PIPADOR REVISION

MRCOG Part 1 Revision Notes

Trisomy 13 (Patau Syndrome)

CELL BIOLOGY • LECTURE 107

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 107: Trisomy 13 (Patau Syndrome)

Cell biology • Visual revision edition — completes the trisomy trio (105–107)

COLOUR CODE USED THROUGHOUT THESE NOTES

T13 / PATAU	TRANSLOCATION / NORMAL	T18 (comparison)	MOSAIC / ALERTS	PALLIATIVE / CONTINUE

LEARNING OBJECTIVES — tick off as you go

- ☐ Know the epidemiology and cytogenetic forms of trisomy 13, including Robertsonian translocations.
- ☐ Recognise the combined test pattern and NIPT limitations.
- ☐ Identify the scan and newborn features, especially holoprosencephaly and polydactyly.
- ☐ Distinguish T13 from T18 and from other causes of holoprosencephaly.
- ☐ Counsel on prognosis, management options and recurrence.

1 Genetics

IN ONE LINE: *Third commonest viable trisomy, defined by MIDLINE defects. Unlike T21 and T18, about one in five cases is a Robertsonian translocation — so parental karyotypes matter.*

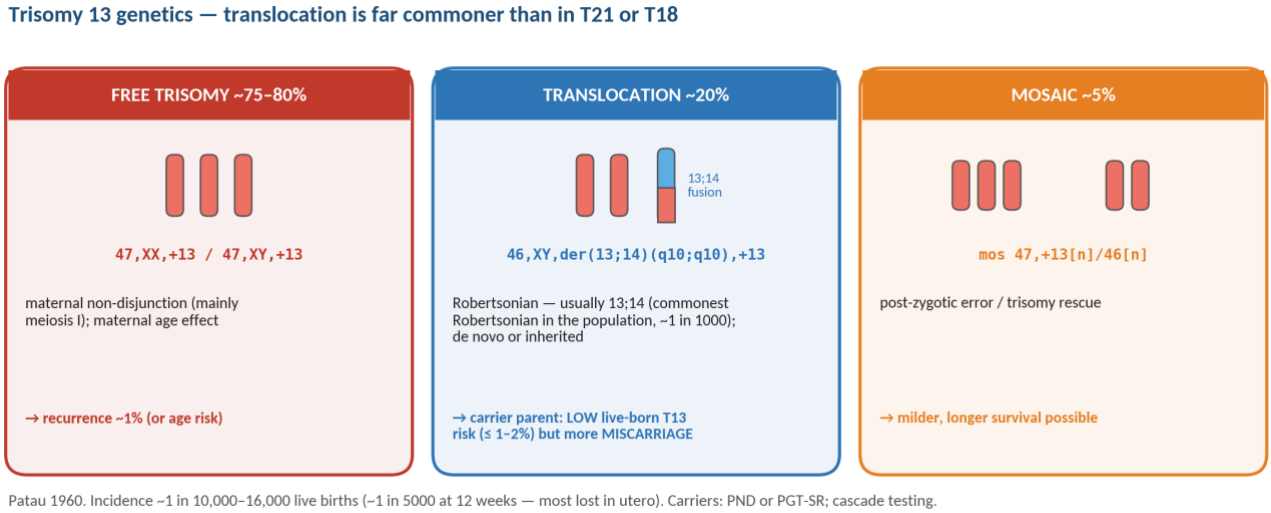


Figure 1. Forms of trisomy 13.

