PIPADOR REVISION

MRCOG Part 1 Revision Notes

Prenatal Screening

GENETICS • LECTURE 85

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 85: Prenatal Screening

Genetics • Visual revision edition

COLOUR CODE USED THROUGHOUT THESE NOTES

FIRST TRIMESTER	NIPT / NEXT STEPS	ANOMALY SCAN	2nd TRIMESTER / PE	RISK / FALSE RESULTS

LEARNING OBJECTIVES — tick off as you go

- ☐ Apply screening principles and performance measures.
- ☐ Describe the UK screening timeline, booking tests and the combined/quadruple tests.
- ☐ Explain NIPT — biology, uses, limitations and confirmation.
- ☐ Know the anomaly scan, soft markers and pre-eclampsia screening.
- ☐ Counsel non-directively, including special situations (twins, IVF, donor eggs).

1 Principles

IN ONE LINE: *Screening gives a CHANCE, not an answer. It must be offered, explained, and freely accepted or declined — diagnosis (CVS, amnio) gives the answer.*

Screening test performance

	HAS condition	does NOT
Screen +ve	a TRUE +ve	b FALSE +ve
Screen −ve	c FALSE −ve	d TRUE −ve

Sensitivity (detection rate)	a / (a + c)	how many affected we find
Specificity	d / (b + d)	how many unaffected test −ve
False-positive rate	b / (b + d) = 1 − specificity	screening fixes this (e.g. ~2–5%)
PPV	a / (a + b)	falls when the condition is RARE
NPV	d / (c + d)	

Wilson & Jungner: important problem • known natural history with a detectable early stage • acceptable test • effective action available • agreed policy on who to treat • cost-balanced • continuous process. Screening identifies RISK; diagnosis gives an ANSWER.

Figure 1. 2×2 table and Wilson–Jungner.

2 The UK Pathway

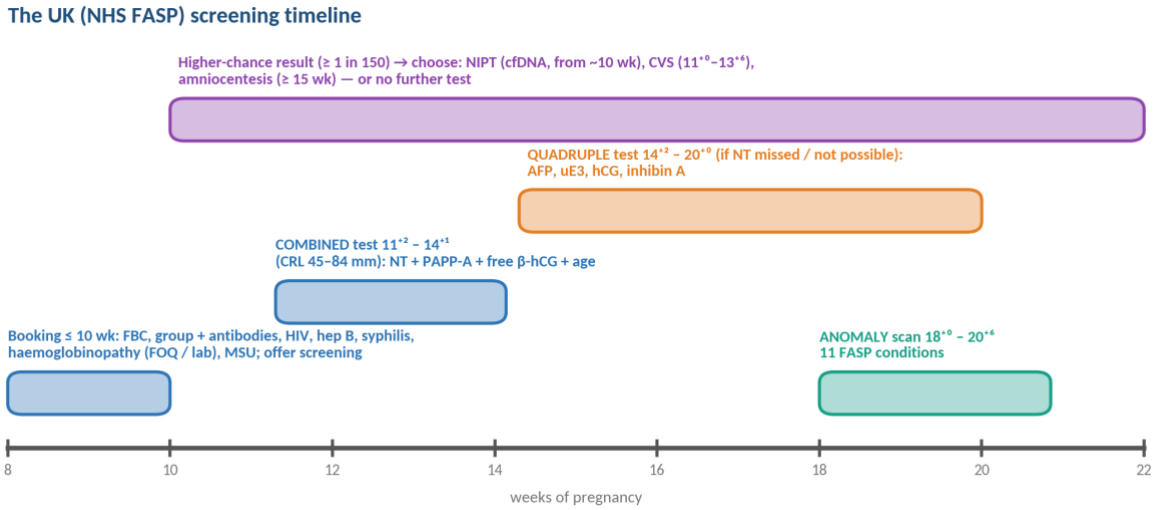


Figure 2. Screening timeline.

Booking (≤ 10 weeks)	Notes
Infections	HIV, hepatitis B, syphilis — all offered (opt-out). Rubella susceptibility screening stopped in England (2016) — check MMR history, vaccinate after birth
Blood	FBC, ABO + RhD, red-cell antibodies
Haemoglobinopathy	Lab screen (high prevalence) or family origin questionnaire + FBC (low prevalence) → partner testing (Lecture 83)
Other	MSU for asymptomatic bacteriuria; BP, BMI; pre-eclampsia + GDM risk assessment

