PIPADOR REVISION

MRCOG Part 1 Revision Notes

Trisomy 18 (Edwards Syndrome)

CELL BIOLOGY • LECTURE 106

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 106: Trisomy 18 (Edwards Syndrome)

Cell biology • Visual revision edition — follows Lecture 105

COLOUR CODE USED THROUGHOUT THESE NOTES

T18 / EDWARDS	T21 (comparison)	T13 / ENDING PREGNANCY	SCAN / ALERTS	PALLIATIVE / CONTINUE

LEARNING OBJECTIVES — tick off as you go

- ☐ Know the epidemiology, cytogenetic forms and recurrence of trisomy 18.
- ☐ Recognise the combined test pattern and NIPT performance.
- ☐ Identify the ultrasound and newborn features.
- ☐ Compare T18 with T21 and T13.
- ☐ Counsel on prognosis and both management paths, including palliative birth planning.

1 Genetics and Epidemiology

IN ONE LINE: *Second commonest autosomal trisomy but far more severe: most affected pregnancies miscarry or are stillborn, and most liveborn babies die within weeks.*

Feature	Key points
Incidence	~1 in 2500 at 12 weeks; ~1 in 5000 live births (≈ 95% lost in utero); liveborn F:M ~3:1 (more male losses)
Free trisomy ~95%	Maternal non-disjunction — mostly MEIOSIS II (unlike T21, mostly MI); strong maternal age effect; recurrence ~1% (or age risk)
Mosaic ~3%	Milder, variable; some survive to adulthood
Translocation ~2%	Unbalanced rearrangement — karyotype parents; carriers → PND or PGT-SR
History	John Edwards, 1960

2 Antenatal Detection

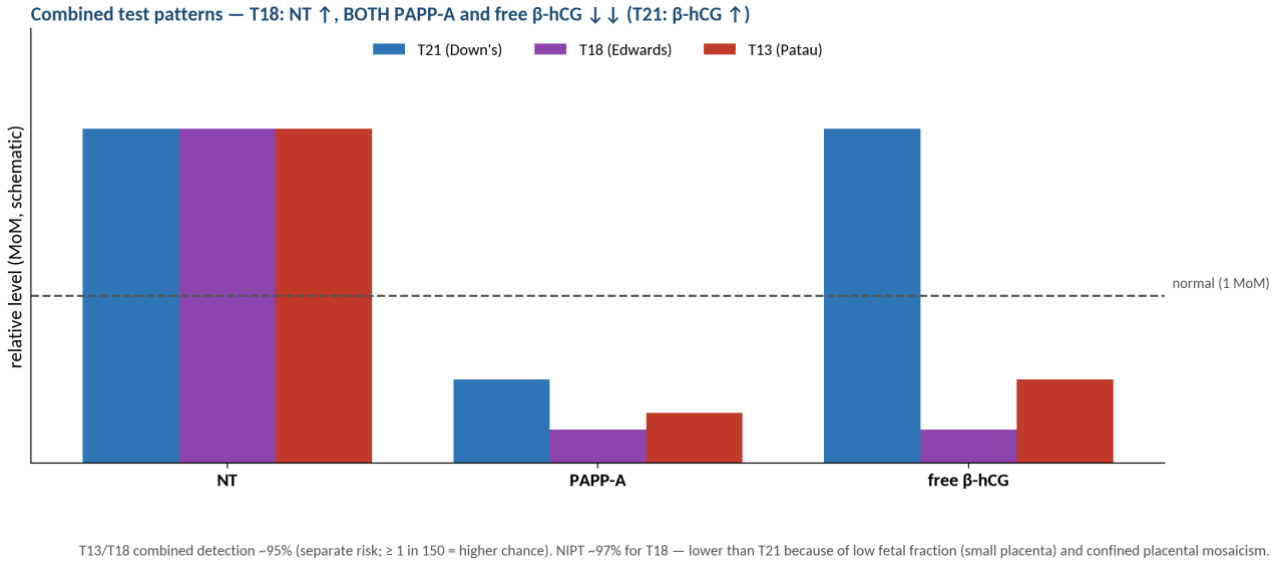


Figure 1. Combined test patterns.

MNEMONIC / MEMORY HOOK

'Edwards drops Everything' — PAPP-A AND free β-hCG both low (Down's: β-hCG UP).

