

Adult Congenital Heart Disease & Cyanosis

The questions signed by * are hard: read their explanation. ● Past Years ● TBL ● Others

This file belongs to the PULSE Cardiology question book: the explanations of the multiple choice answers and the model answers of the essay questions, in the words of the handout.

Answer Key | Multiple Choice Questions

Question no.	1	2	3	4	5	6	7	8	9	10	11	12
Answer	a	d	b	b	b	d	b	d	c	a	a	a

Question no.	13	14	15	16	17	18	19	20	21	22	23	24
Answer	d	b	b	d	b	c	a	b	d	c	c	d

Question no.	25	26	27	28	29	30	31	32	33	34	35
Answer	d	d	b	a	a	c	e	d	c	a	c

► Second | Questions with a Note

Question no.	1	2	3	4	5
Answer	a	a	d	b	d

► Why this answer?

- 1 * The most common congenital cyanotic heart disease in children is:

(a) Fallot tetralogy

The handout calls TOF one of the most common CHDs, and it is one of the two cyanotic CHDs it studies, with reduced pulmonary blood flow. The answer is inferred from these lines; the handout does not rank the cyanotic lesions.

- 2 Which of the following is not used in treatment of cyanotic spells of Fallot's tetralogy?

(d) Hydrocortisone

For cyanotic spells the handout gives the knee-chest position, oxygen, IV morphine and IV propranolol; hydrocortisone is not among them. Sodium bicarbonate is used to correct the acidosis of a severe spell.

- 3 In MOST patients with an ASD, the characteristic physical finding is:

(b) A wide and fixed splitting of 2nd heart sound

The RV impulse is also increased in ASD, but wide and fixed splitting of S2 is the most characteristic sign of ASD. There is no audible murmur because of flow across the ASD, so a pansystolic murmur is not expected.

- 4 Infant 8 months old diagnosed as secundum atrial septal defect ASD, the characteristic clinical sign is:
(b) Wide fixed splitting of the 2nd heart sound
 A soft ejection systolic murmur at the left upper sternal border is also heard in ASD, but wide and fixed splitting of S2 is the most characteristic sign.
- 5 A 6-month-old child presented with respiratory distress (RD), tachycardia, difficult feeding and pansystolic...
(b) VSD
 Medium and large VSDs present with congestive heart failure (dyspnea, tachypnea, sweating and failure to gain weight) and a holosystolic murmur best heard at the left lower sternal border. In ASD there is no audible murmur because of flow across the defect.
- 6 Regarding pulmonary stenosis in children, which is false?
(d) Left ventricular hypertrophy
 Pulmonary stenosis is a right ventricular outflow obstruction and a pressure overload on the right ventricle, and hypertrophy occurs mainly in pressure overload. So the right ventricle hypertrophies, not the left.
- 7 The cardiovascular disease that is diagnosed by just palpation:
(b) Coarctation of the aorta
 Palpation of femoral pulses to detect radio-femoral delay picks up coarctation. VSD and PDA are diagnosed by their murmurs.
- 8 Differential Cyanosis is a possible complication of:
(d) Patent ductus arteriosus (PDA)
 Differential clubbing may occur in a patient with patent ductus arteriosus associated with pulmonary artery hypertension and right to left shunt, sparing the upper limbs. The PDA joins the descending aorta at the level of the left subclavian artery.
- 9 Which of the following is considered an acyanotic congenital heart disease?
(c) Ventricular septal defect.
 Acyanotic CHDs are classified to left to right shunting and obstructive lesions, and VSD is one of the left-to-right shunts. Fallot is a cyanotic CHD.
- 10 * Which of the following is the most common congenital cyanotic heart lesion in children?
(a) Fallot's tetralogy.
 The handout calls TOF one of the most common CHDs, and it is one of the two cyanotic CHDs it studies, with reduced pulmonary blood flow. The answer is inferred from these lines; the handout does not rank the cyanotic lesions.
- 11 * The most common cyanotic congenital heart disease is:
(a) Fallot tetralogy
 The handout calls TOF one of the most common CHDs, and it is one of the two cyanotic CHDs it studies, with reduced pulmonary blood flow. The answer is inferred from these lines; the handout does not rank the cyanotic lesions.
- 12 Tetralogy of Fallot consisted of the following, except:
(a) ASD.
 TOF comprises RVOTO (infundibular stenosis), VSD, aorta dextroposition (overriding aorta) and RV hypertrophy. ASD is not one of the four components.
- 13 Infant 20 months old diagnosed as Tetralogy of Fallot, which of the following is not true:
(d) Small VSD
 RV hypertrophy is one of the four components of TOF, and polycythemia is linked to cyanosis. A large VSD is the usual defect of Fallot, so 'small VSD' is the wrong statement; the handout does not give the VSD size or squatting.
- 14 Infant 20 months old diagnosed as Tetralogy of Fallot, which of the following is not true:
(b) Left ventricle hypertrophy
 TOF has RV hypertrophy, not left ventricular hypertrophy. Squatting, polycythemia and a large VSD fit a cyanotic child with Fallot.

