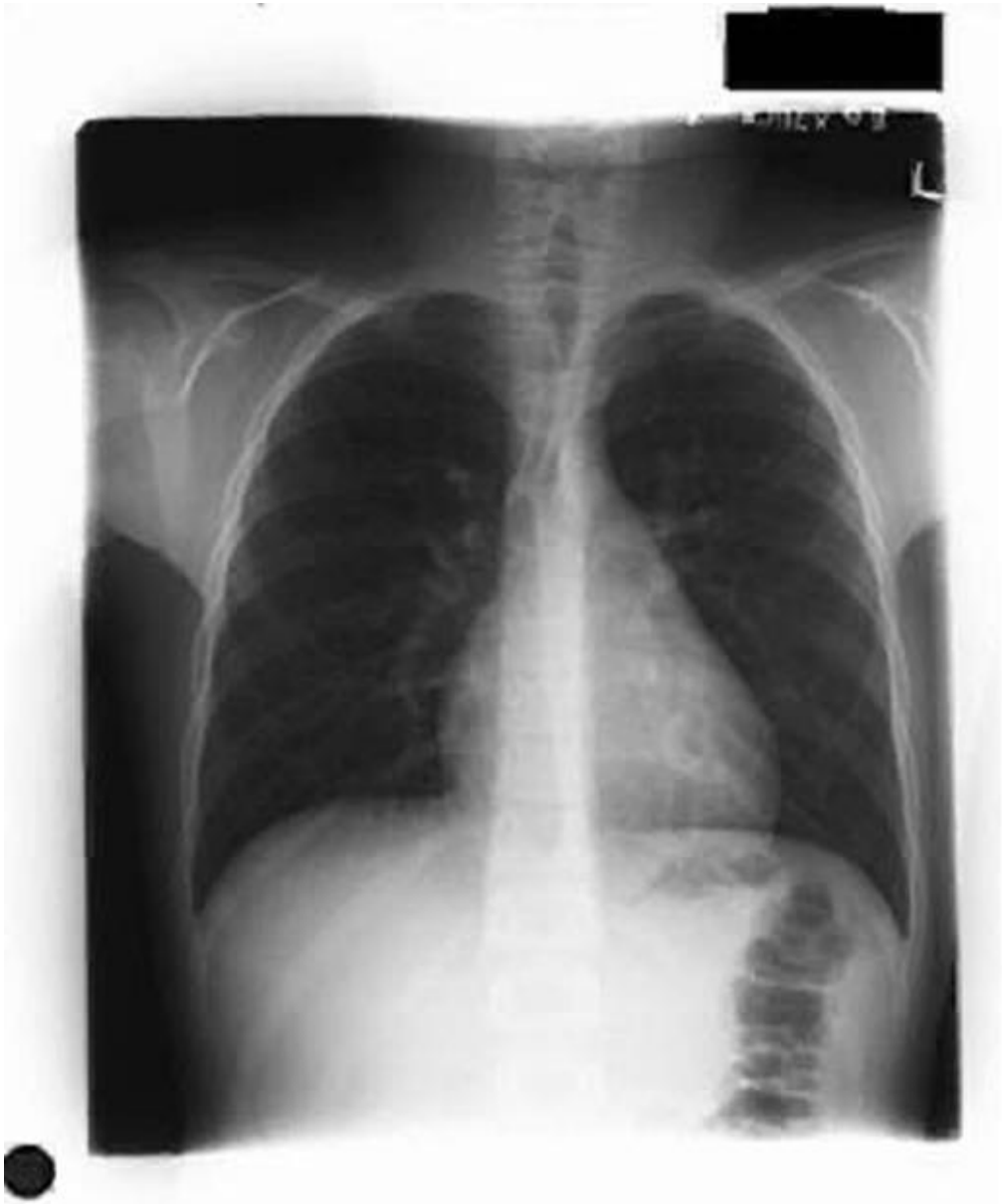
- Patients with a small or moderate ASD or with a PFO are asymptomatic unless a complication occurs.
- There is only trivial shunting in a PFO unless the RA pressure increases or the atrial septum is distorted.
- With larger ASD shunts, exertional dyspnea or heart failure may develop
- Commonly in the fourth decade of life or later

Clinical diagnosis

- Prominent RV and PA pulsations are readily visible and palpable with significant disease
- Systolic ejection murmur in the 2nd-3rd intercostal spaces parasternally
- Decreases in intensity on inspiration (increased RA pressure)
- Increases in intensity on expiration (decreased RA pressure)
- Fixed split of S2.
- Large shunts have diastolic flow murmur at the left lower sternal border (tricuspid valve)

Testing

- Right axis deviation or RVH may be present depending on degree of RV overload
- Incomplete or complete right bundle branch block (RBBB) on EKG
- Superior axis deviation and complete heart block noted in complete AV defect
- The chest radiograph shows large pulmonary arteries, increased pulmonary vascularity, an enlarged RA and RV, and a small aortic knob



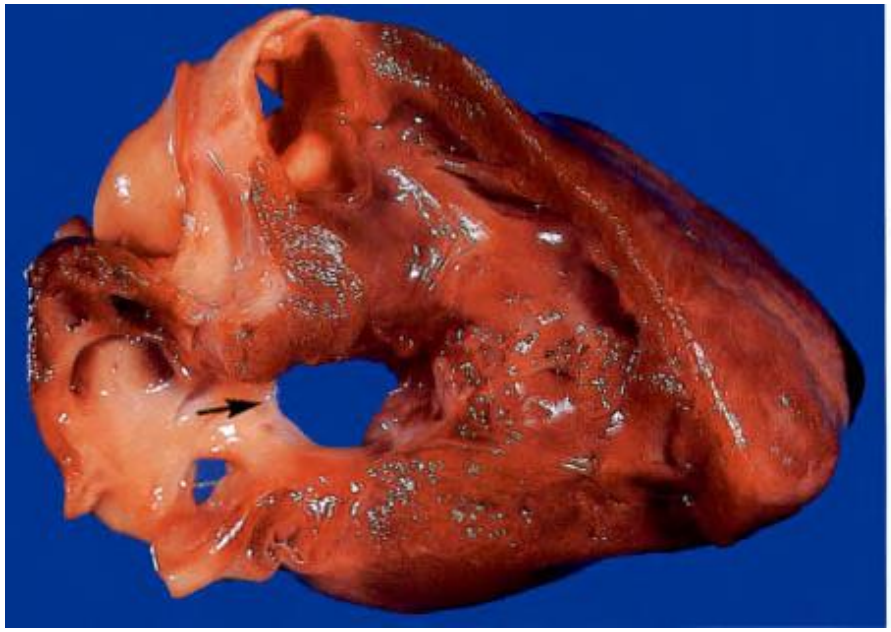
Testing

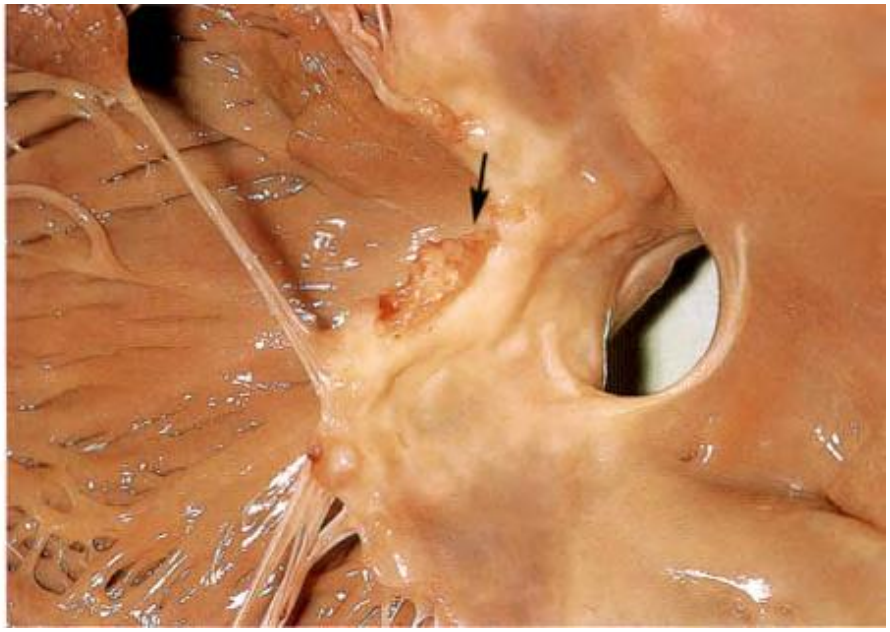
- Echocardiography demonstrates evidence of RA and RV volume overload.
- The atrial defect is usually observed by echocardiography
- Cardiac catheterization can define the size and location of the shunt and determine the pulmonary pressure and PVR

Outcomes

- Patients with small atrial shunts live a normal life span with no intervention.
- Large shunts usually cause disability by age 40 years.
- Left-to-right shunts and RV overload tend to increase with normal age-related reduction in LV (and subsequently LA) compliance.
- ASDs should be closed if there is evidence of an RV volume overload regardless of symptoms.

