PIPADOR REVISION

MRCOG Part 1 Revision Notes

Congenital Heart Defects

EMBRYOLOGY • LECTURE 71

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 71: Congenital Heart Defects

Embryology • Visual revision edition

COLOUR CODE USED THROUGHOUT THESE NOTES

L→R SHUNTS	OBSTRUCTIVE	CYANOTIC	DUCT / URGENT	PREVENTION / PREGNANCY

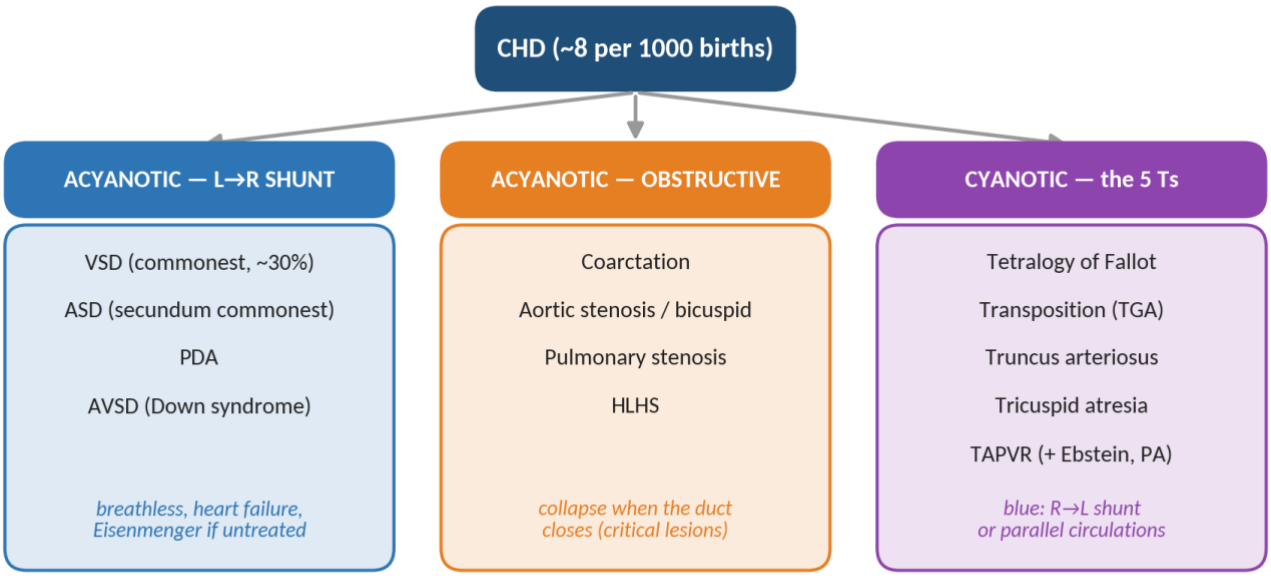
LEARNING OBJECTIVES — tick off as you go

- ☐ Classify CHD into shunts, obstructive and cyanotic lesions.
- ☐ Know the features and management of the main lesions and their embryology.
- ☐ Explain duct-dependent circulation, prostaglandin E1 and PDA closure.
- ☐ Link syndromes and maternal exposures to specific defects.
- ☐ Apply to antenatal detection, newborn screening, recurrence and pregnancy in adult CHD.

1 Classification

IN ONE LINE: *CHD ~8 per 1000 births, ~⅓ of major anomalies. Think: SHUNT (acyanotic), OBSTRUCTION (acyanotic) or CYANOTIC (the 5 Ts).*

Classifying congenital heart disease



Commonest cyanotic lesion at BIRTH = TGA; commonest beyond infancy = TETRALOGY.

Figure 1. Three groups.

