PIPADOR REVISION

MRCOG Part 1 Revision Notes

Turner Syndrome (45,X)

CELL BIOLOGY • LECTURE 108

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 108: Turner Syndrome (45,X)

Cell biology • Visual revision edition

COLOUR CODE USED THROUGHOUT THESE NOTES

45,X / FEATURES	MOSAIC / SERUM	HEART / PREGNANCY	SCAN / ENDOCRINE	GROWTH / ROUTES

LEARNING OBJECTIVES — tick off as you go

- ☐ Know the karyotypes, epidemiology and pathophysiology of Turner syndrome.
- ☐ Recognise antenatal findings and interpret NIPT/amnio results.
- ☐ List the clinical features and lifelong surveillance.
- ☐ Manage puberty induction and HRT.
- ☐ Counsel on fertility and the cardiac risks of pregnancy; know Y-mosaicism and Noonan.

1 Genetics

IN ONE LINE: *Loss of all or part of one X in a female — the only monosomy compatible with life. ~1 in 2000–2500 girls; ~99% of 45,X conceptions miscarry; no maternal age effect.*

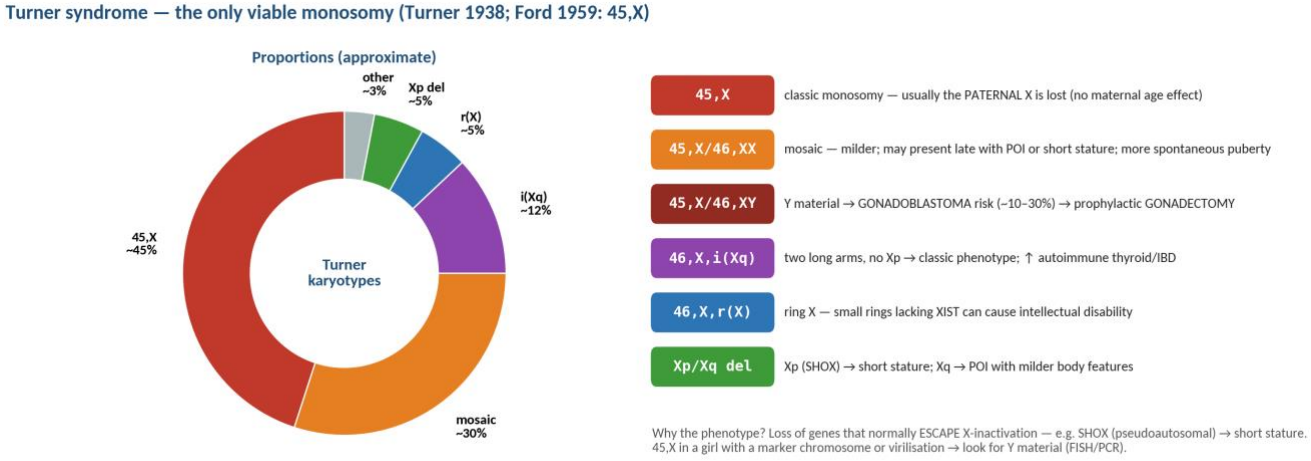


Figure 1. Karyotypes in Turner syndrome.

