FIRST TRIMESTER SCREENING

AUSTRALIAN Clinicallabs

Further information:
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What is First Trimester Screening (FTS)?

First Trimester Screening is the current recommended screening program to identify women with an increased risk of having an affected foetus with chromosomal aneuploidy such as Down syndrome (Trisomy 21), Edwards syndrome (Trisomy 18) or Patau syndrome (Trisomy 13). Combined FTS (cFTS) includes the Multiple of Medians (MoMs) of two blood chemistry results, pregnancy-associated placental protein-A (PAPP-A) and free β-human chorionic gonadotropin (free β-hCG), along with the ultrasound Nuchal Translucency (NT) measurement to assess the risk of aneuploidy. This assessment has 90% sensitivity and 95% specificity for Down syndrome.

Benefits of cFTS

- Recommended by clinical guidelines for pregnant women of all age groups.
- Early detection of Down syndrome with a rate of >90%.
- Reduces the number of invasive tests.
- The test is safe.
- Non-invasive.
- It can be offered to pregnant women with failed Non-Invasive Pre-natal Testing (NIPT).
- MoM values of PAPP-A could be predictive of IUGR or Pre-eclampsia risk.

PIGF for early Pre-eclampsia (PE) screening

Pre-eclampsia (PE) is one of the most common serious complications of pregnancy. Early identification of PE is an important step towards improved management and outcomes of such cases. Placental Growth Factor (PlGF) is the preferred serum marker for Pre-eclampsia prediction. PlGF biomarker has a known value in improving the performance of Down syndrome screening.

When should combined First Trimester Screening (cFTS) be tested?

- A blood chemistry test can be performed between 9 - 13.6 weeks of gestation (ideally taken at 10 weeks).
- An ultrasound examination is normally performed at 11 - 13 weeks of gestation (ideally performed at 12 weeks).

What does the result mean?

Blood chemistry results (PAPP-A & free β-hCG) are assayed using an accredited Foetal Medicine Foundation (FMF) approved platform for FTS.

FTS is not a diagnostic test. The results of cFTS will be reported as either increased risk or not at increased risk of Down syndrome. Not at increased risk means the chance of having Down syndrome is unlikely. This does not guarantee the absence of Down syndrome.

A cFTS result of increased risk does not mean the foetus definitely has Down syndrome. 1-5% of normal pregnancies are reported to have a false-positive result. Women in this group may not have a Down syndrome baby but they will be offered a highly accurate NIPT which can detect nearly all pregnancies affected by chromosomal aneuploidy or a diagnostic amniocentesis or chorionic villus sampling.

What are the Risks of cFTS?

The blood test is non-invasive and there are no known risks to the foetus in having an ultrasound in pregnancy.

Limitations of cFTS:

- Screening will not detect all cases of foetal abnormalities.
- In multiple gestation pregnancies (twins, triplets, etc.) calculation of the risk of Down syndrome or Edwards syndrome can be difficult because the amount of PAPP-A and free β-hCG is increased.

When will the results be available?

Results will be available in the next 1-2 working days following the ultrasound scan date.

How to order?

Health Practitioners can order FTS for patients using the Australian Clinical Labs Antenatal request form. Please ensure you document on the request form the following:

- Calculated gestational age and clinical due date.
- Number of foetuses (if known).
- Patient weight, height, ethnicity and family history.

It also requires history of non-insulin dependent diabetes mellitus (NIDDM) and smoking. For IVF patients, the source of egg (patient or donor), patient age at egg retrieval and the date of egg extraction will also be required. *Please note that Health Practitioners can also order Non-Invasive Prenatal Testing (NIPT-Harmony) on the same antenatal request form.*

Specimen Requirement

One serum sample (gel tube/gold top) is required.

Cost

There may be out-of-pocket costs for both the FTS and PlGF. There will be a Medicare rebate available for FTS.

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Clear **ANSWERS**
to Questions that Matter

HARMONY PRENATAL TEST is a cell-free DNA test that evaluates the risk for trisomies 21, 18, and 13.

Three Simple Steps to Clarity

1. Draw a maternal blood sample at 10 weeks or later in pregnancy
2. Submit sample directly to Australian Clinical Labs
3. Receive results in 7 days after sample receipt