

■ Evelyn & Rose Women's Health

Antenatal Testing for Chromosomal Conditions

This factsheet explains screening and diagnostic tests for chromosomal conditions in pregnancy — what they look for, who may be offered them, and their benefits and limitations. Your obstetrician will help tailor the best approach for you.

■ Who May Be Offered Testing (Indications)

- Personal preference for information about baby's chromosomes
- Maternal age or family history of chromosomal conditions
- Previous pregnancy or child with a chromosomal condition
- Abnormal or increased-risk result on a screening test
- Ultrasound findings suggesting higher chance of a chromosomal difference
- Known parental chromosomal rearrangement (e.g. translocation)

■ Screening Tests (Estimate Chance — Do Not Diagnose)

Non Invasive Prenatal Testing (NIPT) — from 10 weeks

- Analyses placental DNA fragments in the mother's blood.
- Screens mainly for **Trisomy 21 (Down syndrome), Trisomy 18, and Trisomy 13**; many panels also assess sex chromosomes; some offer optional microdeletion add ons.
- High detection rates for common trisomies; result usually within 3–7 business days.

First Trimester Combined Screening (FTCS) — 11+0 to 13+6 weeks

- Nuchal translucency ultrasound + maternal blood tests (PAPP-A and free β -hCG).
- Provides a risk estimate for common trisomies and assesses early anatomy and placental markers.
- Also allows a risk assessment for pre-eclampsia to be performed

Second Trimester Maternal Serum Screening (quadruple test) — ~15–20 weeks

- Blood test that estimates risk of certain chromosomal conditions and open neural tube defects (with ultrasound).

■ Diagnostic Tests (Confirm a Condition)

Chorionic Villus Sampling (CVS) — ~11–13 weeks

- A small placental sample is taken via abdomen or cervix under ultrasound guidance.
- Provides a definitive result for many chromosomal conditions; rapid results possible (e.g. PCR/FISH) with full karyotype/microarray taking longer.

Amniocentesis — from ~15 weeks

- A small amount of amniotic fluid is sampled with a fine needle under ultrasound.
- Diagnostic for chromosomal conditions; can also test for certain infections and genetic conditions when indicated.

■ Benefits & Limitations

Screening tests

- *Benefits:* Non invasive; early information; helps decide on further testing.
- *Limitations:* Do not diagnose; may give false positives/negatives. Results can be affected by low fetal fraction (early gestation, higher BMI), vanishing twin, placental mosaicism, or certain maternal factors. Microdeletion addons have lower accuracy.

Diagnostic tests (CVS/Amniocentesis)

- *Benefits:* Definitive diagnosis for many chromosomal conditions; can perform microarray for submicroscopic changes.
- *Risks:* Small procedure related miscarriage risk (commonly quoted **~0.1–0.3%**); cramping/spotting; infection is rare. Anti-D may be recommended if you are Rh(D) negative.

■ What Conditions Can Be Detected?

- **Trisomy 21 (Down syndrome), Trisomy 18, Trisomy 13**
- **Sex chromosome differences** (e.g. Turner syndrome, Klinefelter syndrome) — depending on the test panel
- **Sub-microscopic copy number changes** (chromosomal microarray, if performed on CVS/amnio)
- NIPT may *not* reliably detect all chromosomal or genetic conditions; detailed anatomy ultrasound remains essential.

■ Choosing the Right Test for You

- Consider how quickly you want information and whether you'd act on diagnostic results.
- Ultrasound remains important at 12 and 20 weeks for structural assessment.
- Genetic counselling is available if screening is high chance or if you have specific family history.

■ Key Takeaway

Screening gives a *chance estimate*; diagnostic tests give a *yes/no answer*. Your care team will help you balance timing, accuracy, and personal preferences to choose the option that's right for you.

Evelyn & Rose Women's Health
Personalised, evidence based antenatal care.

#PrenatalScreening #NIPT #CVS #Amniocentesis #AntenatalCare #EvelynAndRose