Atrial septal defect

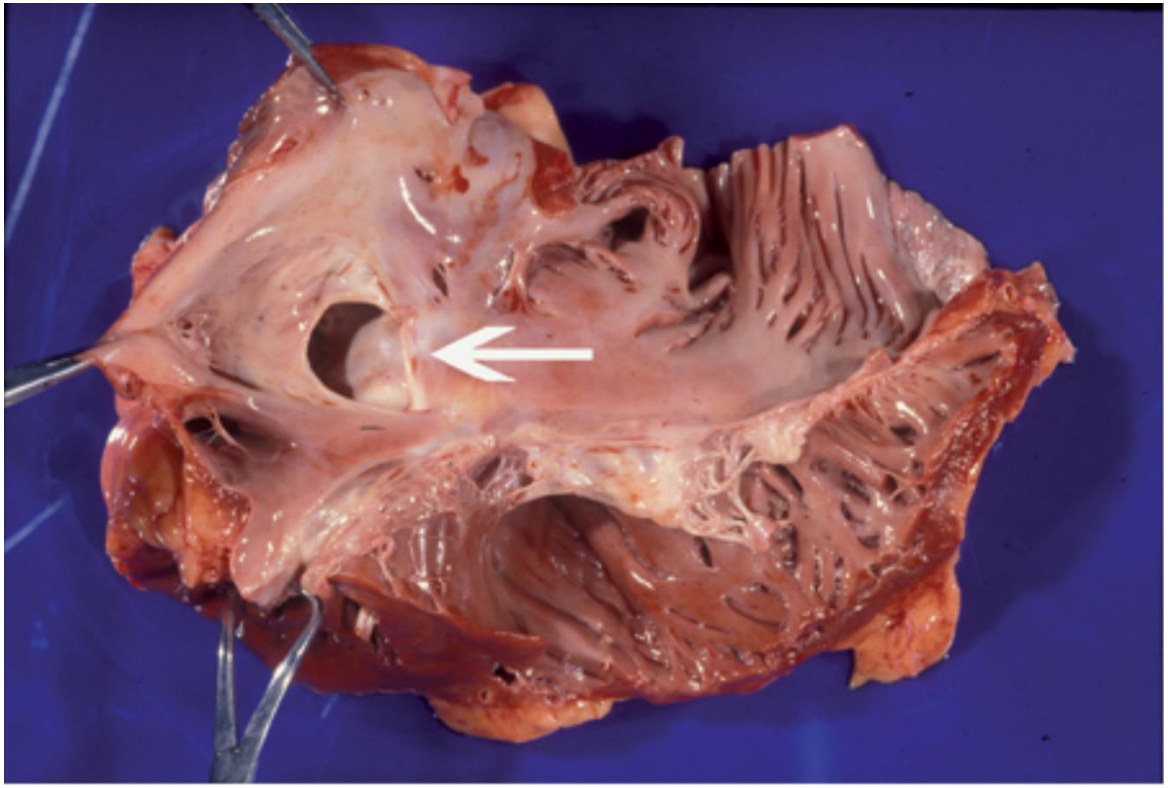




Left. Septum primum atrial septal defect viewed from the right atrium.
Right. Septum primum atrial septal defect plus a cleft in the anterior cusp of the mitral valve. There is a small vegetation on the mitral valve.

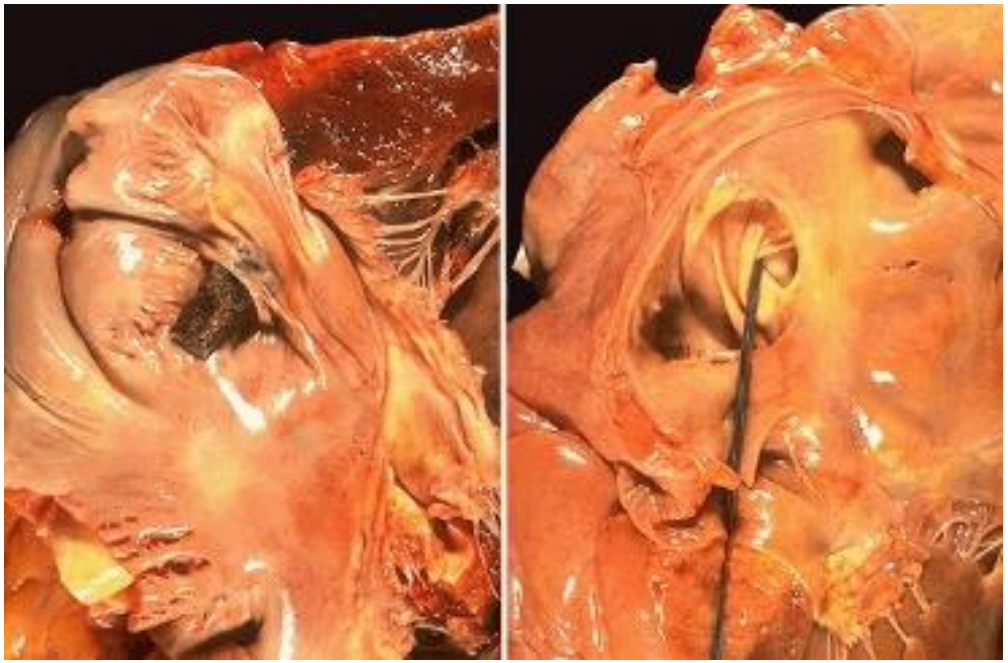
Sheppard, MN, Herrington, CS, "The Cardiovascular System," in Herrington, CS (ed), Muir's Textbook of Pathology, 15th edition. 2014. CRC Press. Boca Raton, Florida. Figs. 1-68 and 1-69

Atrial septal defect



Source: Kemp WL, Burns DK, Brown TG: *Pathology: The Big Picture*: www.accessmedicine.com

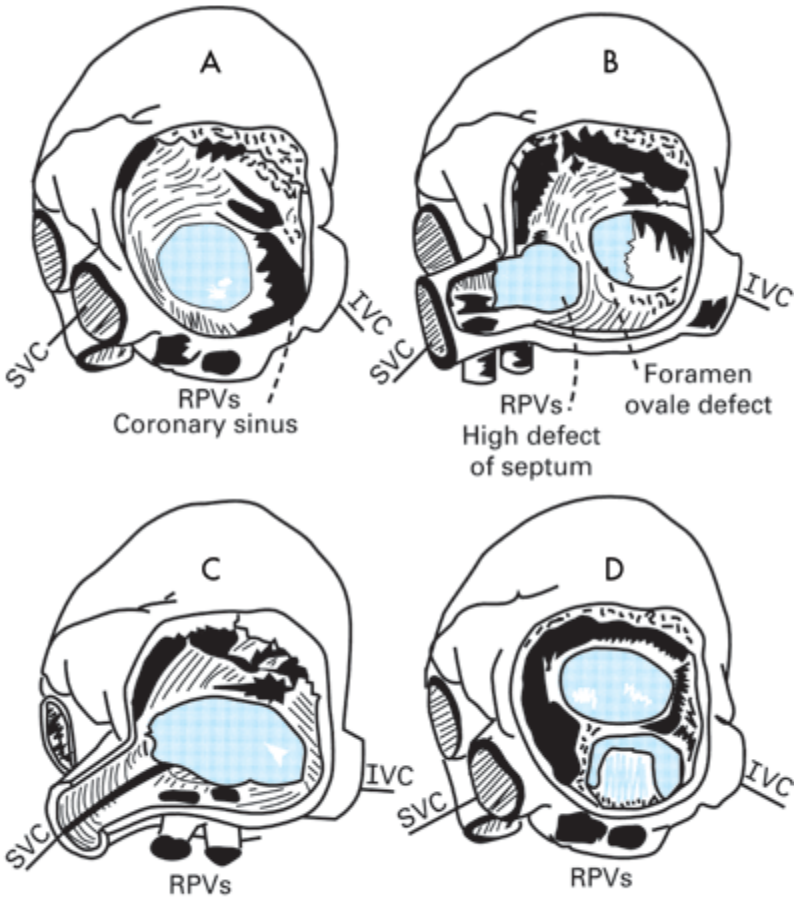
Patent foramen ovale with paradoxical embolus



https://webpath.med.utah.edu/CVHTML/CV115.html

At the right is a probe patent foramen ovale in an adult. A metal probe lifts the septum secundum and reveals the opening. Normally, the left atrial pressure keeps the foramen closed, but if right atrial pressures rise with pulmonary hypertension (as with pulmonary embolus), the foramen may open and even allow a thrombus to go from right to left.

Types of intra-atrial communications



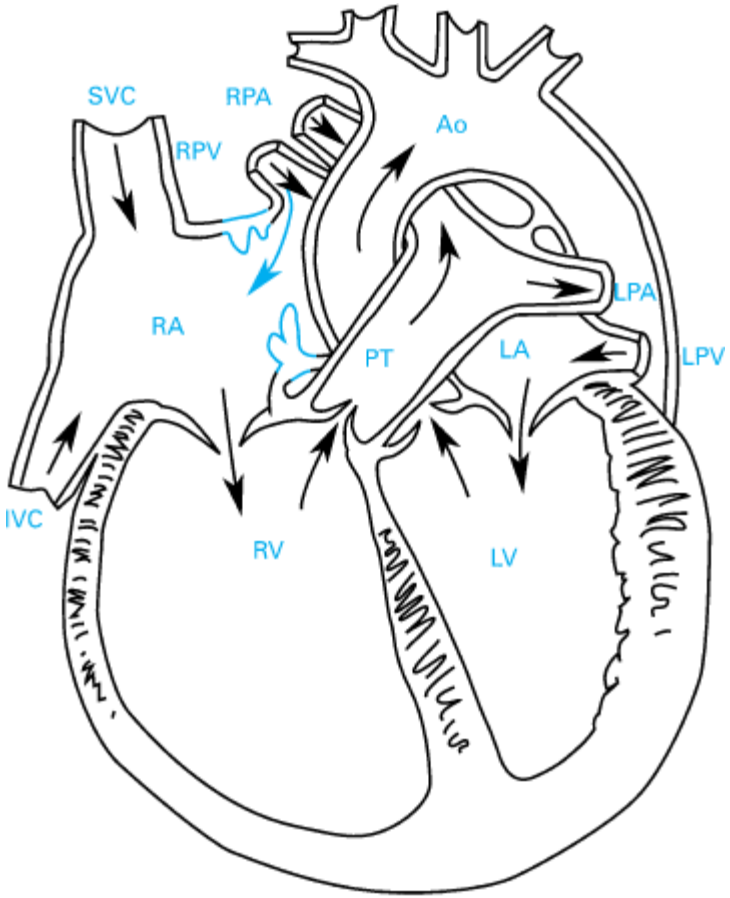
A. Ostium secundum defect.
B. Sinus venosus type of defect.
C. Ostium secundum defect with absence of the posterior rim.
D. Partial form of common atrioventricular canal with cleft mitral valve.