3 Combined and Quadruple Tests

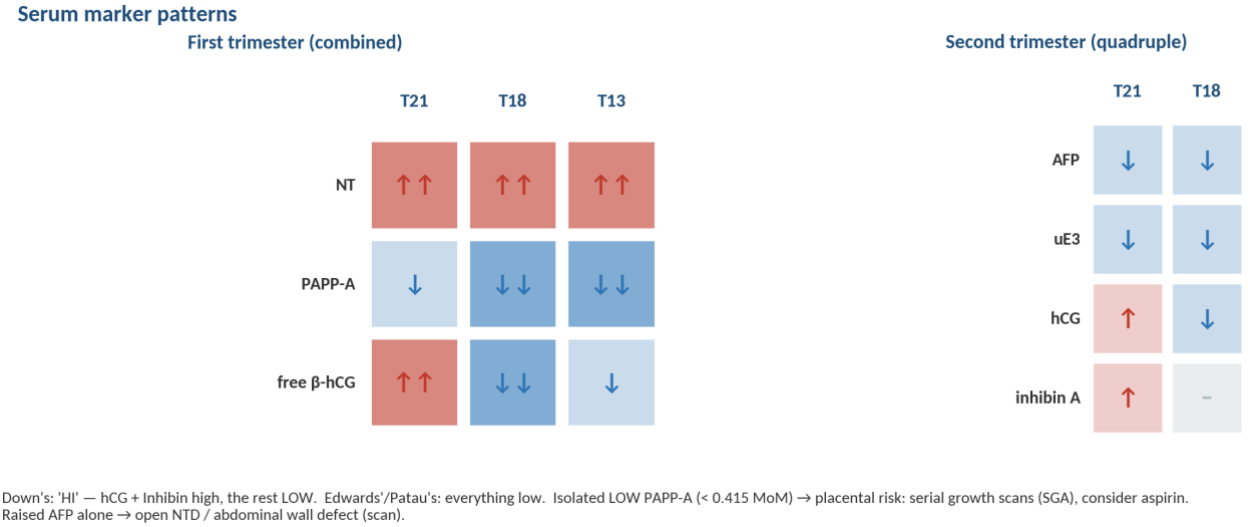


Figure 3. Marker patterns.

- **Combined test** 11^{+2} to 14^{+1} weeks (CRL 45–84 mm): age + NT + PAPP-A + free β-hCG → chance for T21 and for T18/T13; **≥ 1 in 150 = higher chance**. NT > 3.5 mm → offer detailed scan + fetal echo + diagnostic testing even if chromosomes are normal (heart, skeletal, syndromes).

- **Quadruple test** 14^{+2} to 20^{+0} weeks (T21 only) if NT not obtained or booked late. Nasal bone, ductus venosus and tricuspid flow are specialist add-ons.

MNEMONIC / MEMORY HOOK

Down's = 'HI': hCG and Inhibin HIGH, everything else LOW. **Edwards'/Patau's: all LOW.**

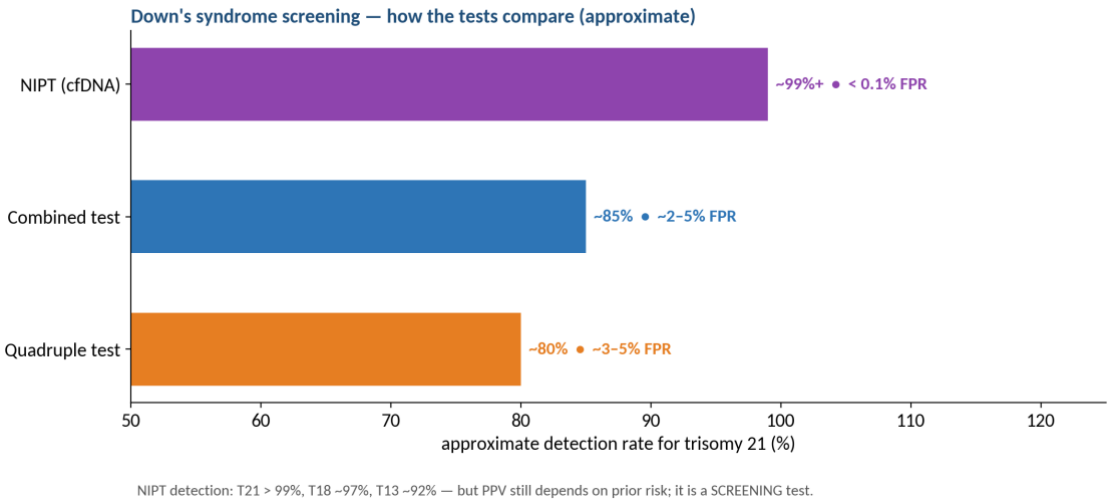


Figure 4. Detection rates.

