Adelaide Mums & Babies Clinic

OPTIONS FOR FIRST TRIMESTER SCREENING

Below is a summary of optional screening tests in the first trimester for chromosomal trisomies in the fetus. As always, not screening is always an option. Choosing a screening test does not mean you need to progress to an invasive diagnostic test if it comes back at increased risk, and it certainly does not mean you need to progress to a termination of the pregnancy.

There is a specialist obstetric unit at the Women's and Children's Hospital that is very good at guiding families through all their options if they have an increased risk report. As always, the team at Adelaide Mums and Babies Clinic can explain things in more detail and help you make the right decision for your family.

THE FIRST TRIMESTER SCREENING TEST (FTS)

This test has been around for a long time. It is done in the first trimester and involves a blood test from the pregnant person, and an ultrasound of the fetus at 11-13 weeks, to estimate the likelihood of a baby having certain chromosomal trisomies, like Down syndrome.

This combined screening (cFTS) measures nuchal translucency (the fluid at the back of the baby's neck) via ultrasound and analyses levels of PAPP-a and free β-hCG in the mother's blood between 9 and 13+6 weeks of pregnancy. These results, along with age, ethnicity, weight, smoking status etc are put into a calculator and a risk report is generated based on statistical analysis. Results are given as “at increased risk” or “not at increased risk” with the cut off usually being 1:250. If an “increased risk” report is generated, further tests can be offered, such as NIPT and/or a diagnostic test like a Chorionic Villus Sampling (CVS) or an amniocentesis.

What it detects

Chromosomal Conditions:
Down syndrome (Trisomy 21)
Edwards syndrome (Trisomy 18)
Patau syndrome (Trisomy 13)

The ultrasound can also identify major structural abnormalities in the fetus and confirm your due date.

Accuracy

The FTS has a high detection rate, identifying approximately 85-90% of babies with Down syndrome. However, it is a screening test, not a diagnostic test, and will not definitively tell you if your baby has a condition.

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NIPT (NON-INVASIVE PRENATAL TESTING)

A safe, simple blood test that screens for chromosomal abnormalities, by analysing parts of the fetus' DNA in the pregnant person's blood. This is performed after 10 weeks of pregnancy. NIPT is another screening test. High-risk results require confirmation with invasive diagnostic tests such as amniocentesis or chorionic villus sampling (CVS). While offering greater accuracy than older methods and greatly reducing the need for invasive testing, NIPT results should be interpreted with all available clinical information, and it is not covered by Medicare or private health insurance in Australia.

Regardless of the result of this test, you will also be recommended to have an early anatomy scan at 12-13 weeks as the NIPT does not rule out structural abnormalities of the fetus caused by other issues.

How *NIPT* Works

Blood Sample: The test requires a single blood sample.
Cell-Free DNA (cfDNA): The blood sample contains cell-free DNA fragments from the developing fetus and the placenta.
Analysis: A laboratory analyses this blood to check for the chance of specific chromosomal abnormalities. Results are available in 1-2 weeks.

NIPT screens for the following chromosomal abnormalities and other:

- Trisomy 21: (Down syndrome, 99% accuracy)
- Trisomy 18: (Edwards syndrome)
- Trisomy 13: (Patau syndrome)
- Abnormalities in sex chromosomes: (e.g., Turner, Klinefelter, Triple X, XYY syndromes)
- Specific microdeletions: (e.g., 22q11.2 deletion)
- Fetal sex, if requested
- Extended panels that cost more money, can assess for many more trisomies and genetic conditions, but their sensitivity and specificity falls.

Benefits of NIPT

- *High Accuracy*: NIPT is a highly accurate screening test for chromosomal conditions. It is still a screening test, not a diagnostic test.
- *Reduced Invasive Testing*: Its increased accuracy has significantly reduced the need for invasive diagnostic procedures.
- *Low Risk*: The test is very safe for both the pregnant person and the baby, as it involves a simple blood test.
- *Early Testing*: It can be performed as early as 10 weeks of pregnancy, giving a result at 11 weeks. This allows for an earlier diagnostic test (CVS), and an earlier final result. The First Trimester Screening Test result is often not available until after 12-13 weeks, meaning a delay until 15 weeks for a diagnostic test can be done with an amniocentesis. If termination is requested, it is a simpler process the earlier this is undertaken.

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- *Cost*: In Australia, NIPT is generally a private, out-of-pocket cost and is not subsidised by Medicare or private health insurance. The cost varies, but is around $530 and done at various pathology labs. Common names include the Nest test, the Harmony test, or Generations.

Where to Get NIPT

NIPT is available through private pathology centres and specialised clinics in Australia. You will need a referral from your doctor, midwife, or genetic counsellor to have the test.

EARLY PREGNANCY STRUCTURAL ULTRASOUND

An early structural scan, also known as the first-trimester scan or nuchal translucency scan, is an ultrasound performed between 11 and 14 weeks. This scan assesses the fetus's early development and identifies potential major structural abnormalities such as issues with the brain, spine, heart, and limbs. The scan also measures the nuchal translucency (NT), a fluid-filled space at the back of the baby's neck, which can be an indicator of chromosomal conditions like Down syndrome. A thickened nuchal fold can also indicate other medical issues such as cardiac conditions, or viral infections affecting the fetus.

What the scan involves:

- *Fetal Anatomy Assessment*: A detailed examination of the fetus's early anatomy, including the brain, face, heart, spine, and limbs.
- *Nuchal Translucency (NT) Measurement*: Measurement of the fluid-filled space at the back of the fetal neck.
- *Confirming Due Date*: The scan can provide an accurate due date by measuring the fetus's length.
- *Placental and Cervical Assessment*: The location of the placenta and the length of the cervix are assessed.

Why it's done:

Early Detection of Abnormalities: To identify major structural anomalies that may not have been apparent in earlier ultrasounds, and have the potential to be picked up before the formal Anatomy scan at 20 weeks, allowing for early assessment of a potentially abnormal fetus.

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