IVC, inferior vena cava; SVC, superior vena cava; RPV, right pulmonary veins.

Source: From Lewis FJ, Winchell P, and Bashour FA.[67] Copyright 1957, American Medical Association. Reproduced with permission from the publisher and authors.

Fig. 82-8 Accessed 04/01/2010

Atrial septal defect at fossa ovalis with left-to-right shunt



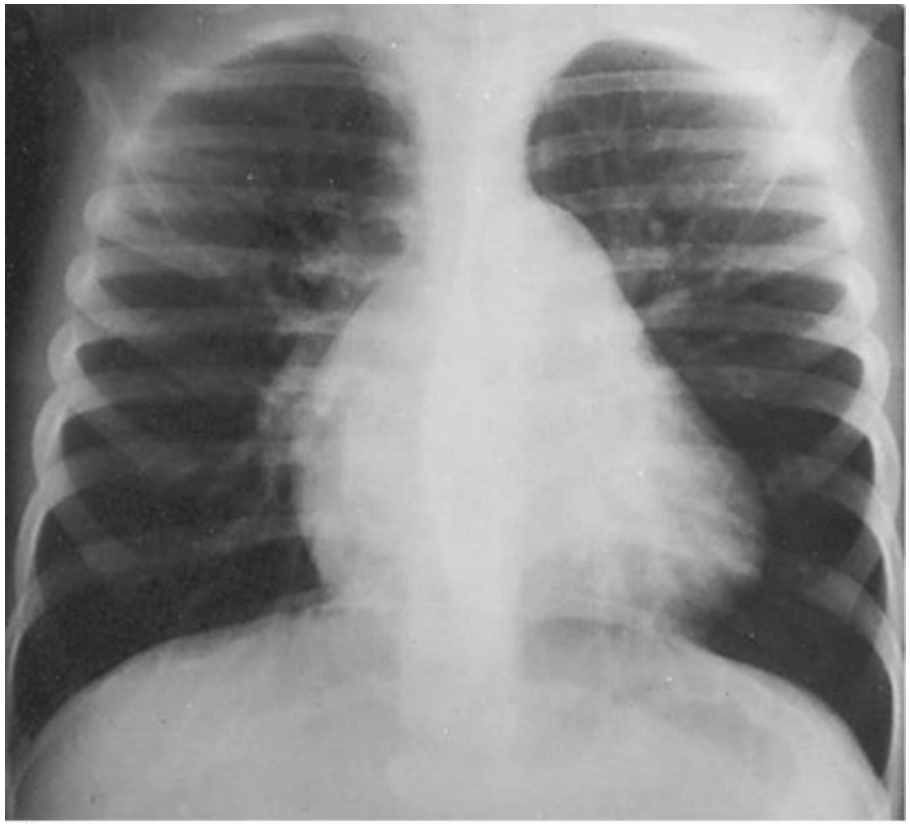
Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

Ao, aorta; IVC, inferior vena cava; LA, left atrium; LPA, left pulmonary artery; LPV, left pulmonary vein; LV, left ventricle; PT, main pulmonary arterial trunk; RA, right atrium; RPA, right pulmonary artery; RPV, right pulmonary vein; RV, right ventricle; SVC, superior vena cava.

Source: From Edwards JE. Classification of congenital heart disease in the adult. In: Roberts WC, ed. *Congenital Heart Disease in Adults*. Philadelphia: Davis; 1979:1. Reproduced with permission from the publisher and author.

Fig. 82-9 Accessed 04/01/2010

Atrial septal defect (secundum) with left-to-right shunt

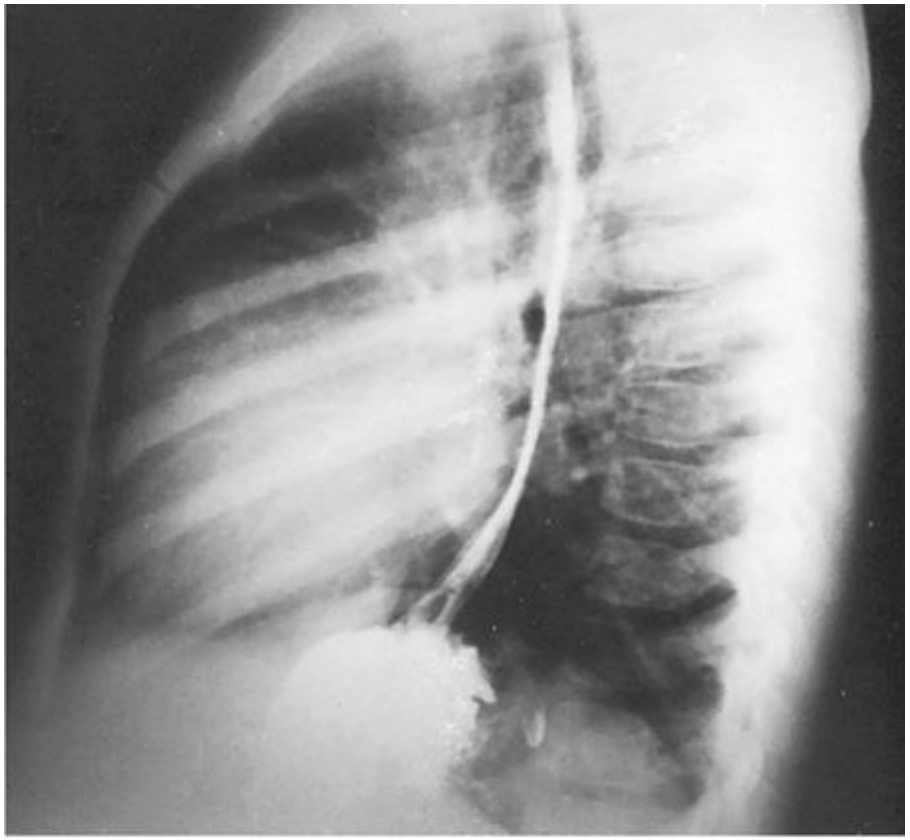


A

Prominence of the main pulmonary arterial segment and increased blood flow. No left atrial dilation is present.

Fig. 82-10 Accessed 04/01/2010

Atrial septal defect (secundum) with left-to-right shunt



B

B. Lateral view. Right ventricular enlargement accompanies prominence of the main pulmonary arterial segment and increased blood flow. No left atrial dilation is present.

Fig. 82-10 Accessed 04/01/2010

Patent ductus arteriosus

- Embryonic ductus arteriosus allows shunting of blood from the PA to the aorta in utero (thus bypassing the lungs)
- Kept patent by circulating prostaglandins
- Spontaneous closure of the ductus arteriosus in response to an increase in P_aO_2 associated with breathing normally occurs within the first 12 hours of life.
- All right heart blood flows to the pulmonary arteries.
- Failure to close results in a persistent left-to-right shunt connecting the left PA and aorta, usually near the origin of the left subclavian artery
- Females 2:1

Patent ductus arteriosus

- Increase incidence in those born at altitudes >10,000 feet (3,000m)
- A small ductus may be well tolerated until adulthood.
- There are no symptoms unless LV failure or pulmonary hypertension develops.
- The hands appear normal while the toes are cyanotic and clubbed (differential cyanosis).
- Neonates with a large ductus present with tachypnea and diaphoresis with feeding

Patent ductus arteriosus

- Loud S_2
- The classic sign of a persistent PDA is a continuous “machine-like” murmur at left upper sternal border. May develop left (or bi) ventricular hypertrophy.
- Use of prostaglandin E will keep the shunt open.
- Use of indomethacin will close a PDA.
- Banding the PDA is a surgical approach.
- If the ductus is large enough, pulmonary hypertension (Eisenmenger physiology) may occur

Testing

- LVH may be found on EKG
- On chest x-ray, the PA, aorta, and LA are prominent because they are in the shunt pathway
- Cardiac MRI and CT are the best noninvasive modalities to demonstrate the ductus size and shape and to assess the size of the pulmonary arteries
- Cardiac catheterization can establish the shunt size and direction

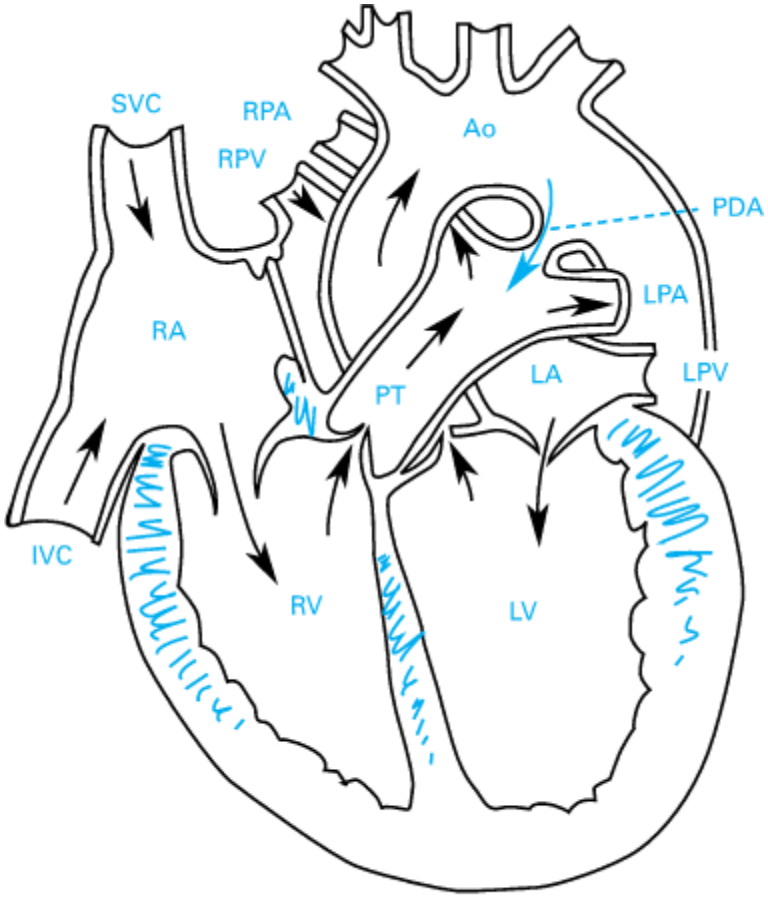
Outcomes

- Large shunts cause a high mortality rate from cardiac failure early in life.
- Smaller shunts are compatible with long-term survival, heart failure being the most common late complication.
- Infective endocarditis or endarteritis may rarely occur
- Antibiotic prophylaxis for dental procedures continues to be recommended