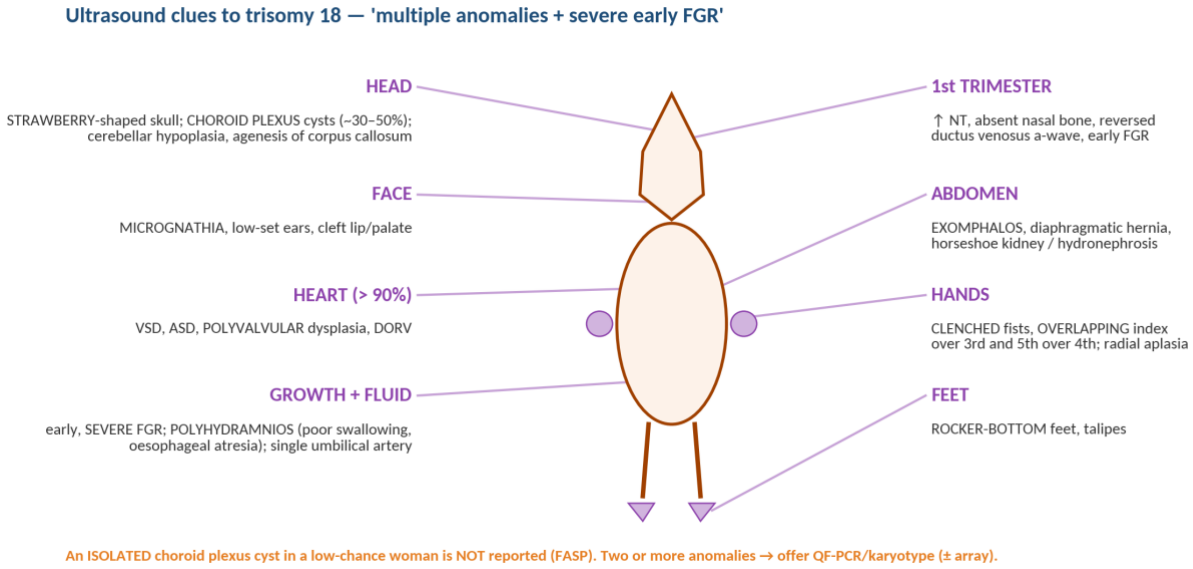


Figure 2. Scan clues.

3 Features and Comparison

At birth	Features
General	Severe FGR (~2 kg at term), microcephaly, prominent occiput, HYPERTONIA
Face	Micrognathia, low-set malformed ears, short palpebral fissures, clefts
Hands/feet	Clenched fists with OVERLAPPING fingers, nail hypoplasia, ROCKER-BOTTOM feet
Organs	Heart > 90%; renal (horseshoe), exomphalos, oesophageal atresia; brain malformations

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 3. T21 vs T18 vs T13.

- **Differential antenatally**: T13 (holoprosencephaly, polydactyly), TRIPLOIDY (digynic: severe FGR + small placenta — Lecture 103), Pena–Shokeir/arthrogryposis, CHARGE, other syndromic FGR.

4 Prognosis and Counselling

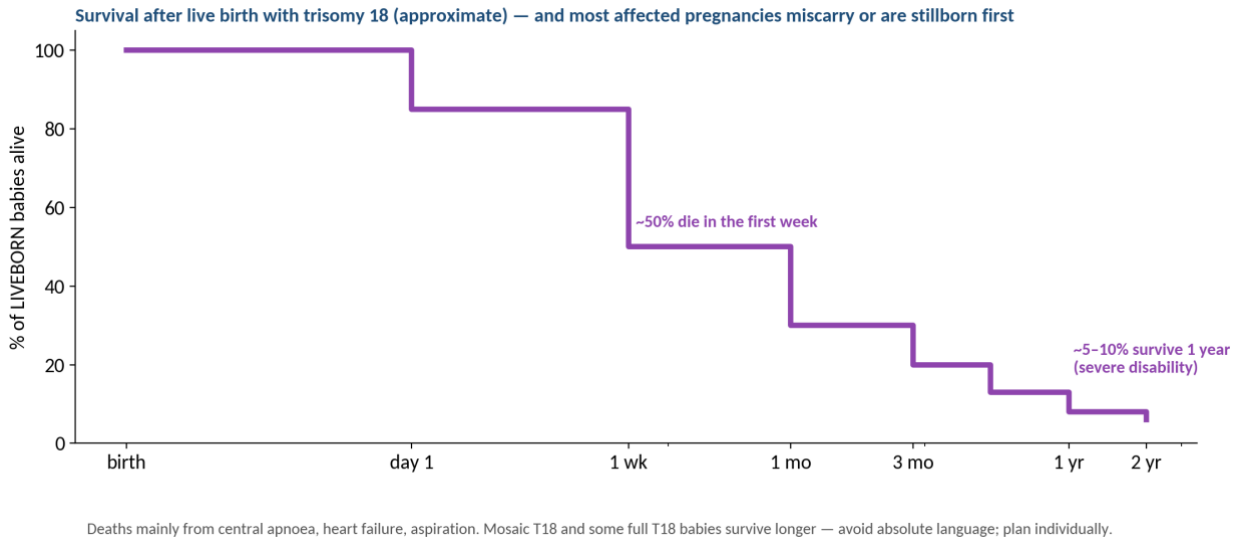


Figure 4. Survival after live birth.