2 Antenatal Detection

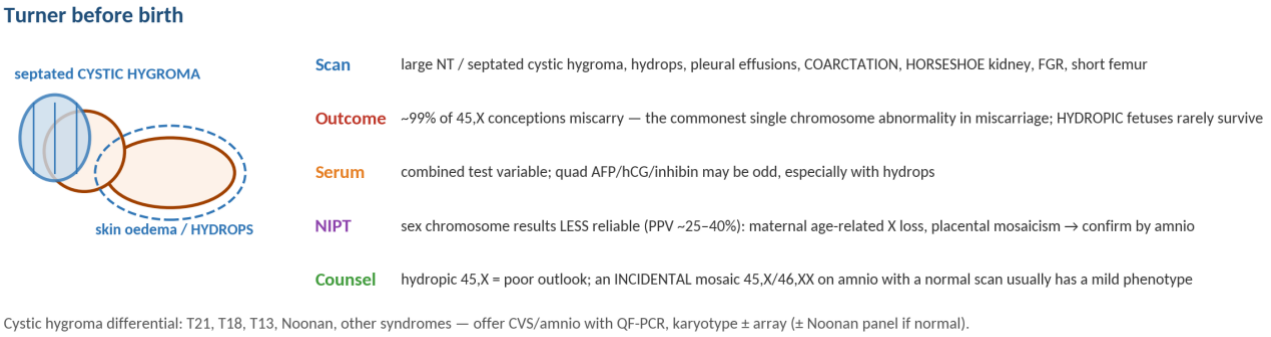


Figure 2. Turner before birth.

3 Clinical Features

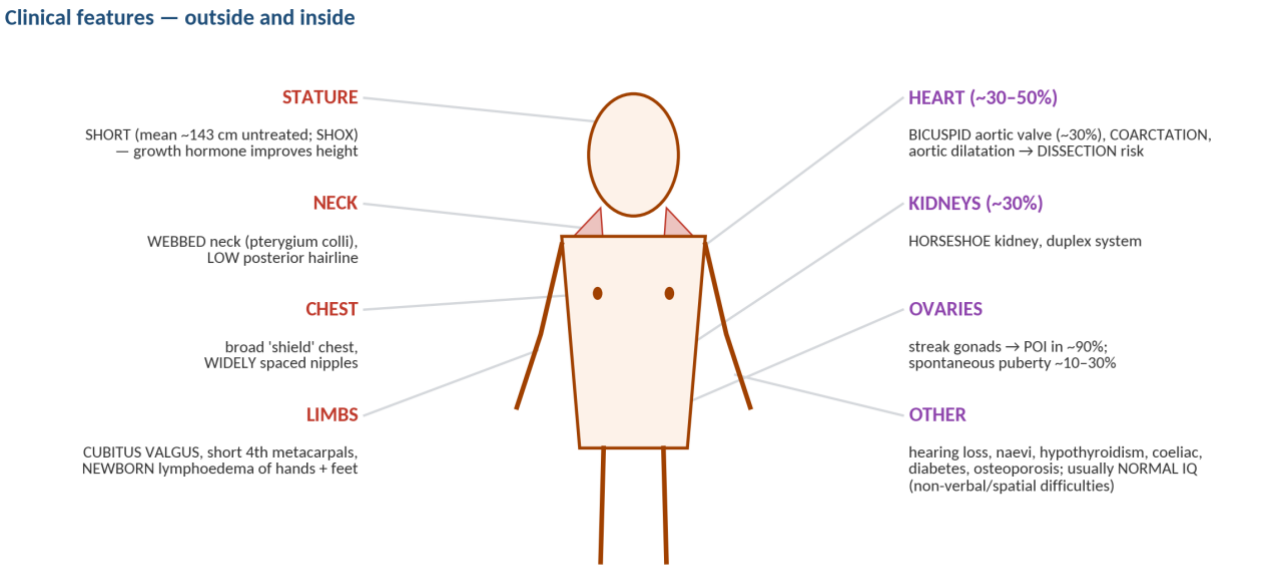


Figure 3. Clinical features.

> **MNEMONIC / MEMORY HOOK**
>
> **'CLOWNS'** — Coarctation, Lymphoedema, Ovarian failure, Webbed neck, Nipples widely spaced, Short stature (+ bicuspid valve, horseshoe kidney).

- **Diagnosis can be late**: always karyotype a girl with unexplained SHORT STATURE or delayed puberty, and women with POI (sex chromosome abnormalities in ~10% of POI; plus fragile X premutation).