2 Antenatal Detection

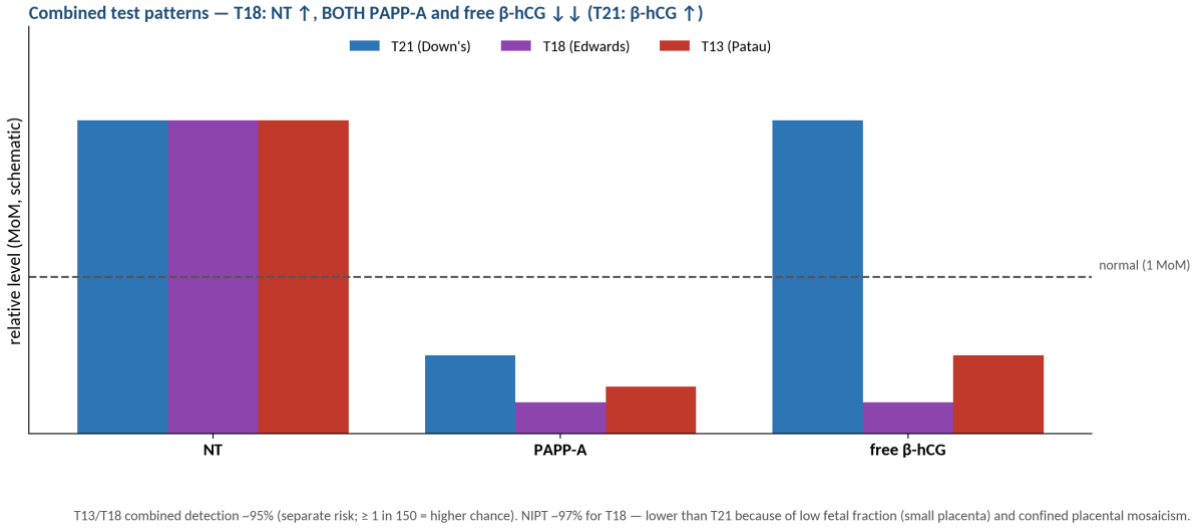


Figure 2. Combined test patterns (from Lecture 106).

- **Combined test**: NT ↑, PAPP-A ↓, free β-hCG ↓ — same direction as T18; T13/T18 risk reported together (≥ 1 in 150 = higher chance). **NIPT** detects ~90% of T13 — the lowest of the three, with more false positives (placental mosaicism) — so always confirm invasively.

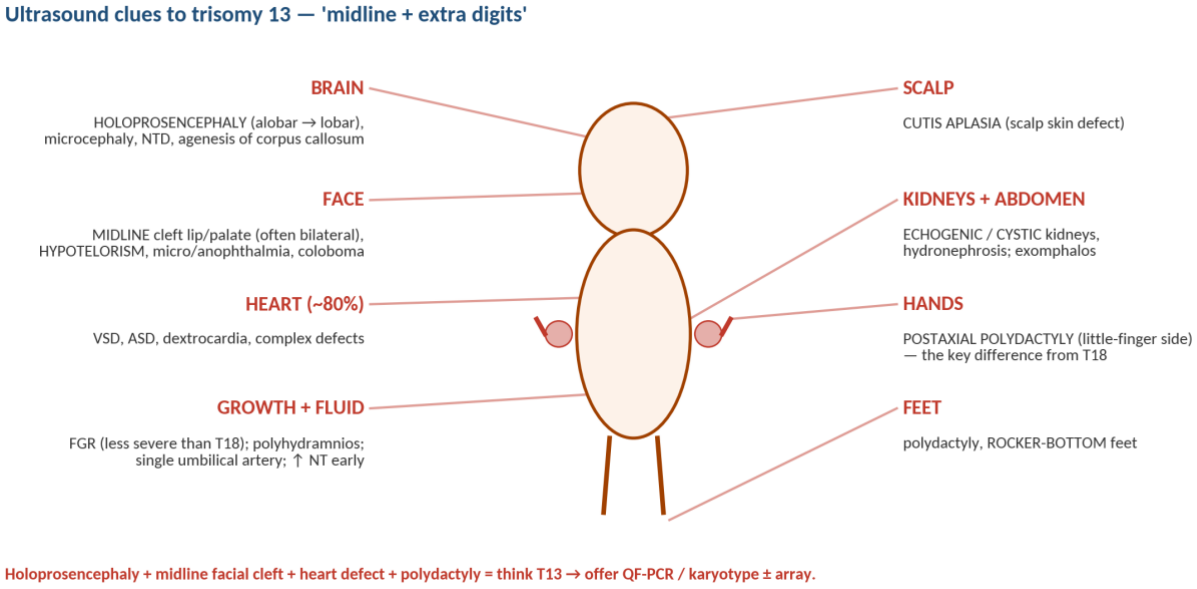


Figure 3. Scan clues.

MNEMONIC / MEMORY HOOK

'Patau = Polydactyly, Palate (midline cleft), Prosencephalon (holoprosencephaly)' — the three P's.