Outcomes

- Surgical ligation of the patent ductus can be accomplished with excellent results
- Pulmonary hypertension treated with vasodilators

Patent ductus arteriosus



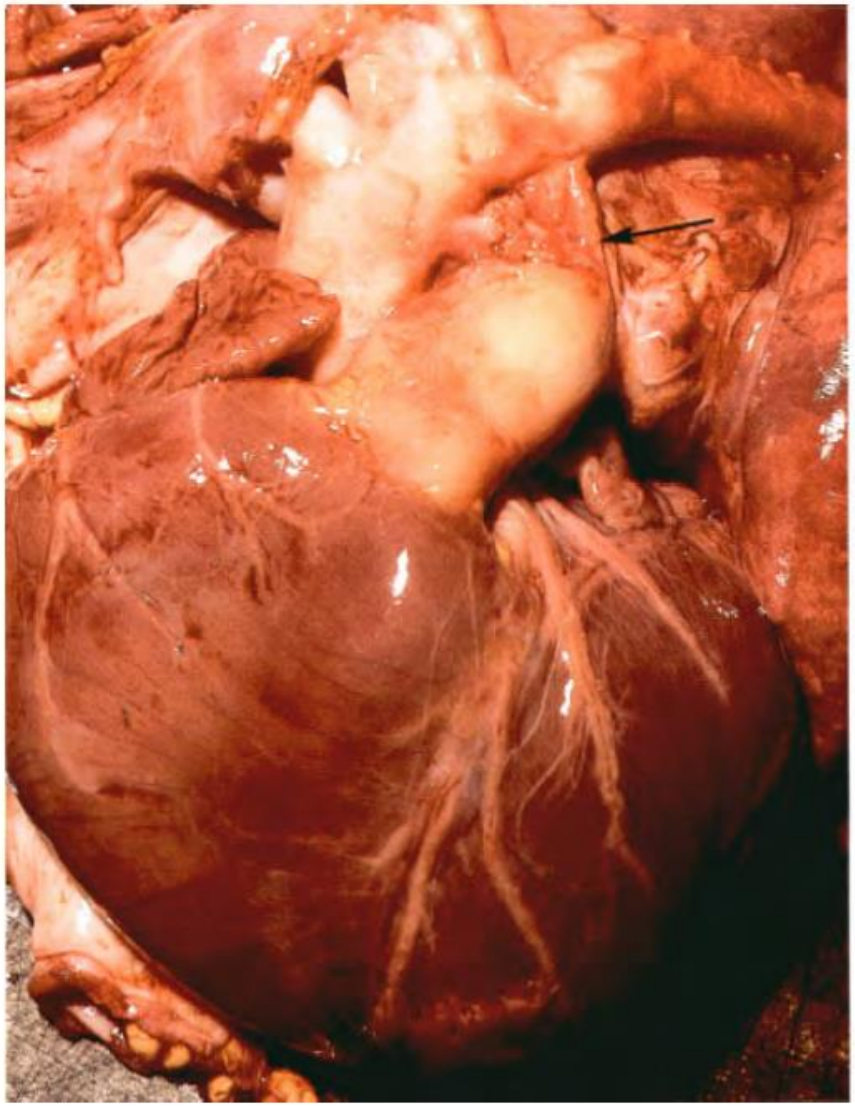
Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

Ao, aorta; IVC, inferior vena cava; LA, left atrium; LPA, left pulmonary artery; LPV, left pulmonary vein; LV, left ventricle; PT, main pulmonary arterial trunk; RA, right atrium; RPA, right pulmonary artery; RPV, right pulmonary vein; RV, right ventricle; SVC, superior vena cava.

Source: From Edwards JE. Classification of congenital heart disease in the adult. In: Roberts WC, ed. *Congenital Heart Disease in Adults*. Philadelphia: Davis; 1979:1. Reproduced with permission from the publisher and author

Fig. 82-17 Accessed 04/01/2010

Patent ductus arteriosus



Patent ductus arteriosus joining the pulmonary artery to the aorta (arrow).

Sheppard, MN, Herrington, CS, "The Cardiovascular System," in Herrington, CS (ed), Muir's Textbook of Pathology, 15th edition. 2014. CRC Press. Boca Raton, Florida. Fig. 1-60

Coarctation of the aorta

- Usual presentation is systemic hypertension.
- Delayed pulse in femoral artery compared to brachial artery.
- Systolic pressure is higher in upper extremities than in lower extremities
- Diastolic pressures are similar.

Coarctation of the aorta

- 50-80 % have bicuspid aortic valve.
- Associated with saccular cerebral aneurysms.
- One-third of patients with Turner's syndrome have coarctation.
- Part of congenital rubella syndrome.
- Preductal if before ligamentum arteriosum. (70%)
- Usually discovered in infancy.
- If patent ductus arteriosum is present, may see cyanosis of lower half of body.

Coarctation of the aorta

- Blowing systolic murmur in the back or left axilla
- Echocardiography/Doppler is diagnostic.
- A peak gradient of more than 20 mm Hg may be significant due to collaterals around the coarctation reducing gradient despite severe obstruction.

Coarctation of the aorta

- Coarctation may present in infants as congestive heart failure.
- May see right bundle branch block.
- Coarctation may present in children as hypertension with systolic ejection murmur at left upper sternal border (and between scapulae)
- Left ventricular hypertrophy on EKG
- If bicuspid aortic valve, ejection click at right upper sternal border.

Coarctation of the aorta

- Postductal if after ligamentum arteriosum.
- Clinically apparent in adolescence.
- Systemic hypertension with lower blood pressure in the arms rather than in the legs.
- May see rib notching caused by dilated collateral vessels.
- Arteries are absent below collateral connections

Post ductal coarctation of the aorta

- Blood pressure in upper limbs is greater than blood pressure in lower limbs.
- S_aO_2 difference > 5% between upper limb and lower limb.
- May have claudication in leg muscles
- Renovascular hypertension as a result of diminished renal artery pressure
- Increased cerebral arterial pressure may lead to saccular aneurysm rupture

Embryology

- Coarctation of the aorta consists of localized narrowing of the aortic arch just distal to the origin of the left subclavian artery
- Related to accessory ductal artery that contracts soon after birth
- Not usually present in the fetus though it may be associated with aortic root hypoplasia
- Pre-ductal, ductal, post-ductal locations

Physiology

- If the stenosis is severe, collateral circulation develops around the coarctation site through the intercostal arteries and the branches of the subclavian arteries
- The renin-angiotensin system is often abnormal as well
- 50-80% have bicuspid aortic valves
- 10% have saccular cerebral aneurysms
- Associated with Turner's syndrome

Clinical diagnosis

- If LV failure does not occur in infancy, there are usually no symptoms until the hypertension produces LV failure
- Strong arterial pulsations are seen in the neck and suprasternal notch.
- Hypertension is present in the arms, but the pressure is normal or low in the legs.
- This difference is exaggerated by exercise.

Clinical diagnosis

- Systolic ejection murmurs heard in the left upper lung field anteriorly and near the spine on the left side posteriorly.
- There may be an aortic regurgitation or stenosis murmur due to an associated bicuspid aortic valve.
- A continuous murmur heard superiorly and midline in the back or over the left anterior chest is correlated with severe coarctation.

Testing

- EKG may show LVH
- Scalloping of the inferior portion of the ribs (rib notching) due to enlarged collateral intercostal arteries noted on chest x-ray
- Dilation of the left subclavian artery and post-stenotic aortic dilation along with LV enlargement may be present
- The coarctation region and the poststenotic dilation of the descending aorta may result in a "3" sign along the aortic shadow on the PA chest radiograph
- The notch in the "3" represents the area of coarctation.

Outcomes

- Cardiac failure is common in infancy and in older untreated patients when the coarctation is severe.
- Die of hypertension, rupture of the aorta, infective endarteritis, or cerebral hemorrhage before the age of 50
- Aortic dissection also occurs with increased frequency.
- Coarctation of any significance may be poorly tolerated in pregnancy because of the inability to support the placental flow.