4 Lifelong Care

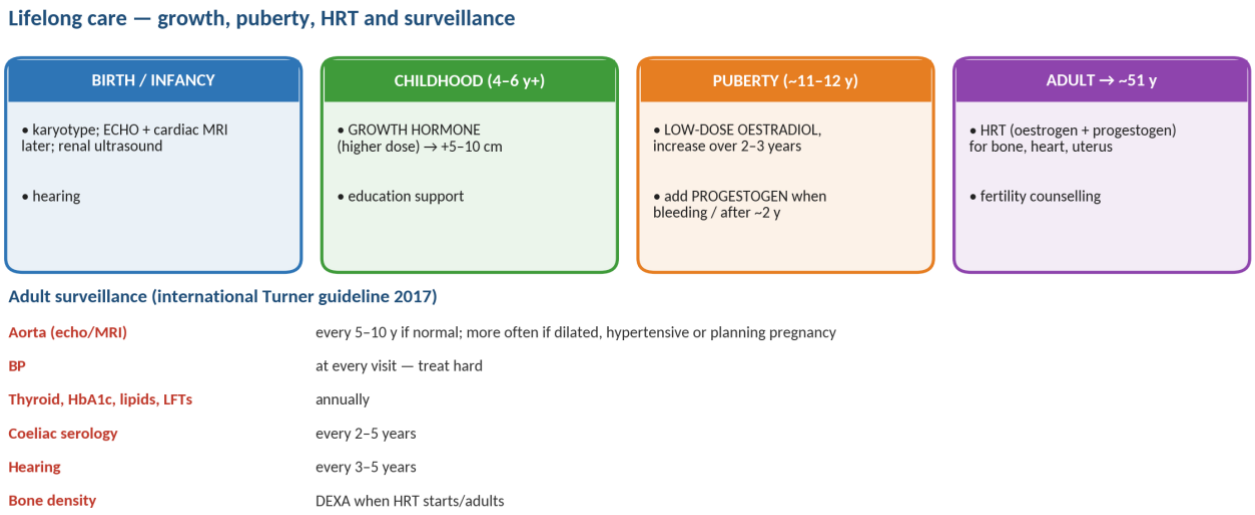


Figure 4. Growth, puberty, HRT and surveillance.

5 Pregnancy

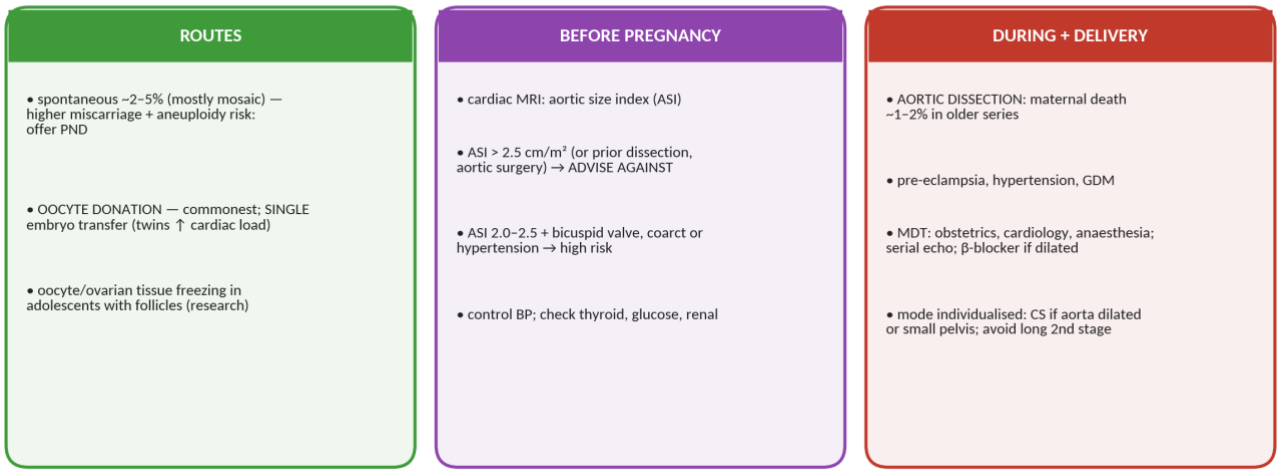


Figure 5. Pregnancy in Turner syndrome.