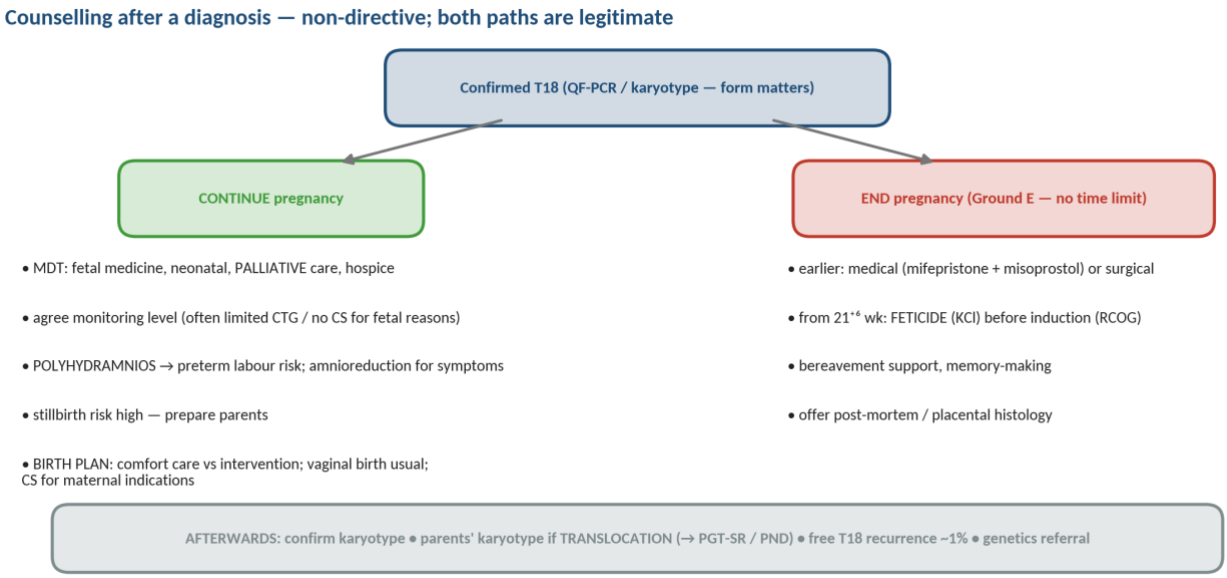


Figure 5. Counselling and management.

- **Postnatal**: confirm karyotype, offer photographs, placental histology and post-mortem; genetics referral; screening, NIPT or invasive testing offered in future pregnancies.

> ★ **PEARL**
>
> **T18 combined test: NT ↑, PAPP-A ↓↓, free β-hCG ↓↓. Clenched overlapping fingers, rocker-bottom feet, strawberry head, exomphalos, early severe FGR.**
>
> **~50% die in the first week, ~90% by a year. Continuing with palliative care or ending under Ground E are both legitimate — non-directive counselling.**

5 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

~1 in 5000 Live births	**~95%** Lost before birth	**3:1** F:M among liveborn	**95/3/2%** Free / mosaic / transloc
~95% Combined T13/18 detection	**~97%** NIPT detection	**> 90%** Heart defects	**5–15 days** Median survival

HIGH-YIELD CHECKLIST

- ☐ Edwards 1960; ~1 in 5000 live births; most lost in utero; F > M.
- ☐ 95% free (maternal MII), 3% mosaic, 2% translocation.
- ☐ Combined: NT ↑, PAPP-A ↓↓, β-hCG ↓↓; detection ~95%; NIPT ~97%.
- ☐ Scan: strawberry head, CPCs, heart, exomphalos, CDH, clenched hands, rocker feet.
- ☐ Severe early FGR + polyhydramnios + single umbilical artery.
- ☐ Isolated CPC in low-chance woman not reported.
- ☐ Hypertonia (vs Down's hypotonia); heart > 90%.
- ☐ ~50% die by 1 week, ~90% by 1 year; mosaic better.
- ☐ Ground E (no limit); feticide from 21^{+6}; or palliative birth plan.
- ☐ Recurrence free ~1%; translocation → parents' karyotype, PGT-SR.

Self-assessment — cover the right-hand column

Question	Answer
Combined test pattern in T18?	NT ↑, PAPP-A ↓, free β-hCG ↓
Which meiotic division usually fails in T18?	Maternal meiosis II
Characteristic hand finding?	Clenched fists with overlapping fingers
Characteristic foot finding?	Rocker-bottom feet
Skull shape on scan?	Strawberry-shaped
Tone in T18 vs T21?	Hypertonia vs hypotonia
Proportion with heart defects?	> 90%
Survival to 1 year?	~5–10%
Legal ground for termination at any gestation?	Abortion Act Ground E
Recurrence after free T18?	~1% (or age-related risk)

6 References

- Cereda A, Carey JC. The trisomy 18 syndrome. Orphanet J Rare Dis 2012.
- RCOG. Termination of pregnancy for fetal abnormality in England, Scotland and Wales. 2010; NHS FASP handbook.
- BAPM. Perinatal palliative care framework. 2023.