Treatment

- Endovascular stenting usual
- Surgical resection with end-to end repair is of low mortality but does carry a risk of spinal cord injury
- 25% of patients remain hypertensive after correction

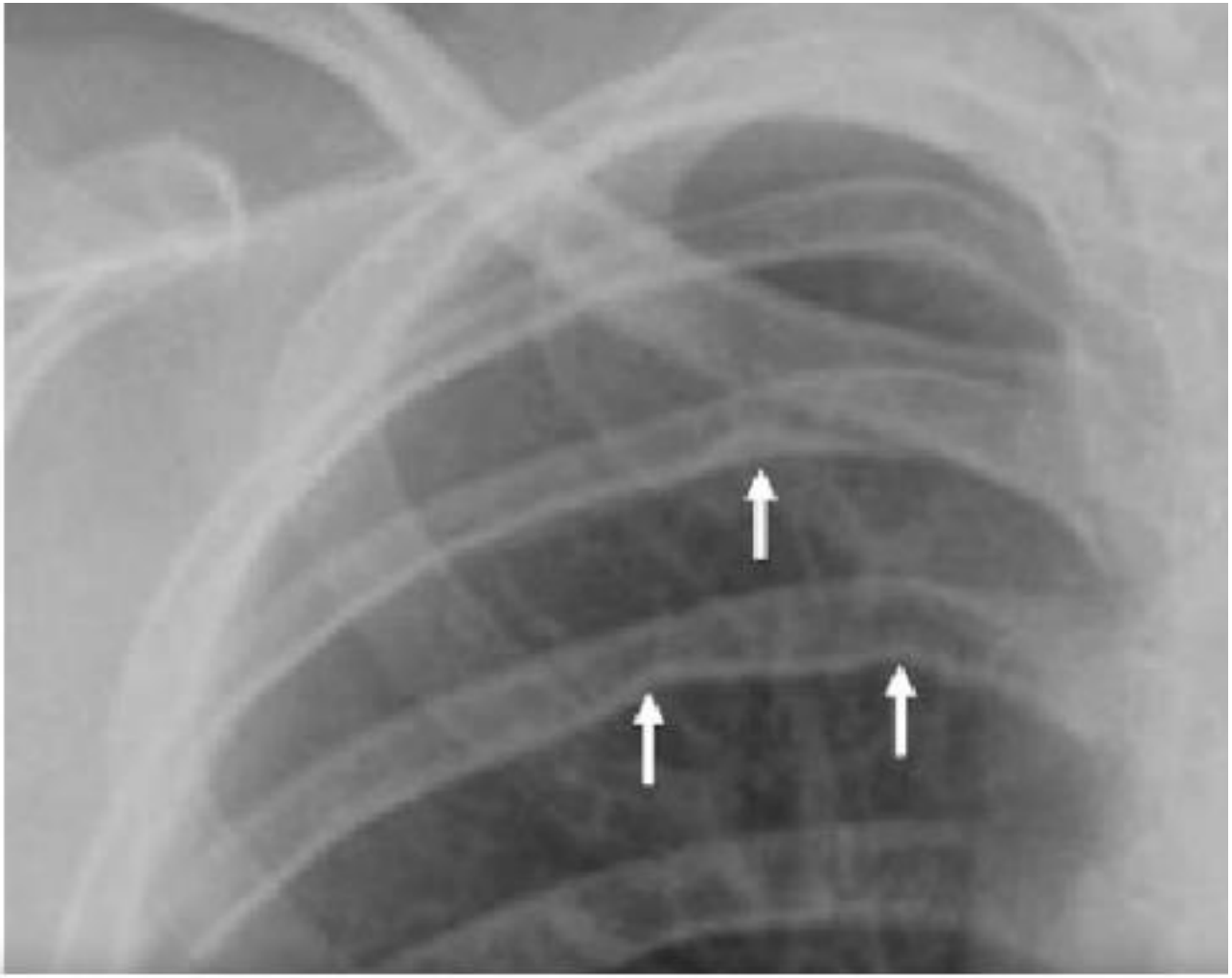
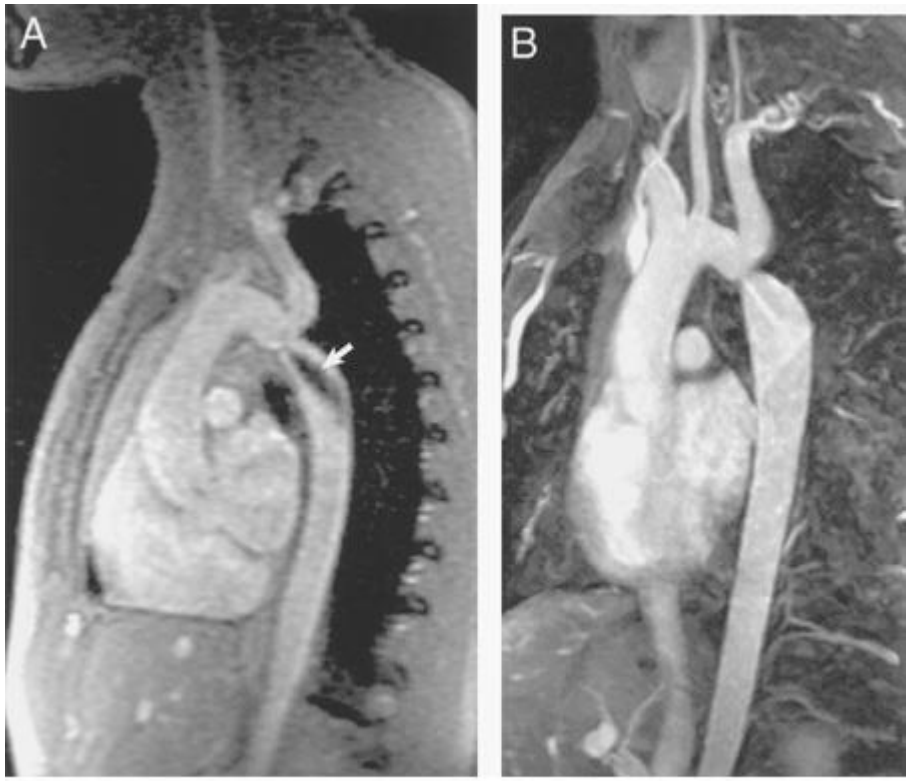


Figure 1. Chest x-ray (anteroposterior view). White arrows indicate rib notching.

Published in Circulation 2009

Aortic coarctation

Coarctation of the aorta



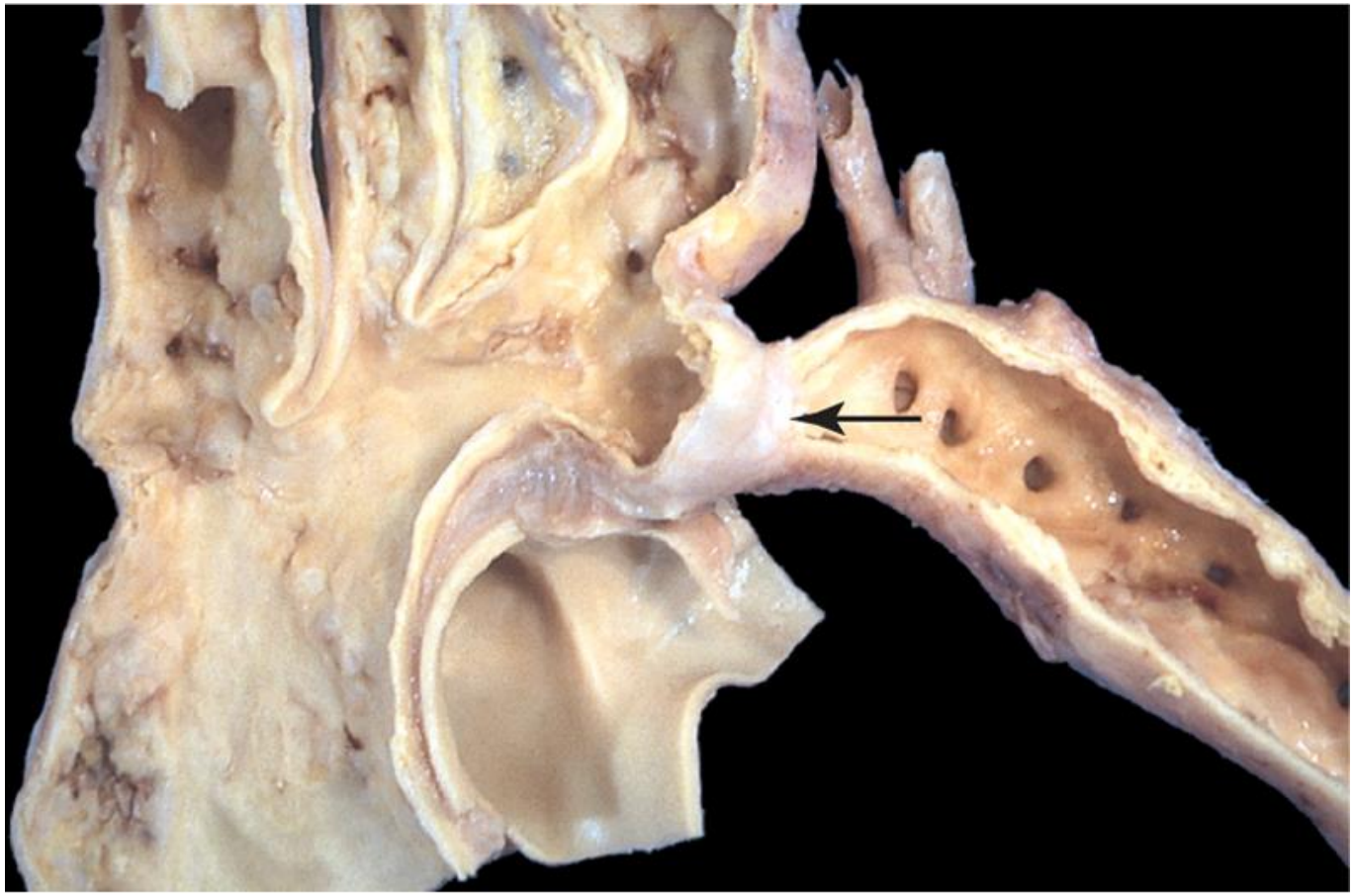
Source: Fuster V, O'Rourke RA, Walsh RA, Poole-Wilson P: *Hurst's The Heart*, 12th Edition: http://www.accessmedicine.com

