Patient Information

Early pregnancy screening: NIPT and CFTS, a comparison

Congratulations on your pregnancy! During this special time, your doctor may recommend different tests to ensure the health and well-being of your baby. Two of these tests are Non-Invasive Prenatal Testing (NIPT) and Combined First Trimester Screening (CFTS). This leaflet aims to provide you with easy-to-understand information about these screening options, their differences, and what you need to know.

What is NIPT?

NIPT, also known as cell-free DNA testing or non-invasive prenatal screening (NIPS), is a modern screening test for foetal chromosomal abnormalities. It is particularly effective in detecting conditions like trisomy 21 (Down syndrome), trisomy 18, and trisomy 13. NIPT involves a simple blood draw from the mother, and it analyses the baby's DNA present in the mother's blood to assess the risk of certain genetic conditions.

What is CFTS?

CFTS, or Combined First Trimester Screening, is another screening test carried out between 11+0 and 13+6 weeks of pregnancy. It involves combining ultrasound measurements (including nuchal translucency) with maternal serum analytes (hCG, oestradiol, PAPP-A) and maternal age to calculate a risk score for chromosomal abnormalities.

NIPT vs. CFTS: Key Differences

1. Accuracy: NIPT is highly accurate, with a detection rate of over 99% for trisomy 21, 96% for trisomy 18, and 91% for trisomy 13. CFTS, on the other hand, has an accuracy of around 90% for these conditions.
2. False Positive Rate: NIPT has a very low false positive rate of 0.2%, while CFTS has a higher false positive rate of about 5%.
3. Screening Range: NIPT can detect specific chromosomal abnormalities, but it does not screen for all genetic conditions. CFTS also targets certain abnormalities but may not cover the same range as NIPT.
4. Additional Testing: NIPT can reduce the need for invasive tests, such as amniocentesis or chorionic villus sampling, which carry a small risk of miscarriage. CFTS may require further testing if the risk score indicates a higher chance of chromosomal abnormalities.

Limitations of NIPT

While NIPT is a valuable screening tool, there are some important limitations to be aware of:

- It does not screen for all chromosomal abnormalities.

- It cannot replace diagnostic testing for definitive results.

- It may not be as accurate in women who are overweight, carrying more than one foetus, or have had an organ transplant or blood transfusion.

Limitations of CFTS

CFTS, too, has its limitations:

- It has a lower detection rate compared to NIPT.

- The false positive rate is higher than NIPT.

- If the risk score indicates a higher chance of chromosomal abnormalities, further invasive testing may be required.

Recommendations

- An early ultrasound scan is recommended in combination with NIPT to assess the overall health of the baby and the pregnancy.

- You also have the option to undergo both NIPT and CFTS, but this may increase the cost.

Medicare Rebates

At the time of writing, there is no Medicare item number for NIPT for the purpose discussed above. However, Medicare rebates may be available for CFTS if there is an increased risk of Down syndrome based on maternal age or other factors.

Consult Your Healthcare Provider

It's essential to discuss these screening options with your healthcare provider to determine which test is most suitable for you. They will consider factors such as your medical history, age, and any specific concerns to recommend the best screening approach.

Remember, the goal of these tests is to provide you with valuable information to make informed decisions about your pregnancy. Feel free to ask your healthcare provider any questions you may have to ensure you feel confident and supported throughout this journey.

To learn more about NIPT and CFTS, you can visit the following links:

- https://www.genetics.edu.au/PDF/Non_invasive_prenatal_testing_fact_sheet-CGE.pdf
- https://www.clinicallabs.com.au/media/1614/first-trimester-screening-patient-brochure.pdf
- https://ranzcog.edu.au/wp-content/uploads/2022/06/Prenatal-Screening-for-Chromosomal-and-Genetic-Conditions.pdf

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