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# NON INVASIVE PRENATAL TESTS (NIPT)

***Information on NIPT***

***Key points***

- Near-diagnostic test for trisomy 21 (Down syndrome), >99% detection
- Tests for the 80% common major chromosome problems
- Results now often available in around 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 as yet.

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*Read on if considering this test.....*

**Non-Invasive Prenatal Tests (NIPT)** are blood tests for pregnant women which give a near-diagnostic level of accuracy for the commonest chromosome problems in pregnancy - trisomies 21, 18 and 13, as well as the option to check for sex chromosome anomalies. Results are based on the "fetal" fraction of cell-free DNA detected in the mother's blood. It is important to bear in mind that the DNA tested is mainly from the placenta, more than the fetus.

NIPT **can be performed from 10 weeks** for women who want to proceed regardless of the results of first trimester screening. The earlier in pregnancy the test is performed, the greater the chance of not getting a result, in which case the test is usually repeated at no further cost.

The test is available for **low risk women** 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 abnormalities.

The test is also suitable for **moderately high risk** situations. However where a definite problem or very high risk scenario is identified, NIPT is not usually 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 as accurate as with a single fetus, you will need further information. If you received a **donor egg or embryo** – you will need further information.

***Decision about how much you wish to test:***
NIPT is mainly to check for trisomies and is most accurate for trisomy 21 (Down syndrome). You also will need to make some other choices.

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 the NIPT test is checking mainly placenta not fetus, so be aware the NIPT is not as accurate for these conditions. No extra cost.
3. **Added microdeletion screens** for some rare but serious conditions can be offered at extra cost but also have more problems with accuracy than the main part of the test, both false positives and negatives.

It is important to understand that **if a chromosome problem such as Trisomy 21 is detected on NIPT, a definitive test (amnio or CVS) is then required to be sure**, since there are occasional false positive results.

***Limitations of NIPT:***
**T**he test only covers the 80% most common chromosome defects found in pregnancy including trisomy 21, trisomy 18, trisomy 13 and potentially, abnormalities of the sex chromosomes. It does not detect **mosaicism** (where only some cells are affected) or most partial chromosome deletions. Although NIPT is called a “fetal” test, it is really mainly **placental** DNA that is analysed. This is not usually a problem with the common trisomies but requires careful consideration if a sex chromosome anomaly is found.

Many major fetal health problems are not covered by NIPT, so the usual 12-week and 19-week ultrasounds are still recommended for structural information.

***Some other considerations are:***
- **Cost** In Perth NIPT is around $400, with no Medicare rebate as yet.
- **Length of time it takes to get a result** and
- The chance of **test failure**.

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