4 NIPT

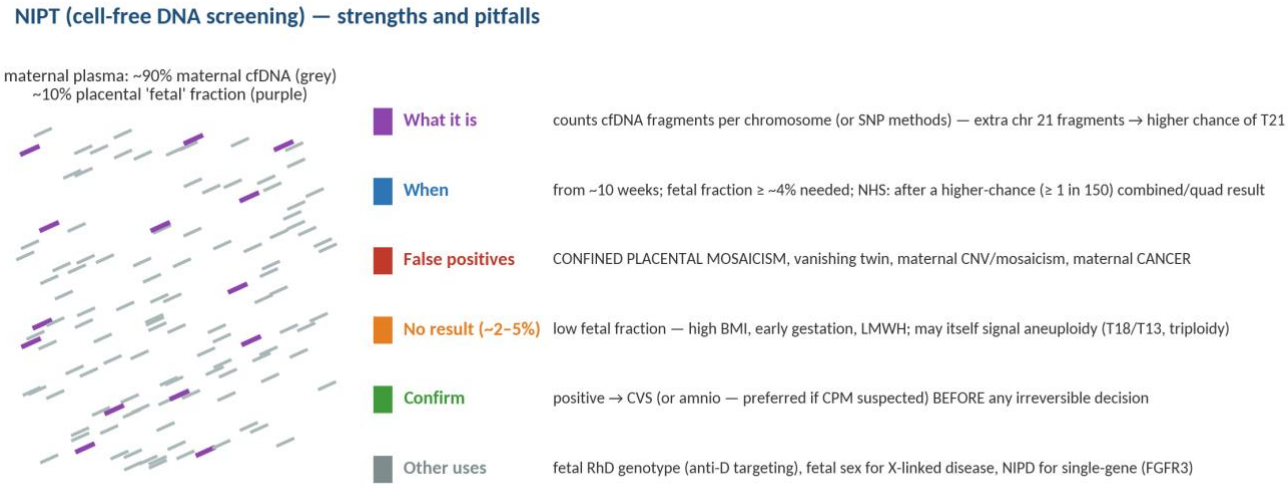


Figure 5. NIPT.

- **Rarer targets** (sex chromosomes, microdeletions) have lower PPV; NHS NIPT covers T21/T18/T13. **Twins**: combined test (NT-based) is used; the quadruple test is not offered; NIPT performs less well, especially with a vanished twin.

5 The 20-Week Anomaly Scan

- 18^{+0} to 20^{+6} weeks. FASP's **11 conditions**: anencephaly, open spina bifida, cleft lip, diaphragmatic hernia, gastroschisis, exomphalos, serious cardiac anomalies, bilateral renal agenesis, lethal skeletal dysplasia, T18, T13. Also placenta site (low-lying → rescan at 32 wk).

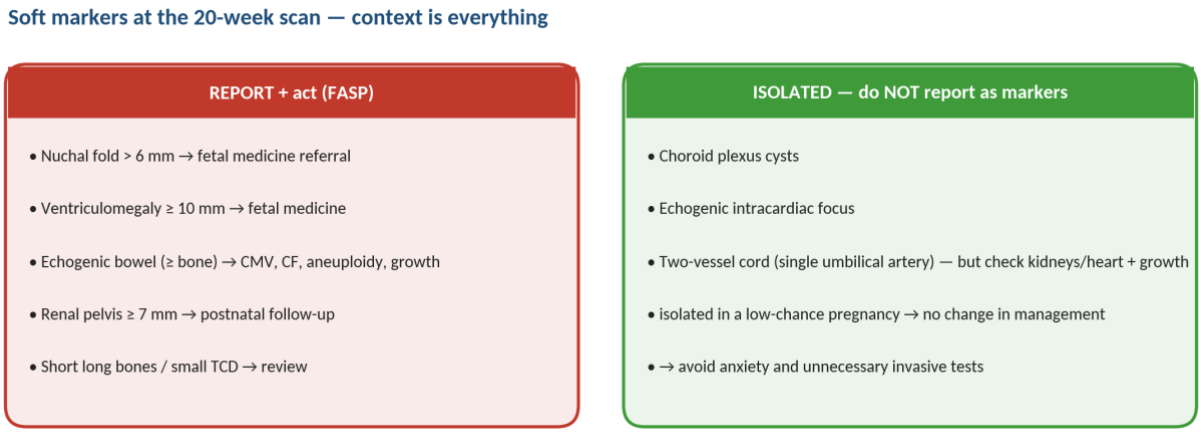


Figure 6. Soft markers.