2 Acyanotic Lesions

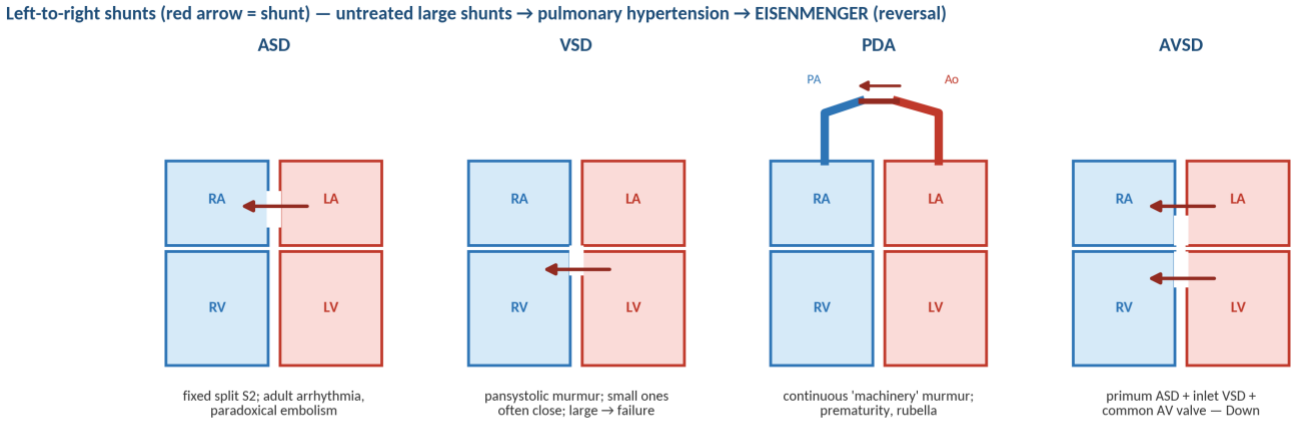


Figure 2. Left-to-right shunts.

Lesion	Key points
VSD	Commonest CHD (~30%); usually perimembranous. Small: loud pansystolic murmur, often closes. Large: heart failure, pulmonary hypertension → Eisenmenger. Device or surgical closure
ASD	Secundum (septum primum deficient) commonest; primum = cushion defect (AVSD spectrum); sinus venosus near SVC with anomalous pulmonary veins. Fixed split S2; adult arrhythmia, paradoxical embolism
PDA	Prematurity, rubella, altitude; machinery murmur. Preterm: ibuprofen/indomethacin/paracetamol; later catheter closure
AVSD	Primum ASD + inlet VSD + common AV valve. ~40–50% of babies with T21 have CHD and AVSD is the commonest; ~40–50% of AVSDs have T21. Repair in the first year
Coarctation	Near the ductus. Critical neonatal form → collapse when the duct closes; later: arm hypertension, weak/delayed femorals. Bicuspid valve ~50–60%; Turner
AS / PS	Bicuspid aortic valve 1–2% of people (stenosis, aortopathy); PS isolated or in tetralogy (Noonan)
HLHS	Tiny LV, mitral/aortic valves, ascending aorta; duct-dependent systemic flow; Norwood → Glenn → Fontan or transplant; antenatal diagnosis = planned delivery at a cardiac centre

3 Cyanotic Lesions — the 5 Ts

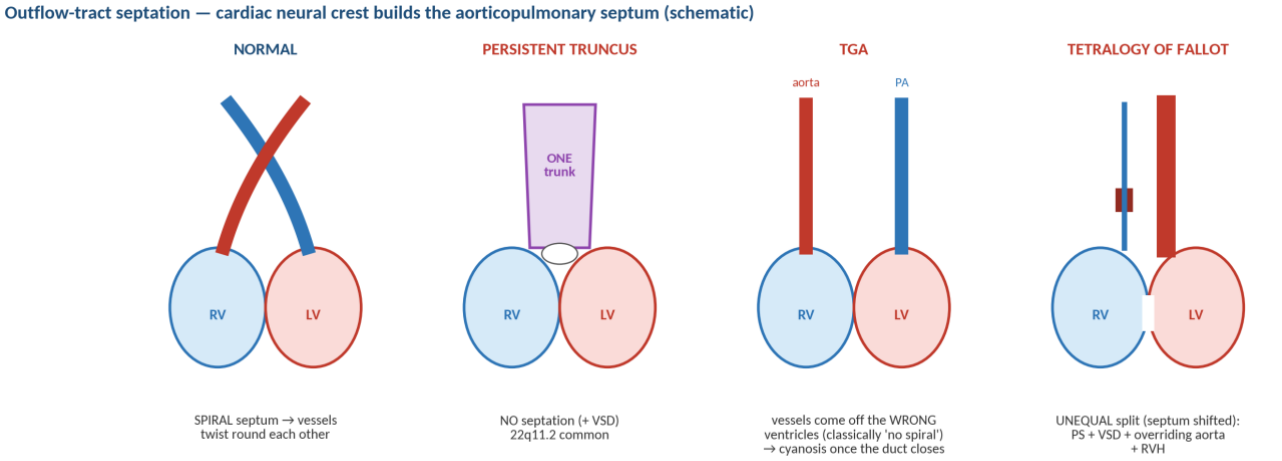


Figure 3. Outflow-tract lesions (from Lecture 70).