- 15 The most common valvular regurgitation in ventricular septal defect is:
(b) AR
Aortic insufficiency develops in approximately 5% of VSD patients, from prolapse of an aortic valve cusp into the VSD or lack of support to the aortic root.
- 16 Cyanotic spells may be a complication of:
(d) Tetralogy of Fallot
Cyanotic spells are treated under tetralogy of Fallot: knee-chest position, oxygen, IV morphine and IV propranolol. PDA, VSD and ASD are acyanotic left-to-right shunts.
- 17 Pulmonary oligemia is seen in X-ray in the following congenital heart lesion:
(b) Tetralogy of Fallot's
TOF is a cyanotic CHD with reduced pulmonary blood flow, seen on CXR as decreased vascularity (oligemia). VSD is a left-to-right shunt with increased pulmonary vascular markings.
- 18 The followings are cyanotic heart lesions with lung oligemia except:
(c) Transposition of the great vessels
Diminished pulmonary vasculature represents obstruction of blood flow to the lungs, as in Fallot with reduced pulmonary blood flow. Transposition of the great vessels is a cyanotic lesion with plethoric lungs, not oligemic.
- 19 Which of the following diseases is characterized by wide and fixed splitting of S2?
(a) Atrial septal defect.
S2 is widely split and fixed (splitting does not vary with respiration) and is the most characteristic sign of ASD.
- 20 The wide and fixed splitting of the second heart sound occurs in:
(b) Atrial septal defect
S2 is widely split and fixed, does not vary with respiration, and is the most characteristic sign of ASD.
- 21 A large ventricular septal defect (VSD) is likely to:
(d) Be commonly associated with plethoric lung.
A large left-to-right shunt gives increased pulmonary vascular markings. Wide and fixed splitting belongs to ASD, and cyanosis from shunt reversal (Eisenmenger) makes the patient inoperable.
- 22 6-month-old baby with PDA, which of the following is true:
(c) Continuous murmur at upper left sternal border
PDA gives a continuous murmur heard best at the left upper sternal border and bounding pulses. Higher arm pressure and rib notching belong to coarctation.
- 23 The characteristic clinical sign of Patent ductus arteriosus is:
(c) Continuous machinery murmur at upper left sternal border
Bounding pulses are also found in PDA, but the continuous (machinery) murmur at the left upper sternal border is its characteristic sign. Rib notching belongs to coarctation.
- 24 Patients with a small PDA have the following criteria EXCEPT:
(d) Risk for endocarditis is extremely low.
Isolated PDA is closed mainly to prevent bacterial endocarditis, so the risk is not extremely low. A small PDA has no symptoms and is detected by a murmur, a continuous murmur at the left upper sternal border.
- 25 Which of the following is a complication of PDA?
(d) All of the above
Moderate to large PDA may present with congestive heart failure, PDA is closed mainly to prevent bacterial endocarditis, and a right-to-left shunt from raised pulmonary vascular resistance is Eisenmenger's physiology.
- 26 Regarding pulmonary stenosis (PS), the most likely is:
(d) It causes cyanosis if critical.
Critical pulmonary stenosis can cause cyanosis by right-to-left shunting at atrial level. The lungs are not plethoric because diminished pulmonary vasculature represents obstruction of blood flow to the lungs; the murmur is not at the apex.

