PIPADOR REVISION

MRCOG Part 1 Revision Notes

Triploidy and Tetraploidy

CELL BIOLOGY • LECTURE 111

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 111: Triploidy and Tetraploidy

Cell biology • Visual revision edition — links Lectures 95, 102, 103

COLOUR CODE USED THROUGHOUT THESE NOTES

PATERNAL / DIANDRIC	MATERNAL / DIGYNIC	PARTIAL MOLE	COMPLETE MOLE	MOSAIC / SURVIVAL

LEARNING OBJECTIVES — tick off as you go

- ☐ Explain the origins of triploidy and the complete mole.
- ☐ Contrast diandric and digynic triploidy, including scan and serum findings.
- ☐ Distinguish partial from complete hydatidiform mole.
- ☐ Manage a molar pregnancy and its follow-up (RCOG GTG 38, 2020).
- ☐ Describe tetraploidy, mosaic triploidy, endopolyploidy and detection.

1 Origins

IN ONE LINE: *Triploidy = 69 chromosomes. Two paternal sets make the trophoblast overgrow (partial mole); two maternal sets make a small placenta and a severely growth-restricted fetus. The complete mole has NO maternal set at all.*

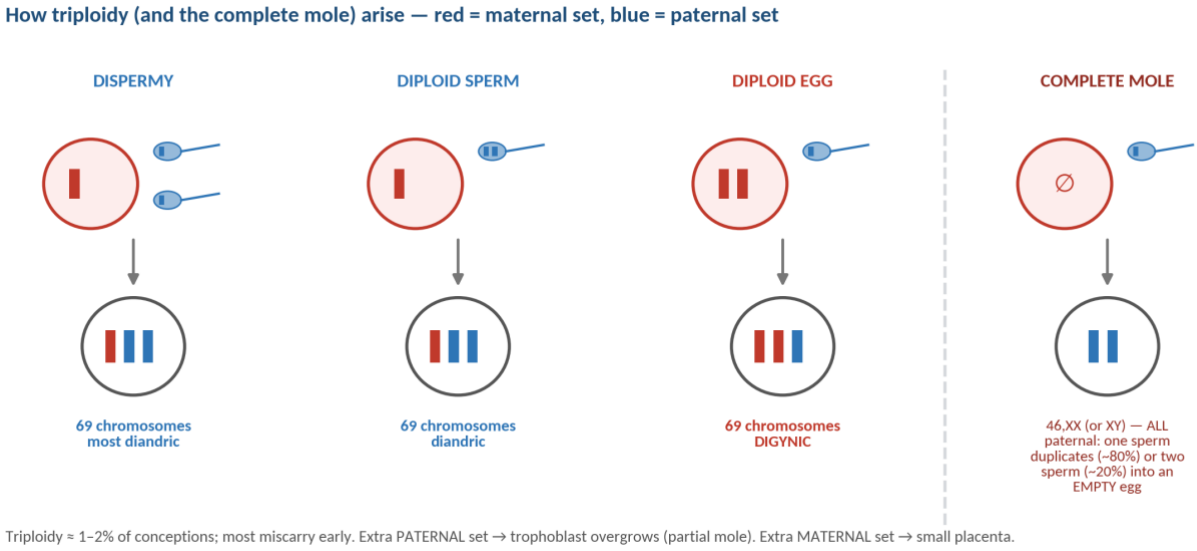


Figure 1. Origins of triploidy and complete mole.

MNEMONIC / MEMORY HOOK

'Dad's extra = big Placenta (Partial mole); Mum's extra = Miniature placenta' — the imprinting rule of thumb.

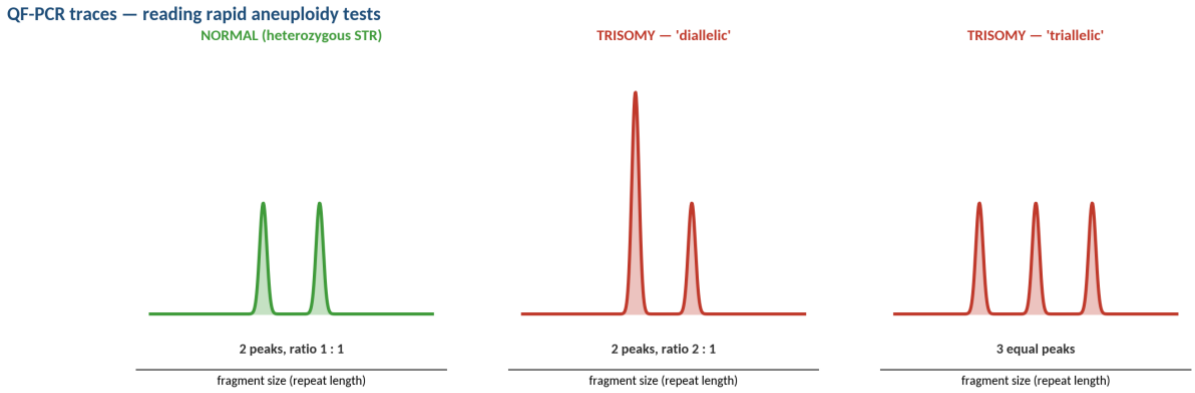
2 Diandric vs Digynic

The parent of origin decides the phenotype (genomic imprinting)