Lesion	Key points
Tetralogy of Fallot	Commonest cyanotic CHD beyond infancy; PS + VSD + overriding aorta + RVH from anterior malalignment of the conotruncal septum; tet spells (squatting ↑ SVR); boot-shaped heart; 22q11.2; full repair in infancy
TGA	Commonest cyanotic lesion presenting at birth; parallel circulations; cyanosis as the duct closes. PGE1 ± balloon atrial septostomy → arterial switch in the first weeks; maternal diabetes
Truncus arteriosus	One trunk + VSD; failed AP septation (neural crest); 22q11.2
Tricuspid atresia	No tricuspid valve, small RV; needs ASD/PFO + VSD or PDA; single-ventricle (Fontan) pathway
TAPVR	Pulmonary veins drain to systemic veins/RA; needs an ASD; obstructed (infracardiac) TAPVR = emergency; 'snowman' heart (supracardiac)
Others	Ebstein (low tricuspid valve, atrialised RV — lithium); pulmonary atresia

MNEMONIC / MEMORY HOOK

The 5 Ts: Tetralogy, Transposition, Truncus, Tricuspid atresia, TAPVR — and 'T21' gives AVSD.

4 Duct-Dependent Circulation

Duct-dependent lesions — the ductus arteriosus decides survival

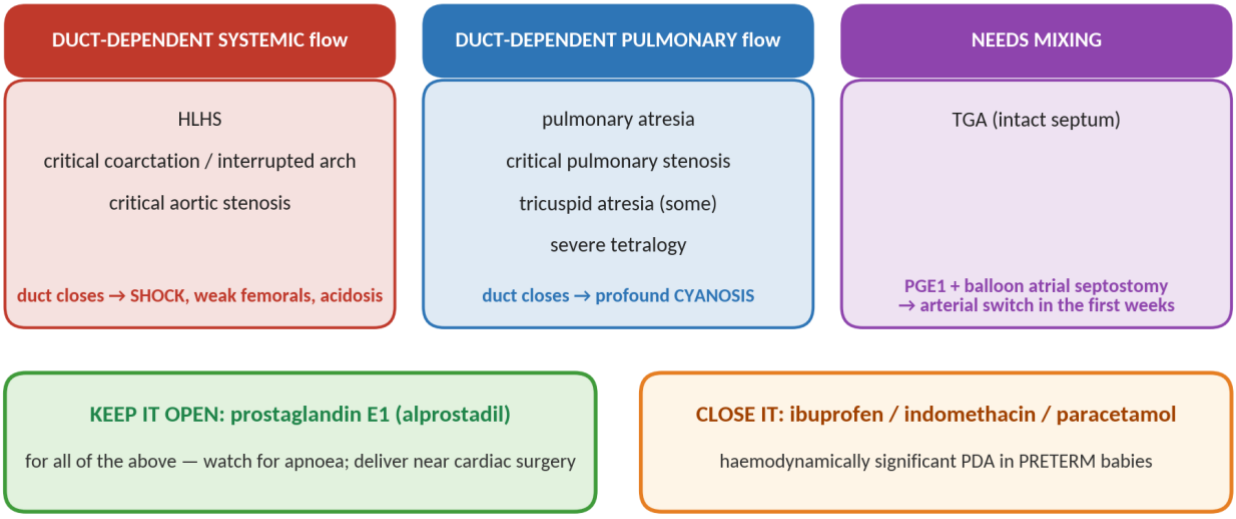


Figure 4. Keep open vs close.

5 Genetic and Maternal Associations

Syndromes and exposures — the hearts they cause



Figure 5. Syndromes and exposures.

