Non-invasive prenatal tests (NIPT)

Key points

- Near-diagnostic accuracy for trisomy 21 (Down syndrome), >99% detection.
- Tests for ≥ 80% common major chromosome problems.
- Results often available in a week.
- Blood test only, so no procedure risk unless an abnormality is detected.
- Many problems are not covered by these tests, ultrasound still recommended.
- No Medicare rebate (nor Australian private health rebate).

Non-Invasive Prenatal Tests (NIPT) are screening blood tests in pregnancy to check for extra chromosomes in the fetus - trisomies 21, 18 and 13 – with options for smaller chromosome defects and for sex chromosome anomalies. Results are based on the "fetal fraction" of cell-free DNA detected in the mother's blood. In reality, tested DNA is mainly from the *placenta*.

A prior ultrasound confirms some basics about the pregnancy (dates, live, location, number of embryos/fetuses and that there are no extra factors to consider). This may be on the day of the blood test if we have not seen you very recently.

The NIPT blood test **is performed from 10 weeks on** for women who plan to use it as the main chromosome screening. This cut off is used since the earlier in pregnancy the test is performed, the greater the chance of not getting a result. The test is usually repeated at no further cost.

Low risk women may choose the test as it is the most accurate test available for trisomies that does not carry any direct miscarriage risk. We or the referring doctor can provide guidance, usually in conjunction with a 13 week ultrasound to check for structural anomalies.

The test is also suitable for **moderately high risk** situations. However if there is a definite problem in the fetus or **very high risk** scenario is identified, NIPT may not be the ideal choice because it is not a definitive test and it does not cover the full range of chromosome problems - 20% of significant chromosome abnormalities are not checked.

Twins – NIPT can be used; like most tests it is not quite as accurate as with a single fetus, and you will need further information.

If you received a **donor egg or embryo** – you will need further information.

Decisions about how much information you wish to receive

NIPT is mainly to check for trisomies and is most accurate for trisomy 21 (Down syndrome). You also will need to make some other decisions.

1) Do you want to **be told the gender** (sex) ? This is > 99% accurate. No extra cost.
2) Do you want the **sex chromosomes checked for anomalies** - a separate decision from whether you want to know the gender. This group covers a variety of conditions ranging from very mild to severe problems. Some can be better managed in childhood if the chromosome anomaly is known from birth rather than diagnosed later in life. However since NIPT tests mainly placental DNA not fetal, it is not as accurate for these conditions. No extra cost.
3) **Extended screening eg Percept to include smaller chromosome defects** to cover some additional rare but serious conditions. This incurs a slight extra cost. The defects have to be large enough for the test to detect reliably, and won't cover all known chromosome defects. Although the test is good and picks up real conditions, there can be more false positives and negatives for parents to deal with.

If a chromosome condition is detected on NIPT, a definitive test (amnio or CVS) is needed to be sure, to avoid occasional false-positive results.

Limitations of NIPT

The test is a screening test, and the results occasionally do not truly represent the fetus for a variety of reasons. It only covers the most common chromosome defects found in pregnancy including trisomy 21, trisomy 18, trisomy 13, conditions with a large enough chromosome change, and potentially, abnormalities of the sex chromosomes. It may not detect **mosaicism** (where only some cells are affected) or many partial chromosome deletions. Although NIPT is called a "fetal" test, mainly **placental** cell-free DNA is analysed. This is not usually a problem with the common trisomies but requires careful consideration if a sex chromosome anomaly is found. Guidance given / arranged.

Many major fetal health problems are not covered by NIPT, so the usual 13+-week and 19+-week ultrasounds are still recommended for structural information.

At QDOS Ultrasound we have chosen not to be aligned with a single lab, so the choice can be tailored for each clinical situation and allow for any changes in best available options. These tests are continually developing, please discuss any specific queries.

Comments : First trimester screening (with blood for PAPP-A and free ß-hCG ideally at 10 weeks, combined with ultrasound at ideally at 12+ weeks), remains a valid alternative for some women, especially low risk women.