	DIANDRIC (type I) — extra PATERNAL set	DIGYNIC (type II) — extra MATERNAL set
Placenta	LARGE, cystic, hydropic villi = PARTIAL MOLE	SMALL, non-molar
Fetus	growth-restricted, often better formed; usually miscarries early	SEVERE ASYMMETRIC FGR (relatively large head), anomalies
Combined test	hCG very HIGH, PAPP-A low (mole-like)	hCG very LOW, PAPP-A very LOW (T18-like)
Mother	risk of persistent GTD (low); rarely early-onset pre-eclampsia	usually no GTD risk
Typically found	1st-trimester miscarriage / molar histology	2nd-trimester scan: FGR + small placenta

Shared anomalies: 3/4 finger SYNDACTYLY, heart defects, NTD/holoprosencephaly, cleft, renal, adrenal hypoplasia. Live-born babies die within hours–days; only diploid/triploid MOSAICS (46/69) survive (asymmetry, pigmentary streaks, ID).

Figure 2. Diandric vs digynic triploidy.



QF-PCR: several polymorphic STR markers on chromosomes 13, 18, 21, X and Y; capillary electrophoresis; result in 24–48 h after CVS/amnio.
Consistent abnormal ratios across markers on one chromosome = trisomy; also detects triploidy, sex-chromosome aneuploidy and maternal-cell contamination.

Figure 3. QF-PCR: three equal peaks suggest triploidy (from Lecture 91).

3 Partial vs Complete Mole

Complete vs partial hydatidiform mole

	COMPLETE MOLE	PARTIAL MOLE
Karyotype	46,XX (or XY) — ALL paternal (androgenetic diploid)	69,XXX / 69,XXY — DIANDRIC triploid
Fetal tissue / RBCs	absent	PRESENT (embryo/fetus, fetal red cells)
Villi	diffuse hydropic swelling, 'bunch of grapes'	MIX of normal + hydropic; scalloped; trophoblast inclusions
Trophoblast	diffuse marked hyperplasia	focal mild hyperplasia
p57 immunostain	NEGATIVE (no maternal genome)	POSITIVE
hCG / symptoms	very high; hyperemesis, thyrotoxicosis, large uterus, theca lutein cysts, early PE	modest rise; usually 'missed miscarriage'
Need for chemo (GTN)	~15%	~0.5–1%
Choriocarcinoma	rare but possible	extremely rare

Also on the differential: placental MESENCHYMAL DYSPLASIA (cystic placenta, normal fetus, normal karyotype; BWS link) and twin pregnancy with a mole + normal co-twin.

Figure 4. Complete vs partial mole.

4 Management

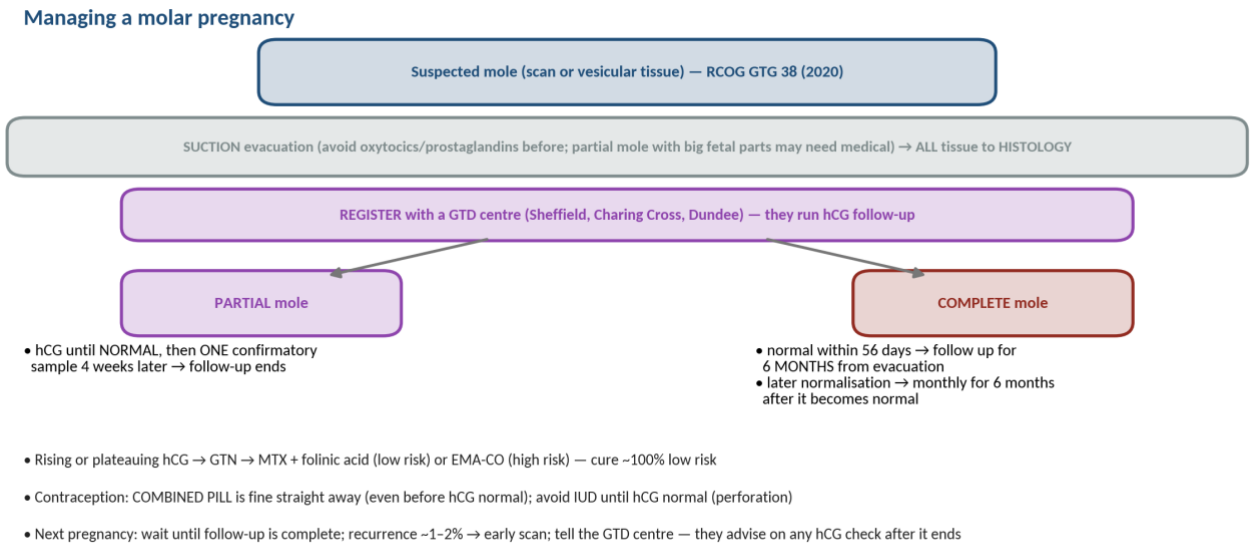


Figure 5. Managing a molar pregnancy.