- **Oocyte donation**: offspring risk is that of the donor's age. **Spontaneous pregnancy**: higher miscarriage and fetal aneuploidy (older series up to ~30%) — offer screening/prenatal diagnosis. Pre-eclampsia risk → consider aspirin per NICE risk factors.

6 Y Mosaicism and Differentials

Turner vs Noonan ('male Turner' — an outdated label)

	TURNER	NOONAN
Who	females only	both sexes
Cause	loss of an X (karyotype abnormal)	AD — PTPN11 (~50%), other RAS–MAPK genes; NORMAL karyotype
Heart	LEFT: bicuspid AV, COARCTATION	RIGHT: PULMONARY STENOSIS, HCM
Gonads	streak ovaries → POI	females fertile; males cryptorchidism
Shared	short stature, webbed neck, cystic hygroma in utero	+ bleeding tendency, learning difficulty more common

Other POI differentials: fragile X premutation, autoimmune, galactosaemia, chemo/radiotherapy, Xq deletion. Short stature: constitutional, SHOX deficiency, GH deficiency.

Figure 6. Turner vs Noonan.

- **45,X/46,XY**: spectrum from Turner phenotype to mixed gonadal dysgenesis/ambiguous genitalia; dysgenetic gonads carry gonadoblastoma risk → prophylactic gonadectomy (MDT, DSD service).

★ **PEARL**

Cystic hygroma/hydrops in early pregnancy → think 45,X (and T21/T18/Noonan). NIPT sex-chromosome results need confirming.

Before pregnancy: cardiac MRI — ASI > 2.5 cm/m² → advise against. Y material → gonadectomy. HRT until ~51.

7 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

1 in 2000–2500 Live female births	**~99%** 45,X conceptions lost	**~45%** Pure 45,X	**~143 cm** Untreated adult height
~30% Bicuspid aortic valve	**~90%** POI	**2–5%** Spontaneous pregnancy	**> 2.5 cm/m²** ASI: avoid pregnancy

HIGH-YIELD CHECKLIST

- ☐ 45,X ~45%, mosaic ~30%, i(Xq), r(X), Xp/Xq deletions; no maternal age effect.
- ☐ SHOX haploinsufficiency → short stature.
- ☐ Antenatal: cystic hygroma, hydrops, coarctation, horseshoe kidney.
- ☐ NIPT sex-chromosome PPV low — confirm; incidental mosaic usually mild.
- ☐ Webbed neck, low hairline, shield chest, cubitus valgus, short 4th metacarpal, lymphoedema.
- ☐ Heart: bicuspid AV, coarctation, aortic dilatation/dissection.
- ☐ GH from ~4–6 y; oestrogen induction ~11–12 y; HRT to ~51.
- ☐ Annual TFT/HbA1c/lipids/LFT; coeliac; hearing; aortic imaging.
- ☐ Pregnancy: oocyte donation, single embryo; ASI; MDT; dissection risk.
- ☐ 45,X/46,XY → gonadectomy; Noonan = PTPN11, pulmonary stenosis, normal karyotype.

Self-assessment — cover the right-hand column

Question	Answer
Only viable human monosomy?	Turner syndrome (45,X)
Gene behind short stature?	SHOX (pseudoautosomal)
Classic first-trimester scan finding?	Septated cystic hygroma (± hydrops)
Commonest heart lesion?	Bicuspid aortic valve
Commonest kidney anomaly?	Horseshoe kidney
When is puberty induced?	~11–12 years with low-dose oestradiol
Key pre-pregnancy test?	Cardiac MRI (aortic size index)
ASI above which pregnancy is advised against?	> 2.5 cm/m²
Why gonadectomy in 45,X/46,XY?	Gonadoblastoma risk
Noonan vs Turner heart?	Pulmonary stenosis vs coarctation/bicuspid AV

8 References

- Gravholt CH et al. Clinical practice guidelines for the care of girls and women with Turner syndrome. Eur J Endocrinol 2017 (update 2024).
- RCOG Scientific Impact Paper. Pregnancy in women with Turner syndrome.
- ESC Guidelines for the management of cardiovascular diseases during pregnancy. 2018.