3 Holoprosencephaly

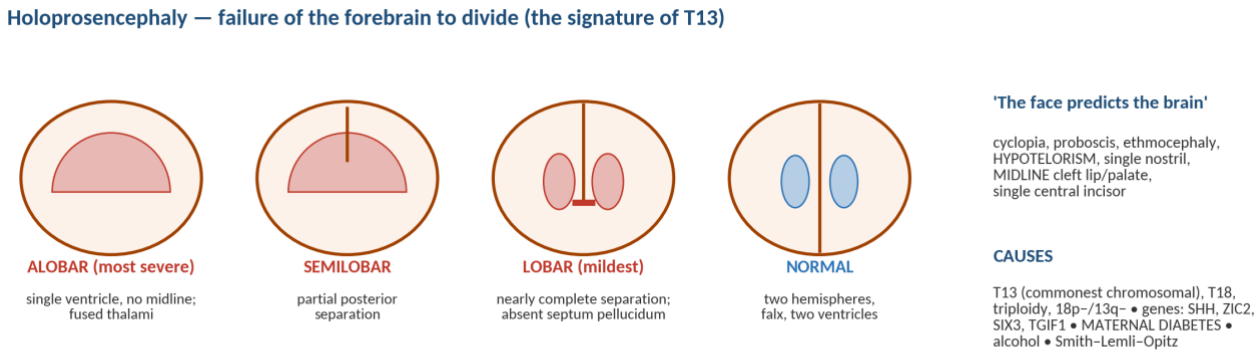


Figure 4. Holoprosencephaly spectrum and causes.

4 Clinical Features and Comparison

At birth	Features
Head + face	Holoprosencephaly, microcephaly, midline cleft lip/palate, hypotelorism, micro/anophthalmia, coloboma, CUTIS APLASIA, low-set ears
Limbs	POSTAXIAL polydactyly, overlapping fingers, rocker-bottom feet
Organs	Heart ~80% (VSD, ASD, dextrocardia); cystic/echogenic kidneys; omphalocele; cryptorchidism / uterine anomalies
Neuro	Apnoea, seizures, profound intellectual disability; tone variable

T13 vs T18 — telling them apart

	T13 — PATAU	T18 — EDWARDS
Signature	MIDLINE: holoprosencephaly, midline clefts	clenched overlapping fingers, strawberry head
Digits	POSTAXIAL POLYDACTYLY	OVERLAPPING fingers, no extra digits
Skin / eyes	cutis aplasia; micro/anophthalmia, hypotelorism	short palpebral fissures
Kidneys	echogenic / cystic	horseshoe, hydronephrosis
FGR	present, usually less severe	early and SEVERE
Translocations	~20% (rob 13;14)	~2%
NIPT detection	~90% (lowest)	~97%
Survival	median ~7–10 days	median ~5–15 days

Both: NT ↑, PAPP-A ↓, free β-hCG ↓ on the combined test; heart defects; rocker-bottom feet; high in utero loss; ~90% die by 1 year.

Figure 5. T13 vs T18.

The three common trisomies side by side

	T21 — DOWN'S	T18 — EDWARDS	T13 — PATAU
Live-birth incidence	~1 in 700–1000	~1 in 5000 (F > M 3:1)	~1 in 10,000
Main meiotic error	maternal meiosis I	maternal meiosis II	maternal meiosis I
Combined test	NT ↑ PAPP-A ↓ β-hCG ↑	NT ↑ PAPP-A ↓↓ β-hCG ↓↓	NT ↑ PAPP-A ↓ β-hCG ↓
Hallmark features	upslanting fissures, sandal gap, AVSD, duodenal atresia	clenched overlapping fingers, rocker feet, strawberry head, exomphalos	HOLOPROSENCEPHALY, midline clefts, polydactyly, cutis aplasia
Tone	HYPOTONIA	HYPERTONIA	variable
Heart defects	~40–50% (AVSD)	> 90% (VSD, valves)	~80%
Survival	~60 years	median 5–15 days; ~5–10% reach 1 year	median ~1–2 weeks; ~5–10% reach 1 year

Figure 6. T21 vs T18 vs T13 (from Lecture 106).

5 Prognosis, Management and Recurrence

- **Prognosis**: median survival ~7–10 days; ~50% die in the first week, ~90% by a year (cardiorespiratory, central apnoea, feeding). Longer survival mainly in mosaic or milder cases, with profound disability.