Fig. 82-20 Accessed 04/01/2010

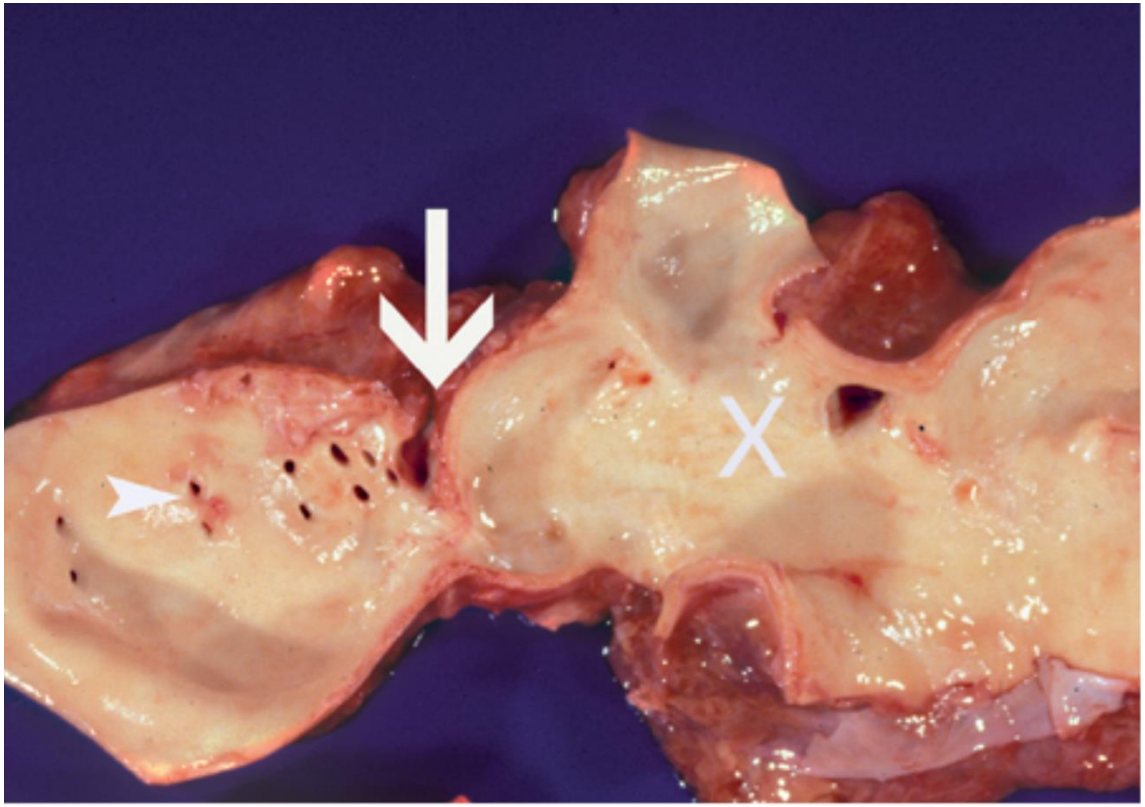
A. MRI angiography systolic frame showing turbulent jet at the coarctation site (*arrow*).
B. Subvolume maximal intensity projection image of the aorta.

Source: From Geva T, Sahn DJ, Powell AC. Magnetic resonance imaging of congenital heart disease in adults. *Prog Pediatr Cardiol* 2003;17:21–39. Reproduced with permission of the authors and publisher.

Coarctation of the aorta postductal



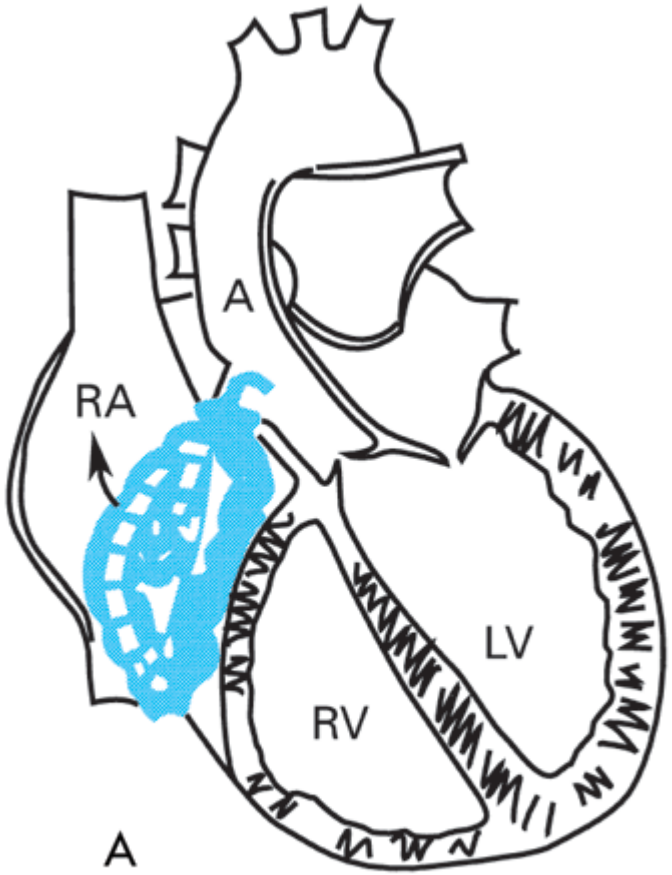
Coarctation of the aorta postductal

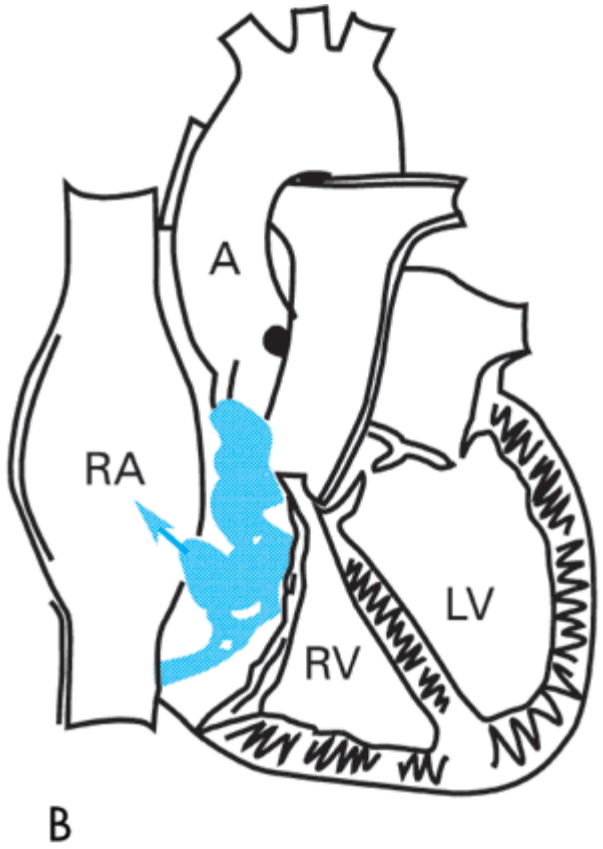


Source: Kemp WL, Burns DK, Brown TG: *Pathology: The Big Picture*: www.accessmedicine.com

Post ductal coarctation (arrow); the aortic arch is marked with an “x”