- 27** The cardiovascular disease that is diagnosed by just palpation:
(b) Coarctation of the aorta
 Palpation of femoral pulses (to detect radio-femoral delay) is necessary to avoid a delay in the diagnosis of coarctation.
- 28** Which of the following is the most probable etiology?
(a) Eisenmenger
 Late cyanosis with clubbing, a loud P2 and central dilation with peripheral pruning of pulmonary arteries plus RV hypertrophy mean a right-to-left shunt in response to raised pulmonary vascular resistance: Eisenmenger. In TOF the pulmonary artery is hypoplastic, not enlarged.
- 29** What is the best treatment?
(a) Medical treatment
 Eisenmenger's syndrome makes the patient inoperable, and large VSDs with irreversible pulmonary vascular obstructive disease are not candidates for surgery; symptomatic treatment is given. Closing the defect, a shunt or prostaglandin E1 are therefore not the answer.
- 30** A female 16 years old, with shortness of breath. She has normal pulse, RR, BP, no murmurs and S2 wide fixed...
(c) Infective endocarditis
 Wide fixed splitting of S2 with dilated right heart means ASD. Infective endocarditis is rare in ASD due to low pressure gradient between the two atria, while heart failure and supra-ventricular arrhythmias are recognised.
- 31** Which is the most probable diagnosis?
(e) PDA
 A machinery murmur at the left upper sternal border with bounding pulses and no cyanosis means PDA. A big VSD gives a holosystolic murmur at the left lower sternal border.
- 32** * A full-term 4-day-old sick male baby comes with severe heart failure, metabolic acidosis and shock. On...
(d) Critical aortic coarctation
 Femoral pulses are palpated to detect coarctation, and early onset COA may reveal cardiomegaly, pulmonary edema and other signs of congestive heart failure. The answer is inferred from these lines; a large PDA gives bounding pulses instead.
- 33** Central cyanosis is not found in:
(c) Left-to-right shunt
 Central cyanosis is caused by congenital heart diseases with right to left shunt and is seen in acute pulmonary oedema. In a left-to-right shunt the oxygenated blood is shunted from left to right, so the lesion is acyanotic.
- 34** Which of the following congenital heart defects causes fixed splitting of the second heart sound?
(a) Atrial septal defect
 Fixed splitting of S2 is the most characteristic sign of ASD. In Ebstein's anomaly the second heart sound is usually normal.
- 35** * Pedal pulse is 'absent' in all except:
(c) Coarctation of aorta
 In coarctation collateral vessels connect the upper arteries to the vessels below the coarctation, and the femoral pulse shows radio-femoral delay. Embolism can make pulsations absent, and Leriche syndrome narrows the terminal abdominal aorta; the answer is inferred from these lines.

► Second | Questions with a Note: why this answer?

- 1** 9 months old baby with small muscular VSD, which of the following is not true:
(a) Heart failure is common
 In small VSDs the patients are usually asymptomatic and the only abnormality is a loud murmur at the left lower sternal border; heart failure belongs to medium and large defects. Endocarditis is more common in small than in big VSD.

2 9 months old infant with small VSD, which of the following is true?

(a) Loud pansystolic murmur over the left parasternal border

In small VSDs the patients are usually asymptomatic and the only abnormality is a loud murmur at the left lower sternal border. Heart failure and chest infection belong to medium and large defects, and pulmonary vascular obstructive disease follows high pressure and high flow.

3 3 years old boy with aortic coarctation, which of the following is not true:

(d) Recurrent lower respiratory infections

Radio-femoral delay, upper limb hypertension and rib notching are features of coarctation. Respiratory infections belong to medium and large left-to-right shunts such as VSD.

4 * 10 months old baby with large muscular VSD, which of the following is false:

(b) Loud harsh pansystolic murmur

Respiratory infection, heart failure and a loud single S2 when PAP is elevated are expected in medium and large VSDs. The answer is inferred from 'The intensity of the murmur may vary between grades II-V/VI', so the murmur of a large defect is not always loud.

5 All of the following are true about pulmonary stenosis except:

(d) Accentuated second heart sound

A loud P2 is a sign of pulmonary hypertension, not of pulmonary stenosis, where S2 is soft. Pulmonary stenosis is a pressure overload that causes right ventricular hypertrophy.

Model Answers | Essay Questions

1 Enumerate four complications of ventricular septal defect.

- ① **Congestive heart failure:** patients with medium and large defects may present with symptoms of congestive heart failure (dyspnea, tachypnea, sweating and failure to gain weight) or with symptoms related to bronchial obstruction and/or respiratory infection.
- ② **Pulmonary vascular obstructive disease**
 - May develop in **10%** of VSDs.
 - This is probably related to the exposure of the pulmonary vascular bed to high pressure and high flow.
- ③ **Development of infundibular stenosis "Gasul's transformation of the VSD"**
 - May occur in **8%** of the defects.
 - It is more with right aortic arch and increased angle of the right ventricular outflow tract.
 - While development of infundibular stenosis eventually requires the patient to have surgery, it indeed protects the pulmonary vascular bed and prevents development of pulmonary vascular obstruction disease.
- ④ **Aortic insufficiency**
 - Develops in approximately **5%** of patients.
 - This may either be related to prolapse of an aortic valve cusp into the VSD or lack of support to the aortic root.
 - More with supracristal VSDs.
 - Surgical correction is indicated if moderate to severe aortic insufficiency is present.
- ⑤ **Eisenmenger's complex:** flow reversal across a VSD when pulmonary vascular resistance exceeds systemic levels.
- ⑥ **Infective endocarditis:** congenital anomalies, esp. VSD, PDA and coarctation.
 - Narrow orifice: more common in small than in big VSD.