Point	Detail
Histology	Send products of conception from ALL failed pregnancies after medical/surgical management where possible — molar change is often unsuspected; ploidy/p57 resolve doubt
Follow-up length	Partial: until hCG normal + one confirmatory sample 4 weeks later. Complete: 6 months (from evacuation if normal ≤ 56 days, otherwise from normalisation)
GTN after partial mole	~0.5–1% need chemotherapy (complete ~15%)
Counselling	Triploid fetus is not viable; options are expectant/medical/surgical management or ending pregnancy (Ground E); recurrence ~1–2%

5 Other Polyploidy

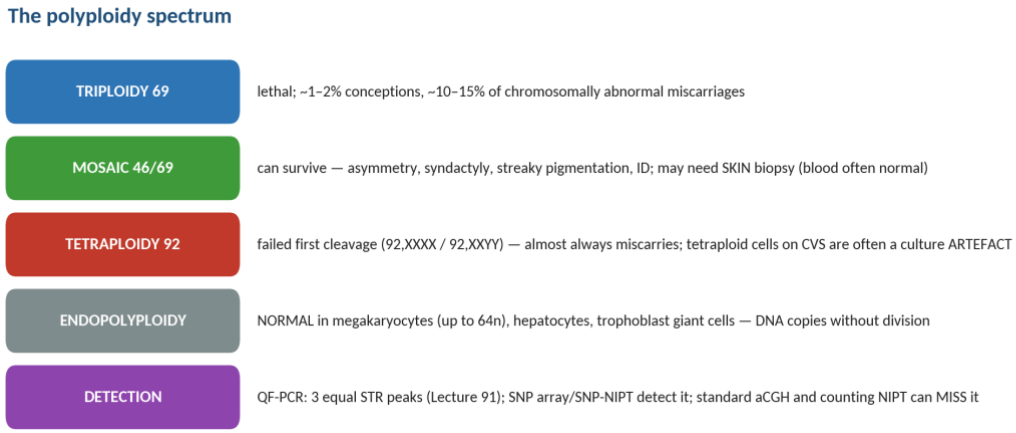


Figure 6. The polyploidy spectrum.

★ PEARL

Partial mole = diandric TRIPLOID with fetal tissue, p57 positive, GTN ~0.5–1%. Complete mole = androgenetic DIPLOID, no fetus, p57 negative, GTN ~15%.

Digynic triploidy: tiny placenta, severe asymmetric FGR, hCG + PAPP-A very low. Register all moles with a GTD centre.

6 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

69 / 92 Triploid / tetraploid	**1–2%** Conceptions triploid	**~0.5–1%** GTN after partial mole	**~15%** GTN after complete mole
4 weeks Confirmatory sample (partial)	**56 days** Complete-mole cut-off	**6 months** Complete-mole follow-up	**~1–2%** Recurrence of mole

HIGH-YIELD CHECKLIST

- ☐ Triploidy: dispermy (commonest), diploid sperm, diploid egg.
- ☐ Diandric = partial mole; digynic = small placenta + severe FGR.
- ☐ Combined test: diandric hCG ↑↑; digynic hCG + PAPP-A ↓↓.
- ☐ Syndactyly 3/4, heart, NTD, holoprosencephaly; non-viable.
- ☐ Complete mole: 46,XX androgenetic, no fetus, p57 negative.
- ☐ Partial mole: 69, fetal tissue, mixed villi, p57 positive.
- ☐ Suction evacuation; histology; register (Sheffield, Charing Cross, Dundee).
- ☐ Partial: hCG normal + 1 sample at 4 weeks; complete: 6 months.
- ☐ COC allowed immediately; IUD after hCG normal.
- ☐ Tetraploidy, 46/69 mosaics, endopolyploidy; QF-PCR/SNP array detect.

Self-assessment — cover the right-hand column

Question	Answer
Commonest cause of triploidy?	Dispermy
Triploidy with an extra paternal set gives…	Partial hydatidiform mole
Placenta in digynic triploidy?	Small, non-molar
Karyotype of a complete mole?	46,XX (or XY), all paternal
Immunostain separating complete from partial mole?	p57 (negative in complete)
Risk of GTN after partial mole?	~0.5–1%
hCG follow-up after partial mole?	Until normal, then one sample 4 weeks later
Is the combined pill allowed after evacuation?	Yes — straight away
QF-PCR pattern in triploidy?	Three equal STR peaks
Where do UK molar pregnancies get followed up?	Sheffield, Charing Cross or Dundee

7 References

- RCOG Green-top Guideline No. 38. Management of Gestational Trophoblastic Disease. 2020.
- McFadden DE, Robinson WP. Phenotype of triploid embryos. J Med Genet 2006.
- Gardner RJM, Amor DJ. Chromosome Abnormalities and Genetic Counselling. 5th ed. 2018.