Anomalous communication of coronary arteries





Anomalous communication of coronary arteries

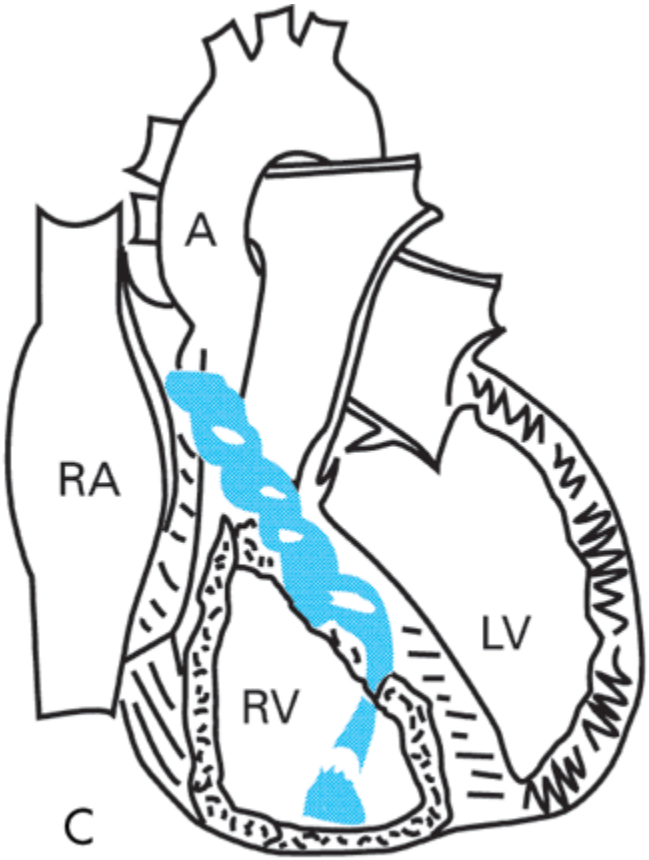


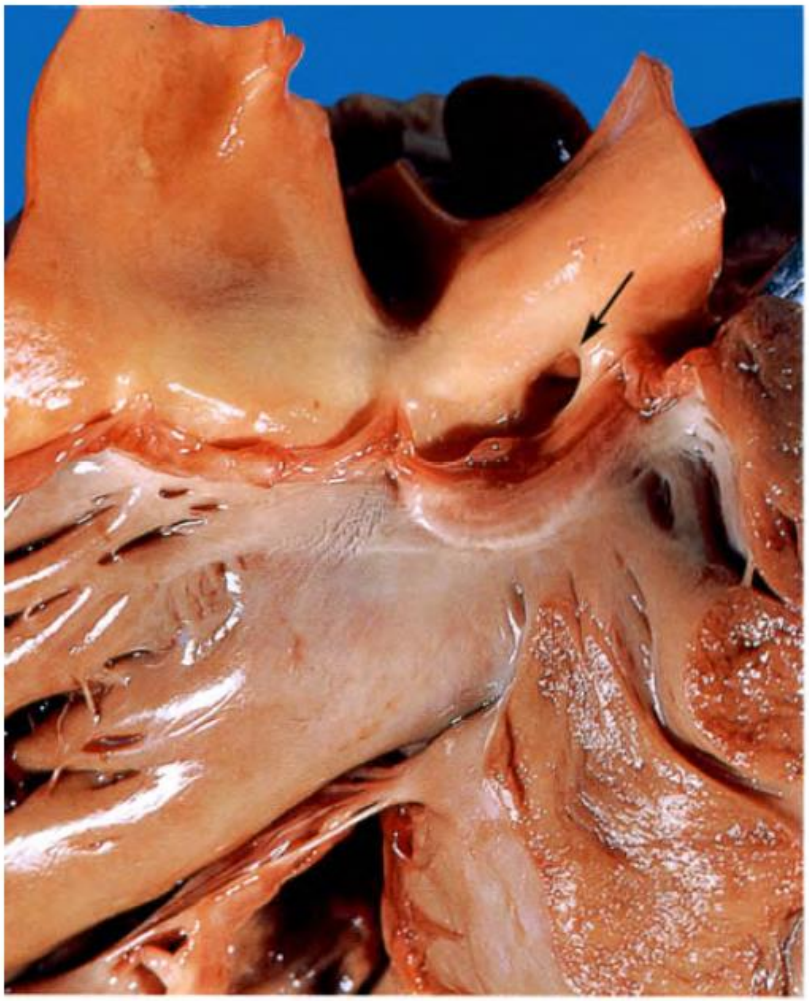
Fig. 82-44 Accessed 04/01/2010

A. Right coronary artery communicates with coronary sinus. **B.** Right coronary artery communicates with right atrium (RA). **C.** Anomalous communication of right coronary artery with right ventricle (RV). **D.** Two coronary arteries arise from the aorta (A) and make collateral communication with accessory coronary artery arising from pulmonary trunk (PT). LV, left ventricle.

Anomalous communication of coronary arteries

- The right coronary artery is the site of origin in somewhat over half the cases, and the two most common sites into which the fistula feeds are a pulmonary artery and the right ventricle.
- Continuous murmur in an unusual location

Anomalous communication of coronary arteries



Left coronary artery arising from the pulmonary trunk.
The orifice of the coronary artery can be seen arising from the sinus above the pulmonary artery cusp on the right (arrow). This is the commonest congenital abnormality of the coronary arteries. Death resulted from myocardial infarction
caused by perfusion of the left ventricle by poorly oxygenated blood.

Sheppard, MN, Herrington, CS, “The Cardiovascular System,” in Herrington, CS (ed), Muir’s Textbook of Pathology, 15th edition. 2014. CRC Press. Boca Raton, Florida. Fig. 1.73

Pulmonic valve stenosis

- Asymptomatic unless lesion is at least moderately severe.
- Severe cases may present with right-sided heart failure.
- Cyanosis and heart failure in ductal dependent lesions (neonates)
- If ventricular septum intact, will shunt via PFO

Pulmonic valve stenosis

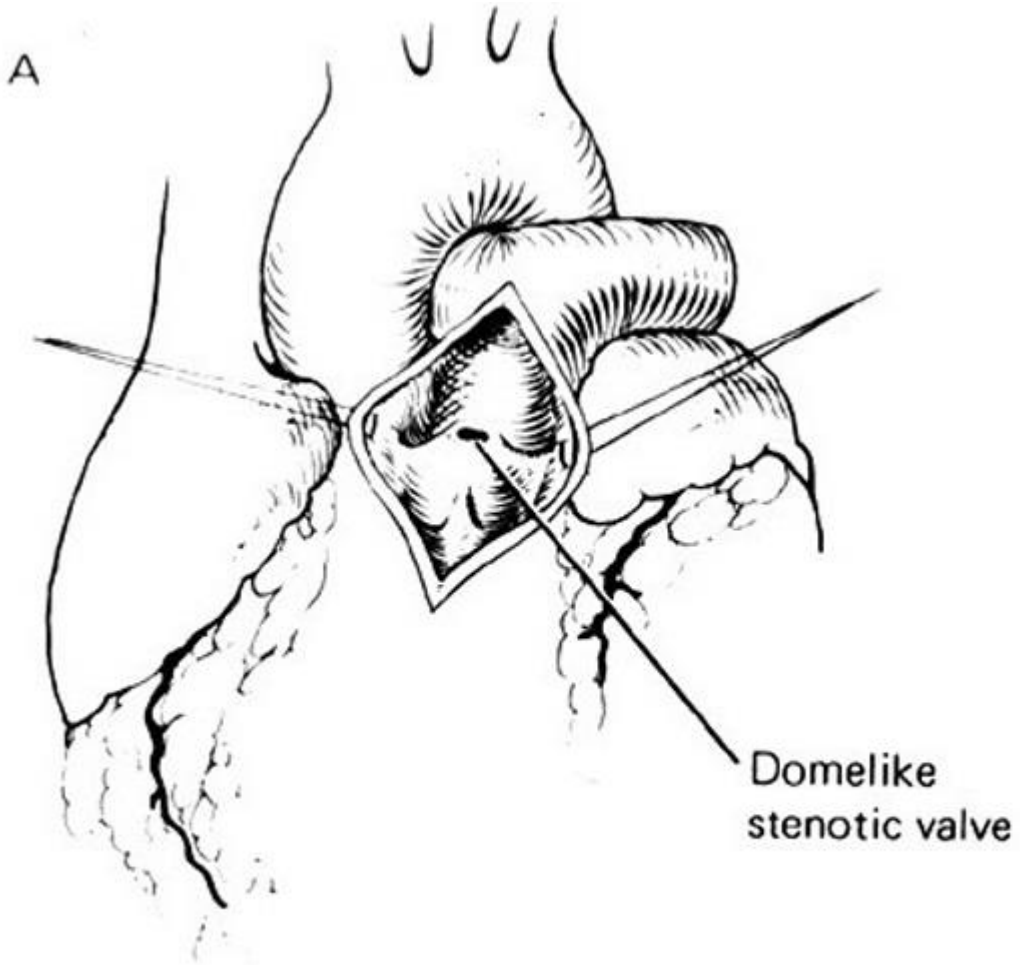
- High-pitched systolic ejection murmur maximal in the 2^{nd}-3^{rd} left interspace with radiation to the left shoulder.
- S_2 widely split
- P_2 delayed and soft or absent.

Pulmonic valve stenosis

- Pulmonary ejection click often present and decreases with inspiration
- The only right heart sound that decreases with inspiration
- RV lift seen
- Patients with peak pulmonic valve gradients greater than 60 mm Hg or a mean of 40 mm Hg by echocardiography/Doppler should undergo intervention regardless of symptoms.

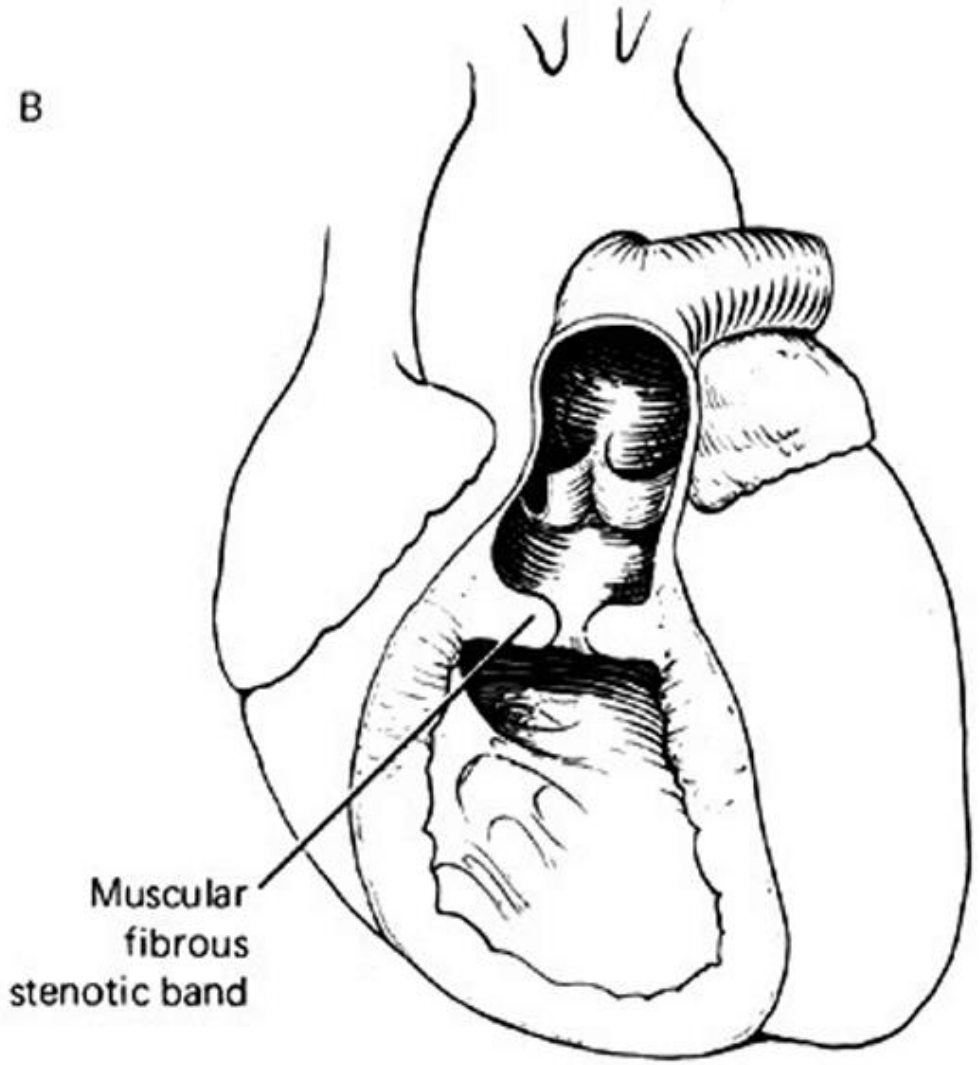
Physiology

- Pulmonic stenosis is often congenital and associated with other cardiac lesions.
- Most patients with valvular pulmonic stenosis have a domed valve
- There is increased resistance to RV outflow, a raised RV pressure, and diminished pulmonary blood flow