2 Describe the complications of Fallot's tetralogy.

- ① The mortality rate in untreated patients reaches **50%** by age 6 years.
- ② **Syncope.**
 - Hypoxic syncope: Fallot's tetralogy and other cyanotic diseases.
- ③ **Right-sided heart failure:** elevated jugular venous pressure [JVP] with large A wave, ascites, peripheral edema, hepatomegaly.
- ④ **Pulmonary valve insufficiency "PR":** low-pitched, short diastolic murmur.
- ⑤ A mild **anemia** may be present; polycythemia is rare in the absence of cyanosis.
- ⑥ Coagulation profile may be abnormal in patients with cyanosis and **bleeding**.
- ⑦ **Endocarditis or sepsis:** obtain blood cultures in febrile patients to rule out endocarditis or sepsis.
- ⑧ **Cyanotic spells** (adults with acute cyanosis).
- ⑨ **Hyperviscosity** in a nondehydrated state, most commonly when hematocrit levels > **65-70**; if symptoms occur with hematocrits < 65, evidence of iron deficiency should be sought.
- ⑩ **Cerebral arterial thrombosis:** prophylactic phlebotomy has no place in the prevention of cerebral arterial thrombosis.

Note The handout gives no separate list of Fallot complications; the points are gathered from its pages.

3 Mention five complications of patent ductus arteriosus (PDA) and PDA diagnosis in pediatrics.

Complications

- ① **Congestive heart failure:** moderate to large PDA may either present with symptoms of easy fatigability, symptoms associated congestive heart failure.
- ② **Bacterial endocarditis:** isolated PDA is an indication for trans-catheter device closure by cardiologists, mainly to prevent bacterial endocarditis.
 - Infective endocarditis, underlying cardiac lesion: congenital anomalies, esp. VSD, PDA and coarctation.
- ③ **Hyperdynamic pulmonary hypertension:** it is due to increased pulmonary arterial blood flow; it occurs in congenital heart disease with left to right shunt e.g. ASD, VSD & PDA.
- ④ **Eisenmenger's physiology:** a right-to-left shunt occurs in response to an elevation in pulmonary vascular resistance.
- ⑤ **Differential clubbing:** occasionally, clubbing may occur in lower limbs, sparing the upper limbs; it may occur in a patient with patent ductus arteriosus associated with pulmonary artery hypertension and right to left shunt.

Diagnosis

Clinical presentation depends upon the size of the ductus.

- * If the PDA is small, there are no symptoms, and it is usually detected because of a murmur detected on a routine examination.
- * Moderate to large PDA may either present with symptoms of easy fatigability, symptoms associated **congestive heart failure**.

Physical findings

- * Left ventricular impulse is normal in small PDA and may be hyperdynamic with large shunts.
- * A thrill may be felt at the left upper sternal border and in the suprasternal notch.
- * S1 is usually normal and S2 may be buried within the murmur.
- * In most cases, a **continuous murmur** is heard best at the **left upper sternal border**. The murmur begins in systole and continues through S2 into the diastole. The systolic component of the murmur crescendos up to S2 while the diastolic part decrescendo to a varying distance (time) into the diastole → **machinery murmur**.

- * In big PDA, a mid-diastolic murmur may be heard at the apex because of increased flow across the mitral valve; such a mid-diastolic murmur suggests a Qp:Qs greater than **2:1** (Pulmonary to systemic flow = Qp:Qs).
- * Arterial pulses are **bounding** in all but patients with very small ductus.

Investigations

	Findings
Chest x-ray	If moderate to large PDA, cardiomegaly, increased pulmonary blood flow and left atrial enlargement may be seen.
Electrocardiogram	The ECG may be normal or may show left atrial and left ventricular enlargement, depending upon the size of the ductus.
Echocardiogram	Doppler echocardiography shows characteristic diastolic flow pattern in the pulmonary artery, indicative of PDA. Also determine the size of PDA.
Cardiac catheterization and selective cine angiography	Integral part of transcatheter closure. Selective aortic arch injection demonstrates the size, shape, and location of the ductus.