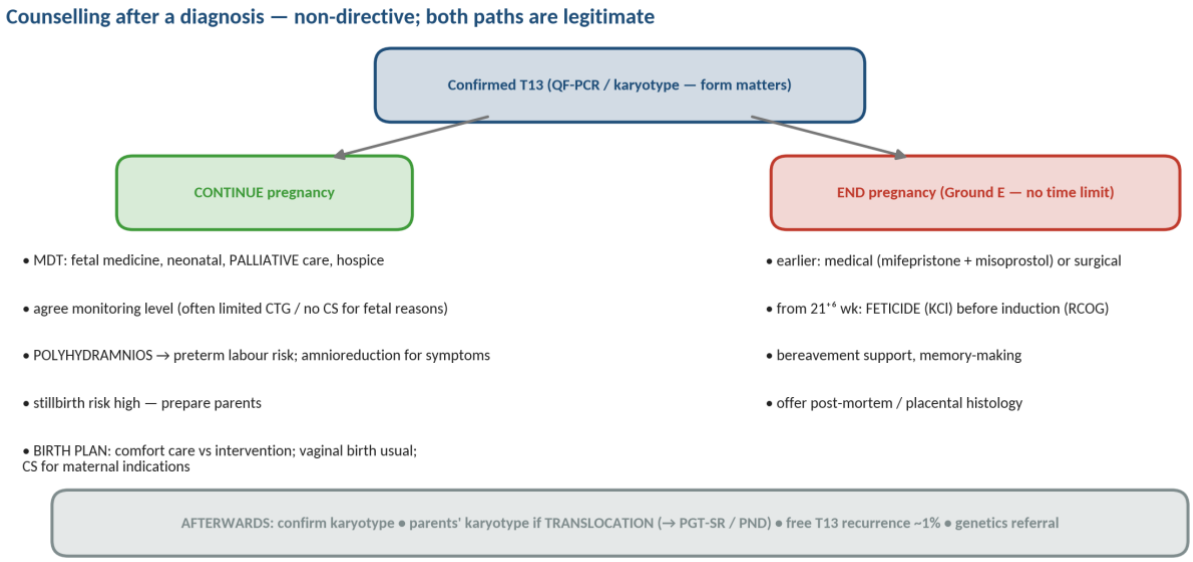


Figure 7. Counselling pathway.

Situation	Recurrence / action
Free T13	~1% (or age-related risk if higher); offer screening/NIPT or diagnostic testing next time
Parent carries rob(13;14)	Live-born T13 risk low (≤ 1–2%) but more miscarriage and recurrent loss; healthy balanced-carrier children possible; offer PND or PGT-SR; cascade testing
De novo translocation	Low recurrence
HPE with normal karyotype	Array/exome (SHH, ZIC2, SIX3, TGIF1); maternal diabetes, alcohol — optimise glycaemic control before next pregnancy

★ PEARL

T13 = midline: holoprosencephaly + midline cleft + POSTAXIAL polydactyly + cutis aplasia; heart ~80%. ~20% are Robertsonian (usually 13;14) → karyotype both parents. NIPT is least reliable for T13 — confirm invasively.

6 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

1 in 10–16k Live births	**~1 in 5000** At 12 weeks	**75–80 / 20 / 5%** Free / transloc / mosaic	**~1 in 1000** rob(13;14) carriers
~90% NIPT detection (lowest)	**~80%** Heart defects	**7–10 days** Median survival	**~1%** Recurrence (free)

HIGH-YIELD CHECKLIST

- ☐ Patau 1960; ~1 in 10,000–16,000 live births; maternal age effect.
- ☐ 75–80% free, ~20% Robertsonian (13;14), ~5% mosaic.
- ☐ Combined: NT ↑, PAPP-A ↓, β-hCG ↓ (like T18).
- ☐ NIPT ~90% — lowest; confirm by CVS/amnio.
- ☐ Holoprosencephaly (alobar → lobar); 'face predicts the brain'.
- ☐ Midline cleft, hypotelorism, cutis aplasia, postaxial polydactyly.
- ☐ Heart ~80%; echogenic/cystic kidneys; omphalocele.
- ☐ ~50% die by 1 week, ~90% by 1 year.
- ☐ Ground E or palliative birth plan — non-directive.
- ☐ Translocation: parental karyotypes, PGT-SR; HPE + normal karyotype → SHH, diabetes.

Self-assessment — cover the right-hand column

Question	Answer
Commonest chromosomal cause of holoprosencephaly?	Trisomy 13
Most severe form of holoprosencephaly?	Alobar
Digit finding distinguishing T13 from T18?	Postaxial polydactyly
Scalp finding in T13?	Cutis aplasia
Combined test pattern?	NT ↑, PAPP-A ↓, free β-hCG ↓
Which common trisomy does NIPT detect least well?	Trisomy 13
Proportion due to translocation?	~20% (usually rob(13;14))
Embryo test for a rob(13;14) carrier?	PGT-SR
Gene to test in non-chromosomal HPE?	SHH (also ZIC2, SIX3, TGIF1)
Maternal condition linked to HPE?	Diabetes (poor glycaemic control)

7 References

- Gardner RJM, Amor DJ. Chromosome Abnormalities and Genetic Counselling. 5th ed. 2018.
- NICE NG3. Diabetes in pregnancy. 2015 (updated 2020).
- RCOG. Termination of pregnancy for fetal abnormality. 2010; BAPM perinatal palliative care framework. 2023.