6 Antenatal Detection and Recurrence

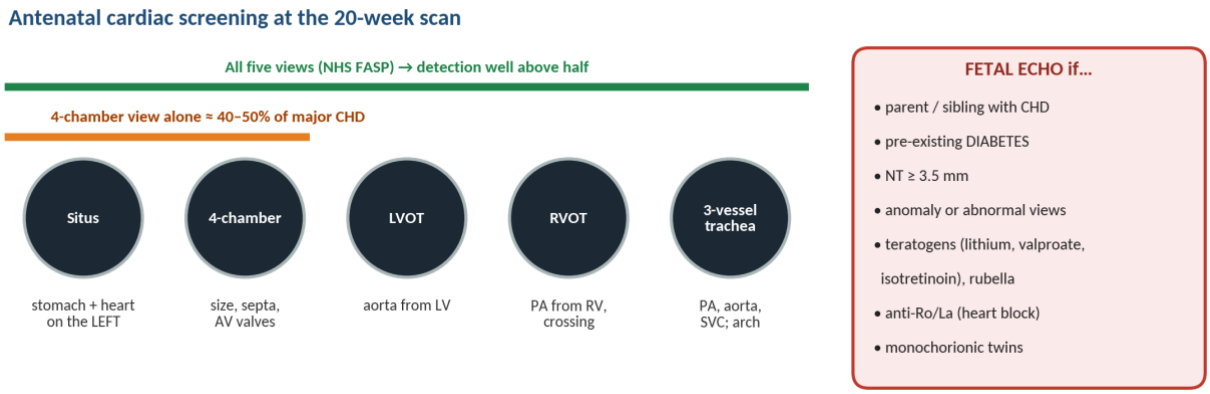


Figure 6. Screening views and fetal echo (from Lecture 70).

Situation	Approximate recurrence
One affected sibling	~2–5%
Two affected siblings	~5–10%
Affected mother	~5–10% (higher for some lesions, e.g. AVSD)
Affected father	~2–3%
Single-gene / chromosomal cause	Depends on the condition (e.g. 22q11.2 deletion 50% if a parent carries it)

7 Newborn Screening and Pregnancy

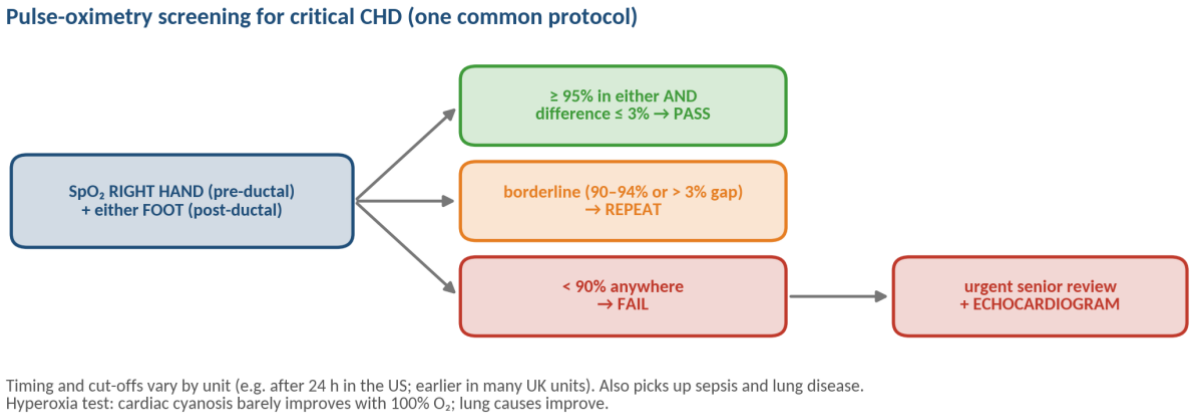


Figure 7. Pulse oximetry screening.

- Pulse oximetry is **not part of the national NHS newborn examination programme** but is used in many UK units; cut-offs and timing vary.