Note The handout gives no separate list of PDA complications; the points are gathered from several pages.

4 Enumerate 3 complications of patent ductus arteriosus.

- 1 **Congestive heart failure:** moderate to large PDA may either present with symptoms of easy fatigability, symptoms associated congestive heart failure.
- 2 **Bacterial endocarditis:** isolated PDA is an indication for trans-catheter device closure by cardiologists, mainly to prevent bacterial endocarditis.
 - Infective endocarditis, underlying cardiac lesion: congenital anomalies, esp. VSD, PDA and coarctation.
- 3 **Hyperdynamic pulmonary hypertension:** it is due to increased pulmonary arterial blood flow; it occurs in congenital heart disease with left to right shunt e.g. ASD, VSD & PDA.
- 4 **Eisenmenger's physiology:** a right-to-left shunt occurs in response to an elevation in pulmonary vascular resistance.
- 5 **Differential clubbing:** occasionally, clubbing may occur in lower limbs, sparing the upper limbs; it may occur in a patient with patent ductus arteriosus associated with pulmonary artery hypertension and right to left shunt.

Note The handout gives no separate list of PDA complications; the points are gathered from several pages.

5 Discuss treatment of patent ductus arteriosus (PDA).

- * Isolated PDA is an indication for **trans-catheter device closure** by cardiologists, mainly to prevent bacterial endocarditis.
- * If the patient is asymptomatic, waiting until **6-12 months** of age.
- * For large-sized PDA, **surgical closure** is available option.
- * If so small PDA, but no continuous murmur on auscultation → called "**silent ductus**" may no need for intervention.

6 Mention clinical picture (CP) and treatment of PDA.

Clinical picture

Symptoms

Clinical presentation depends upon the size of the ductus.

- * If the PDA is small, there are no symptoms, and it is usually detected because of a murmur detected on a routine examination.
- * Moderate to large PDA may either present with symptoms of easy fatigability, symptoms associated **congestive heart failure**.

Physical findings

- * Left ventricular impulse is normal in small PDA and may be hyperdynamic with large shunts.
- * A thrill may be felt at the left upper sternal border and in the suprasternal notch.
- * S1 is usually normal and S2 may be buried within the murmur.
- * In most cases, a **continuous murmur** is heard best at the **left upper sternal border**. The murmur begins in systole and continues through S2 into the diastole. The systolic component of the murmur crescendos up to S2 while the diastolic part decrescendo to a varying distance (time) into the diastole → **machinery murmur**.
- * In big PDA, a mid-diastolic murmur may be heard at the apex because of increased flow across the mitral valve; such a mid-diastolic murmur suggests a Qp:Qs greater than **2:1** (Pulmonary to systemic flow = Qp:Qs).
- * Arterial pulses are **bounding** in all but patients with very small ductus.

Treatment (Management)

- * Isolated PDA is an indication for **trans-catheter device closure** by cardiologists, mainly to prevent bacterial endocarditis.
- * If the patient is asymptomatic, waiting until **6-12 months** of age.
- * For large-sized PDA, **surgical closure** is available option.
- * If so small PDA, but no continuous murmur on auscultation → called "**silent ductus**" may no need for intervention.

7

Clarify hemodynamics, clinical picture (CP) and complications of TOF.

Hemodynamics

TOF is one of the most common CHDs, comprises:

- ① Right ventricular (RV) outflow tract obstruction (RVOTO) (infundibular stenosis).
- ② Ventricular septal defect (VSD).
- ③ Aorta dextroposition (overriding aorta).
- ④ And RV hypertrophy.

Cyanotic CHD with **reduced pulmonary blood flow**.

Clinical picture

Signs and symptoms

Signs and symptoms are directly related to the severity of the anatomic defects and may include the following:

- ① Lack of exercise endurance.
- ② Syncope.
- ③ Palpitations.
- ④ Exertional dyspnea, usually worsening with age.

Physical findings in adults

- * Evidence of **right-sided heart failure** (elevated jugular venous pressure [JVP] with large A wave, ascites, peripheral edema, hepatomegaly).
- * In individuals with pulmonary valve insufficiency "PR": low-pitched, short diastolic murmur.