6 Pre-eclampsia Screening

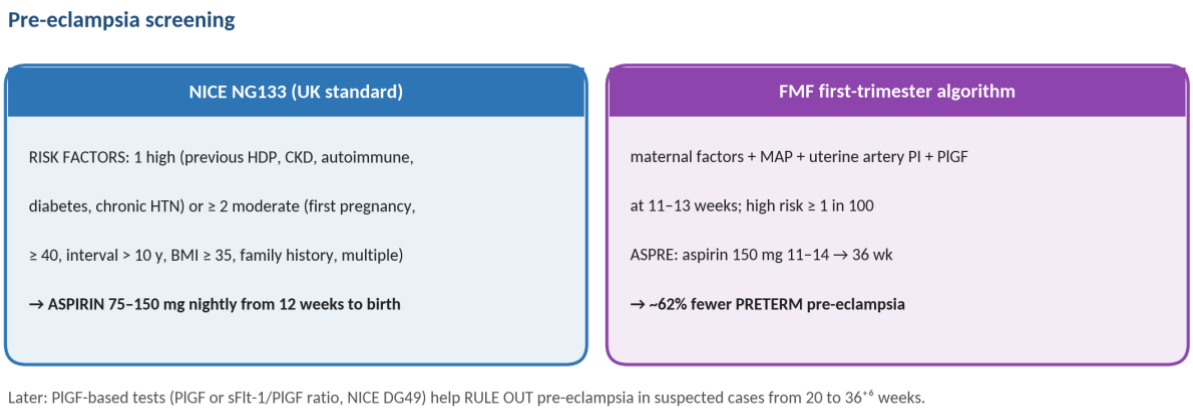


Figure 7. NICE vs FMF approaches.

7 Counselling

BEFORE + AFTER

- Chance, not diagnosis; false-positive and false-negative results; what 'higher chance' leads to
- **Non-directive**; any choice (including declining) is valid
- Higher chance → choose NIPT, CVS (11^{+0}–13^{+6}) or amnio (≥ 15 wk), or nothing; miscarriage risk from invasive tests ~0.5% or lower
- Results within the FASP standard turnaround; fetal medicine for anomalies

SPECIAL SITUATIONS

- **Twins**: chorionicity first; combined test per fetus (NT)
- **IVF/ICSI**: lower PAPP-A, slightly higher hCG — algorithms adjust
- **Donor egg**: use the DONOR's age
- **Vanished twin**: biochemistry and NIPT unreliable → NT-only
- Low PAPP-A (< 0.415 MoM) → serial growth scans

★ PEARL

Combined test 11^{+2}–14^{+1}; quad 14^{+2}–20^{+0}; anomaly scan 18^{+0}–20^{+6}; higher chance ≥ 1 in 150.
NIPT is screening, not diagnosis — confirm before any irreversible decision (CPM causes false positives).

8 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

11^{+2}–14^{+1} Combined test	**45–84 mm** CRL for NT	**1 in 150** Higher-chance cut-off	**~85% / ~80%** Combined / quad DR
> 99% NIPT T21 DR	**> 3.5 mm** NT — act regardless	**11** FASP scan conditions	**~62%** ASPRE ↓ preterm PE

HIGH-YIELD CHECKLIST

- ☐ Sensitivity, specificity, PPV (depends on prevalence), FPR.
- ☐ Booking ≤ 10 wk: HIV, hep B, syphilis, haemoglobinopathy; no rubella screen.
- ☐ Combined 11^{+2}–14^{+1} (CRL 45–84): NT, PAPP-A, β-hCG; cut-off 1 in 150.
- ☐ Quad 14^{+2}–20^{+0}: AFP, uE3, hCG, inhibin; 'HI' for Down's.
- ☐ NIPT: cfDNA ~10% placental; > 99% T21; CPM, vanishing twin, maternal cancer; confirm.
- ☐ NT > 3.5 mm → detailed scan + echo.
- ☐ Anomaly scan 18^{+0}–20^{+6}: 11 conditions.
- ☐ Soft markers: report NF > 6 mm, VM ≥ 10, echogenic bowel, RPD ≥ 7; not CPC/EIF alone.
- ☐ PE: NICE risk factors → aspirin 75–150 mg from 12 wk; FMF/ASPRE.
- ☐ Low PAPP-A → growth scans; donor egg → donor age.

Self-assessment — cover the right-hand column

Question	Answer
Window for the combined test?	11^{+2} to 14^{+1} weeks (CRL 45–84 mm)
Combined markers in T21?	↑ NT, ↓ PAPP-A, ↑ free β-hCG
Quadruple markers raised in T21?	hCG and inhibin A
NHS higher-chance cut-off?	1 in 150
Main biological cause of NIPT false positives?	Confined placental mosaicism
Is PPV affected by prevalence?	Yes — lower in rare conditions / low-risk groups
Action for NT > 3.5 mm with normal karyotype?	Detailed anomaly scan + fetal echo
Are isolated choroid plexus cysts reported?	No (FASP)
Aspirin dose and start (NICE)?	75–150 mg from 12 weeks until birth
Which age is used with a donor egg?	The donor's age at egg collection

9 References

- NHS Fetal Anomaly Screening Programme (FASP) Handbook (current edition).
- NICE NG201. Antenatal care. 2021; NICE NG133. Hypertension in pregnancy. 2019 (updated 2023).
- Rolnik DL et al. Aspirin versus placebo in pregnancies at high risk for preterm pre-eclampsia (ASPRE). N Engl J Med 2017.