What's new in NIPT?

Non-invasive prenatal testing (NIPT) has become an established part of antenatal care, offering women the opportunity to clarify whether their baby is at increased risk of Down syndrome or other chromosomal conditions. Many providers are now offering a tiered model of NIPT. The "standard" NIPT screens for Trisomy 21, Trisomy 13 and Trisomy 18, with the option to "opt in" for fetal sex.

In addition to this, many NIPT platforms can now screen for **additional chromosome conditions**, though it is important to note that the test performance and accuracy may be significantly lower.

Some NIPT providers offer screening for:

- Sex chromosome aneuploidies (eg Klinefelter syndrome)
- Microdeletions and microduplications (eg VCFS or 22q11 deletion)
- Rare autosomal trisomies and segmental deletions/duplications (eg Trisomy 16)
- Triploidy

It is important for women to know the scope of testing and that invasive testing may be indicated for the confirmation of high-risk screening test results.

Patients can be referred for post-test genetic counselling and consideration of CVS/amniocentesis:

F: 9382 60385 E: seslhd-RHWPrenatalGenetics@health.nsw.gov.au

Need a refresher? Complete the 90 minute online module on First Trimester Screening and receive 3 points CPD Activity RACGP or 2 Educational Activity hours ACRRM. Available at www.genetic.edu.au or *gplearning* through the RACGP.