Complications

- ① The mortality rate in untreated patients reaches **50%** by age 6 years.
- ② **Syncope.**
 - Hypoxic syncope: Fallot's tetralogy and other cyanotic diseases.
- ③ **Right-sided heart failure:** elevated jugular venous pressure [JVP] with large A wave, ascites, peripheral edema, hepatomegaly.
- ④ **Pulmonary valve insufficiency "PR":** low-pitched, short diastolic murmur.
- ⑤ A mild **anemia** may be present; polycythemia is rare in the absence of cyanosis.
- ⑥ Coagulation profile may be abnormal in patients with cyanosis and **bleeding**.
- ⑦ **Endocarditis or sepsis:** obtain blood cultures in febrile patients to rule out endocarditis or sepsis.
- ⑧ **Cyanotic spells** (adults with acute cyanosis).
- ⑨ **Hyperviscosity** in a nondehydrated state, most commonly when hematocrit levels > **65-70**; if symptoms occur with hematocrits < 65, evidence of iron deficiency should be sought.
- ⑩ **Cerebral arterial thrombosis:** prophylactic phlebotomy has no place in the prevention of cerebral arterial thrombosis.

Note The handout does not explain the haemodynamics of Fallot or list its complications; points are gathered from its pages.

- 8** Mention the clinical picture (CP), investigations and complications of the following: TOF, aortic coarctation.

(a) Tetralogy of Fallot

Clinical picture

Signs and symptoms

Signs and symptoms are directly related to the severity of the anatomic defects and may include the following:

- ① Lack of exercise endurance.
- ② Syncope.
- ③ Palpitations.
- ④ Exertional dyspnea, usually worsening with age.

Physical findings in adults

- * Evidence of **right-sided heart failure** (elevated jugular venous pressure [JVP] with large A wave, ascites, peripheral edema, hepatomegaly).
- * In individuals with pulmonary valve insufficiency "PR": low-pitched, short diastolic murmur.

Investigations

In adult patients, laboratory studies may be helpful:

	Finding / use
Complete blood cell count	A mild anemia may be present; polycythemia is rare in the absence of cyanosis.
Coagulation profile	May be abnormal in patients with cyanosis and bleeding.
Arterial blood gas (including serum lactate, base excess, oxygen partial pressure [PO₂])	The patient's oxygenation status, serum lactate levels, and base excess value appear to be prognostic markers for mortality in repaired TOF.
Blood cultures	Obtain blood cultures in febrile patients to rule out endocarditis or sepsis.

	Findings
CXR	May be normal or depict cardiomegaly and prominent RV shadowing. Although the classic boot-shaped heart (coeur en sabot) is the hallmark of the disorder in infants, this shape of the heart may not be seen in adult patients.
Echocardiography	Color-flow Doppler echocardiography: assesses overall cardiac function and the status of the valves, and for the presence of any residual VSD, ductus arteriosus, or atrial septal defect. Reveals the grade and severity of any RVOT obstruction.
MRI	Gold standard for assessing RV function and size, and for quantifying the pulmonary regurgitant volume; provides good delineation of the aorta size and assessment of the pulmonary arteries and its branches, the status of the RVOT, and the presence of VSDs, and/or RV hypertrophy.
Cardiac catheterization	Allows: assessment of the pulmonary annulus size and pulmonary arteries; assessment of the severity of RVOTO; location of the position and size of the VSD; ruling out possible coronary artery anomalies.

Complications

- ① The mortality rate in untreated patients reaches **50%** by age 6 years.
- ② **Syncope.**
 - Hypoxic syncope: Fallot's tetralogy and other cyanotic diseases.
- ③ **Right-sided heart failure:** elevated jugular venous pressure [JVP] with large A wave, ascites, peripheral edema, hepatomegaly.
- ④ **Pulmonary valve insufficiency "PR":** low-pitched, short diastolic murmur.
- ⑤ A mild **anemia** may be present; polycythemia is rare in the absence of cyanosis.
- ⑥ Coagulation profile may be abnormal in patients with cyanosis and **bleeding.**
- ⑦ **Endocarditis or sepsis:** obtain blood cultures in febrile patients to rule out endocarditis or sepsis.
- ⑧ **Cyanotic spells** (adults with acute cyanosis).
- ⑨ **Hyperviscosity** in a nondehydrated state, most commonly when hematocrit levels > **65-70**; if symptoms occur with hematocrits < 65, evidence of iron deficiency should be sought.
- ⑩ **Cerebral arterial thrombosis:** prophylactic phlebotomy has no place in the prevention of cerebral arterial thrombosis.

(b) Aortic coarctation

Clinical picture

In adolescents and adults, the diagnosis should always be sought in the event of unexplained hypertension of the upper extremities.