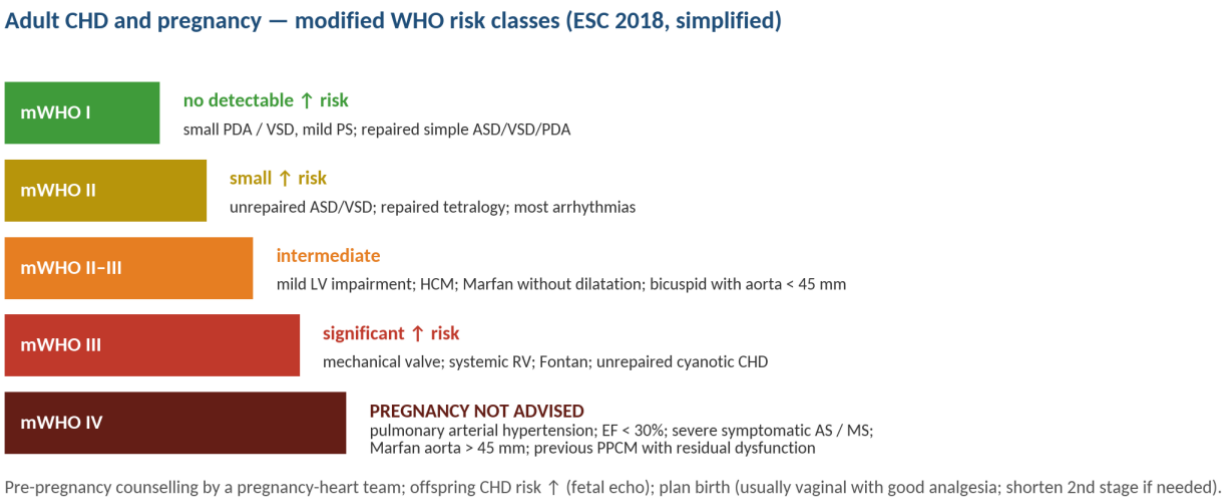


Figure 8. mWHO pregnancy risk classes.

ANTENATAL PLAN

- MDT: fetal medicine, paediatric cardiology, neonatology, surgery
- Karyotype/microarray (22q11.2, T21) when relevant
- Most CHD: **term vaginal birth** locally; **duct-dependent** lesions: deliver at a cardiac centre with PGE1 ready

PREVENTION

- Pre-pregnancy **HbA1c < 48 mmol/mol** in diabetes; **folic acid 5 mg**
- Review lithium, antiepileptics, isotretinoin
- Rubella immunity before pregnancy

★ **PEARL**

Duct-dependent lesion → keep the duct OPEN with PGE1. Preterm PDA → CLOSE it with ibuprofen/indomethacin.

Turner = coarctation + bicuspid valve; Williams = supravalvar AS; Noonan = PS; 22q11.2 = conotruncal; lithium = Ebstein.

8 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

~8/1000 CHD births	**~30%** CHDs that are VSD	**~50–60%** Coarctation + bicuspid	**1–2%** Bicuspid aortic valve
~40–50% AVSDs with T21	**≥ 95% / ≤ 3%** Oximetry pass	**< 48** HbA1c (mmol/mol) target	**~2–5%** Sibling recurrence

HIGH-YIELD CHECKLIST

- ☐ Shunts: VSD (commonest), ASD (secundum), PDA, AVSD.
- ☐ Obstructive: coarctation (Turner, bicuspid), AS, PS, HLHS.
- ☐ 5 Ts; TGA at birth, tetralogy later.
- ☐ Tetralogy = anterior malalignment; PROVe; 22q11.2.
- ☐ Duct-dependent → PGE1; preterm PDA → NSAID/paracetamol.
- ☐ T21 → AVSD; Turner → coarctation; Williams → supravalvar AS; Noonan → PS.
- ☐ Maternal diabetes → TGA, VSD, HCM; lithium → Ebstein; rubella → PDA.
- ☐ Fetal echo indications; outflow views detect more.
- ☐ Recurrence: mother > father.
- ☐ mWHO IV: PAH, EF < 30%, severe AS/MS, Marfan aorta > 45 mm.

Self-assessment — cover the right-hand column

Question	Answer
Commonest CHD overall?	VSD
Commonest cyanotic CHD presenting at birth?	Transposition of the great arteries
Embryological basis of tetralogy?	Anterior malalignment of the conotruncal septum
Drug to keep the duct open?	Prostaglandin E1 (alprostadil)
Which ASD is part of an AVSD?	Ostium primum
Syndrome with coarctation and bicuspid valve?	Turner
Cardiac lesion of Williams syndrome?	Supravalvar aortic stenosis
Maternal drug linked to Ebstein anomaly?	Lithium
Higher recurrence: affected mother or father?	Mother
Name two mWHO class IV conditions.	Pulmonary arterial hypertension; EF < 30% (also severe AS, Marfan aorta > 45 mm)

9 References

- Regitz-Zagrosek V et al. 2018 ESC Guidelines for the management of cardiovascular diseases during pregnancy.
- NICE NG3. Diabetes in pregnancy. 2015 (updated 2020).
- NHS Fetal Anomaly Screening Programme Handbook (current edition).