Physiology

- A dysplastic valve is noted in Noonan's syndrome
- The main PA is normal
- Arterial oxygen saturation is normal
- Peripheral pulmonic stenosis (of the infundibulum) is associated with congenital rubella syndrome

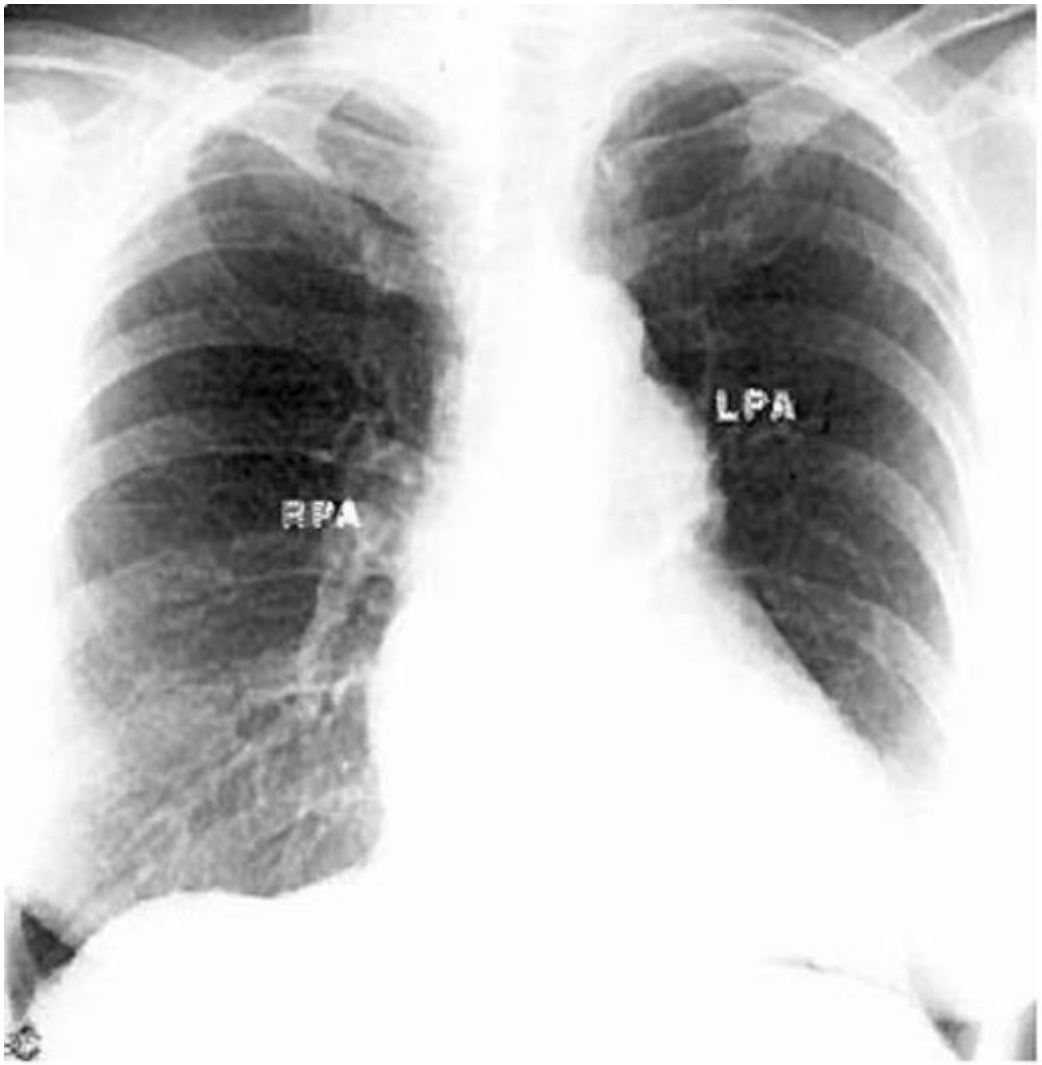


Clinical diagnosis

- Palpable parasternal lift due to RVH
- PA may be palpable as well
- Loud systolic murmur at the 2nd-3rd interspaces parasternally
- Radiate to left shoulder
- Pulmonary ejection click often present and decreases with inspiration
- The only right heart sound that decreases with inspiration

Testing

- Right axis deviation or RVH is noted
- P waves are peaked
- Often post-stenotic dilatation of pulmonary arteries
- Greater perfusion of left lung
- Chen's sign is increased perfusion of left lung base on chest x-ray



Source: Maxine A. Papadakis, Stephen J. McPhee, Michael W. Rabow: Current Medical Diagnosis and Treatment 2020

Testing

- Echocardiography/Doppler is the diagnostic tool of choice
- Distinguish between a dome valve and a dysplastic valve
- Determine the gradient across the valve
- Provide information regarding subvalvular obstruction and the presence or absence of tricuspid or pulmonic valvular regurgitation.
- No need for cardiac catheterization.

Outcomes

- A dysplastic pulmonary valve usually requires surgical treatment
- A domed pulmonary valve stenosis usually can be treated with balloon valvuloplasty.
- RV outflow tract obstruction due to other valve dysfunction may be treated with a percutaneous pulmonary valve replacement.
- Pulmonary insufficiency is a surgical complication.
- If severe, replace the valve.

Ebstein malformation of the tricuspid valve

- The septal leaflet of the tricuspid valve is displaced toward the apex of the heart and is attached to the endocardium of the RV rather than at the tricuspid annulus.
- A large portion of the RV functions physiologically as part of the right atrium and does not contribute to RV output.
- If antegrade pulmonary flow is adequate, symptoms may not present until adulthood.
- Tachyarrhythmias (and RBBB) associated with right atrial dilatation

Testing

- Cardiomegaly with prominent right heart border on chest x-ray
- Association with Wolff-Parkinson-White (WPW) syndrome, in which case a delta wave is present on EKG
- Short PR with a slurred upstroke of the QRS
- Echocardiography confirmatory

Treatment

- In cyanotic neonates, PGE_2 is used to maintain pulmonary blood flow via the ductus arteriosus until PVR decreases, facilitating antegrade pulmonary artery flow.
- Surgical interventions vary according to the severity of the disease.
- Have variable outcomes

Aortic stenosis

- Harsh systolic ejection murmur at the upper right sternal border with radiation to the neck.
- Thrill in suprasternal notch and carotid arteries.
- Systolic click at the apex.
- Dilation of the ascending aorta on chest radiograph

Aortic stenosis

- Valvular aortic stenosis
- 60-75%
- Males 3–5:1
- Typically associated with a bicuspid or unicuspid aortic valve (fusion of three leaflets).
- Leaflet fusion typically results in reduced leaflet mobility and potential obstruction to flow.

Aortic Stenosis

- Subvalvular aortic stenosis
- 10-20%
- Males 2–3:1
- Associated with a discrete membrane or muscular narrowing in the LV outflow tract.
- Supravalvular aortic stenosis
- 8-14%
- Involves narrowing of the ascending aorta, typically at the aortic sinotubular junction.

Supravalvular aortic stenosis

- Autosomal dominant.
- 7q11.23 mutation affects elastin.
- Characterized by narrowing of the ascending aorta above the level of the sinus of Valsalva.
- Left ventricular outlet obstruction
- Williams-Beuren syndrome
- Characterized additionally by transient hypercalcemia, elfin facial appearance with low bridge nose, and stenosis of other major arteries.
- Developmental delay with good language skills; impaired spatial processing.

Clinical diagnosis

- Isolated valvular aortic stenosis seldom causes symptoms in infancy
- Severe congenital stenosis can be associated with severe LV dysfunction and cardiogenic shock and require a PDA to supply systemic cardiac output (“critical” aortic stenosis)

Clinical diagnosis

- Valvular and subvalvular aortic stenosis
- Pulses are diminished with a slow upstroke.
- Palpation reveals an LV thrust at the apex and, possibly, a systolic thrill at the suprasternal notch and over the carotid arteries
- An aortic ejection click at the apex
- Does not vary with respiration
- Systolic ejection murmur is present at the right upper sternal border, radiating to the suprasternal notch and carotids

Clinical diagnosis

- Supravalvular aortic stenosis
- The harsh systolic murmur is best heard in the suprasternal notch (with possible thrill) and carotids but is well transmitted over the aortic area.
- There may be a difference in pulses and blood pressure between the right and left arms
- With more prominent pulse and higher pressure in the right arm (the Coanda effect).
- All forms are progressive.

Mitral valve prolapse

- Midsystolic click.
- Late systolic “whooping” or “honking” murmur.
- Typical symptoms reported include chest pain, palpitations, and dizziness.
- Often over-diagnosed on routine cardiac ultrasound.

Mitral valve prolapse

- As the mitral valve closes during systole, it moves posteriorly or superiorly (prolapses) into the left atrium
- 2% of thin adolescent girls
- Associated with Marfan, Loeys-Dietz, and Ehlers-Danlos syndromes

Clinical diagnosis

- Most patients are asymptomatic
- A midsystolic click (with or without a systolic murmur) is elicited best in the standing position and is the hallmark of this entity.
- Usually is heard at the apex but may be audible at the left sternal border.
- A late, short systolic murmur after the click implies mitral insufficiency
- May have dysrhythmias if mitral insufficiency

Testing and treatment

- Echocardiography is diagnostic
- Propranolol may be effective in treatment of coexisting arrhythmias
- Antibiotic prophylaxis is not needed
- Benign course if no mitral insufficiency