Symptoms

- * Presenting symptoms may include headaches, chest pain, fatigue, or even life-threatening intracranial hemorrhage.
- * True claudication is rare.
- * Many patients are asymptomatic except for the incidentally noted hypertension.
- * Frequently, COA is not recognized by the primary care physician.

Pulse & blood pressure

- * Palpation of femoral pulses (to detect **radio-femoral delay**), and measurement of blood pressure during routine examination is necessary to avoid a delay in the diagnosis.
- * A pressure difference of more than **20 mm Hg** in favor of the arms may be considered evidence of coarctation of the aorta.

Auscultation

- * A murmur in the left infraclavicular area and under the left scapula may be systolic, but the murmur may also sound continuous in the presence of multiple collateral vessels or, occasionally, severe coarctation.
- * An ejection click may be audible when an associated bicuspid aortic valve and a murmur of aortic stenosis or insufficiency are present.
- * Similarly, a murmur of mitral stenosis or LV outflow tract obstruction may also occur.
- * A gallop rhythm may occur in the presence of a hypertrophic noncompliant LV.

Other findings on physical examination

- * Abnormalities of blood vessels in the retina and a prominent suprasternal notch pulsation.
- * A thrill may be present in the suprasternal notch or on the precordium in the presence of significant aortic valve stenosis.
- * In the rare case of abdominal coarctation, an abdominal bruit may be noted.

Investigations

	Findings
ECG	May be normal or may reveal LV hypertrophy and may show signs of LV ischemia or strain. Sometimes, LV hypertrophy may manifest as increased S waves in leads V5 and V6, the so-called posterobasal LV hypertrophy.
CXR	With early onset of COA may reveal cardiomegaly, pulmonary edema, and other signs of congestive heart failure (CHF); with late onset COA may reveal cardiomegaly; an inverted "3" sign of the barium-filled esophagus or a "3" sign on a highly penetrated CXR (frontal view) may be visualized; rib notching secondary to collateral vessels may also be seen.
Echocardiography	The suprasternal notch 2-D view allows evaluation of the aortic arch to assess the transverse aortic arch, isthmus, and severity of coarctation and to measure the pressure gradient "PG" at the site of COA.
MRI and CT	Are useful in older or postoperative patients to assess residual arch obstruction, arch hypoplasia, or formation of aneurysms.

Complications

- ① Complications of COA include: secondary **LVH**, systemic **HTN**, **HF**, **aortic dissection**, premature **CAD**, and **cerebrovascular complications**.
- ② Some patients with COA may have **cerebral aneurysms**, predisposing them to cerebrovascular accidents with severe hypertension later in life.
- ③ With untreated coarctation a mean survival of **35 years** with a 75% mortality by age 50 years can be expected.

Note The handout gives no separate list of Fallot complications; that part is gathered from its pages.

9 Describe treatment of the following congenital cardiac defects: TOF, PDA, VSD, TGA.

(a) Tetralogy of Fallot

Cyanotic spells

For adults with acute cyanosis:

- * Place them in a **knee-chest position**.
- * In addition, provide/administer **Oxygen**, **IV morphine** and **IV propranolol** for severe cases.

Surgery

- * Most symptomatic adults with TOF require some type of surgical intervention.
- * **Pulmonary valve replacement** is the most common procedure performed.

Phlebotomy

N.B. Phlebotomy is indicated under two sets of circumstances: patients symptomatic of hyperviscosity in a nondehydrated state, most commonly when hematocrit levels > 65-70; in asymptomatic patients with hematocrit levels > 65 before surgery to minimize postoperative bleeding.

- * If symptoms occur with hematocrits < 65, evidence of iron deficiency should be sought.
- * When indicated, removal of **500 mL** of blood over 30-45 minutes followed by quantitative volume replacement with normal saline or dextran can be performed.
- * The procedure may be repeated every 24 hours until improvement occurs.
- * An attempt should be made to achieve symptom control with the smallest quantity of blood removal to avoid iron deficiency.
- * Prophylactic phlebotomy has no place in the prevention of cerebral arterial thrombosis.

(b) PDA

- * Isolated PDA is an indication for **trans-catheter device closure** by cardiologists, mainly to prevent bacterial endocarditis.
- * If the patient is asymptomatic, waiting until **6-12 months** of age.
- * For large-sized PDA, **surgical closure** is available option.
- * If so small PDA, but no continuous murmur on auscultation → called "**silent ductus**" may no need for intervention.

(c) VSD

Strategies depend, to a large degree, on the size of the VSD.

Defect size	What to do
In small VSDs	Reassurance and follow up.
In moderate-sized defects	Treatment of heart failure if present, should be undertaken. Markedly enlarged left ventricle are probably indications for surgical closure.
In very large defects	The heart failure should be treated aggressively. If the congestive heart failure is difficult to control with medics, surgical closure should be undertaken.

Resistance	What to do
When the pulmonary vascular resistance is elevated	Its response to oxygen and other vasodilator agents (NO) may be necessary to determine the suitability for surgical closure; patients with calculated pulmonary vascular resistance < 8 wood units with a Qp: Qs > 1.5 are generally considered suitable candidates for surgery; if the resistance drops < 8 units after administering oxygen or other vasodilator agents, the patient becomes a candidate for surgery.
Large VSDs with severe elevation of pulmonary resistance	(Irreversible pulmonary vascular obstructive disease) are not candidates for surgery. Symptomatic treatment and erythropheresis for symptoms of polycythemia should be undertaken. These patients may eventually become candidates for lung transplantation.

- * When surgery is indicated, **open heart surgical technique** is the treatment of choice.
- * **Transcatheter device occlusion** of VSD is amenable now for some types as muscular and peri-membranous types with sufficient septum in the subaortic region so that the device can be implanted without interfering with aortic valve function.
- * Aortic insufficiency: surgical correction is indicated if moderate to severe aortic insufficiency is present.

Note The handout does not cover transposition of the great arteries, so that part is not answered.

10 Classification of congenital heart diseases.

The simplest way is to classify them as **Cyanotic** and **Acyanotic** CHDs.

- * **Cyanotic CHD:** our scope in Cyanotic CHD is on Ebstein anomaly and Fallot Tetralogy only, both with reduced pulmonary blood flow.
 - Ebstein's Anomaly.
 - Tetralogy of Fallot "TOF".
- * **Acyanotic CHDs:** further classified to Left to right shunting and obstructive lesions.
 - Acyanotic heart defects: Left-to-right shunts
 - Atrial Septal Defect (ASD).
 - Ventricular Septal Defect (VSD).
 - Patent Ductus Arteriosus "PDA".
 - Acyanotic Obstructive lesions
 - Pulmonary Stenosis (RVOTO).
 - Aortic Stenosis "AS" (LVOTO).
 - Aortic Coarctation "COA".

11 Using the stethoscope only, mention the diagnostic findings of the following: ASD, VSD, PDA.

(a) ASD

- * S2 is **widely split and fixed** (splitting does not vary with respiration) and is the most characteristic sign of ASD.
- * A soft ejection systolic murmur of grade I-II/VI intensity (due to increased flow across the pulmonary valve and is heard best at the left upper sternal border).
- * A grade I-II/VI mid-diastolic flow rumble is heard (with the bell of the stethoscope) best at the left lower sternal border. This is due to large volume flow across the tricuspid valve.
- * There is no audible murmur because of flow across the ASD.

(b) VSD

- ① In very small defects, murmur, though begins with first heart sound, may not last through the entire systole; the shorter the murmur, the smaller is the defect.
- ② In small defects the only abnormality is a loud Ejection systolic murmur heard best at the left lower sternal border.
- ③ In medium and large defects:
 - S2 is split unless elevated PAP "pulmonary artery pressure", in which case a loud single S2.
 - A **holosystolic murmur** is best heard at the left lower sternal border and does not usually radiate although it may be heard widely over the precordium.
 - The intensity of the murmur may vary between grades II-V/VI.
 - There is no significant variation of this murmur with respiration.
- ④ A grade I-II/VI mid-diastolic flow rumble may be heard at the apex in patients with medium to large-sized VSD and large left-to-right shunts; this murmur is heard best with the bell of the stethoscope. The mid diastolic murmur is due to increased flow across the mitral valve and usually indicates a Qp: Qs greater than **2:1**.

(c) PDA

- * S1 is usually normal and S2 may be buried within the murmur.
- * In most cases, a **continuous murmur** is heard best at the left upper sternal border. The murmur begins in systole and continues through S2 into the diastole. The systolic component of the murmur crescendos up to S2 while the diastolic part decrescendo to a varying distance (time) into the diastole → **machinery murmur**.
- * In big PDA, a mid-diastolic murmur may be heard at the apex because of increased flow across the mitral valve, such a mid-diastolic murmur suggests a Qp:Qs greater than 2:1.