Antenatal Testing at

AUSTRALIAN Clinicallabs

First Trimester Screening & NIPT Testing
NOW AVAILABLE at Australian Clinical Labs

First Trimester Screening at Australian Clinical Labs

Australian Clinical Labs is now performing Harmony (NIPT) & First Trimester Screening in-house.

- **No longer sending samples off to secondary laboratories.**
- **We will be providing results directly to the Sonographers, therefore clinics with trisomies risk profile, such as Down syndrome (Trisomy 21), Edwards syndrome (Trisomy 18) or Patau syndrome (Trisomy 13) will be able to give patients immediate results.**
- **For ultrasound clinics without trisomies risk profile, Clinical Labs will provide the risk profile back to the referrers (see results below).**
- **Quick turnaround times, i.e. results will be available in the next 1-2 days following the ultrasound.**

What is First Trimester Screening (FTS)?

First Trimester Screening (FTS) 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 (PIGF) is the preferred serum marker for Pre-eclampsia prediction. PIGF 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 10 - 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 an increased risk of Down syndrome. Not at an 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 by 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 PIGF. There will be a Medicare rebate available for FTS.

How will you receive your results?

There are 2 way's in which you may receive the Down's Risk Profile.

1. The ultrasound clinic enters the NT measurements and Clinical Labs will send you the results (as shown here).
2. The ultrasound clinic will send you the results.

Ultrasound Data

Date of Ultrasound	29/06/16	
Calculated EDD	25/12/16	
Gestational Age at US	14 + 3	
Number of Foetuses	01	
Nasal Bone	Present	
CRL	45.2	mm
Measured by	C COMPUTER CENTRE	

Biochemistry / Risk Calculations — **Screen Result : Low Risk**

Parameters	Value	Corrected MoM	Age Risk	Combined Risk	Twin Risk
Free B HCG	68 ng/mL	2.30			
PAPP-A	145.0 mU/L	0.34			
PIGF	N/A	N/A			
Nuchal Translucency	3.4 mm	1.2			
Nuchal Translucency (twin)	N/A	N/A			
Trisomy 21 (Down's)			1 in 199	1 in 350	N/A
Trisomy 18 (Edwards')			1 in 325	< 1 in 20000	N/A
Trisomy 13 (Patau's)			1 in 75	1 in 4568	N/A

Antenatal Test Available at Australian Clinical Labs

With the introduction of the FTS and NIPT testing at Australian Clinical Labs, we now provide a comprehensive range of antenatal testing. The test schedule below is the suggested range of tests to be performed throughout the course of pregnancy.

4 - 12 WEEKS

- HCG
- FBCE
- Iron Studies
- Blood Group & Antibodies
- HepBsAg
- Treponemal Serology
- Hep C
- MSU
- Vitamin D
- TSH
- HIV
- Toxo
- CMV
- Folate / B12
- Syphilis

11 - 13.6 WEEKS

- First Trimester Screen (Free B HCG, PAPP-A)
- PIGF
- Harmony - NIPT (10+ Weeks)

26 - 28 WEEKS

- Fasting Blood Glucose (FBG)
- Glucose Tolerance Test in Pregnancy
- Iron Studies

35 + WEEKS

- FBE
- Blood Group & Antibody Screen
- Strep B Streptococcal

performed in Australia

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

NOW PERFORMED BY AUSTRALIAN Clinicalabs

Australian Clinical Labs is now offering the Harmony Non-Invasive Prenatal Test (NIPT), a blood test for Down syndrome using cell free DNA (cfDNA) technology. The test can be performed as early as 10 weeks of gestation and carries a **>99% accuracy rate for Down syndrome with a low false-positive rate of <0.1%**. The test also screens for trisomies 18, 13 and it can also evaluate foetal sex and sex chromosome (X, Y) conditions.

The Harmony test can be offered to pregnant women of any age or risk category. It can be ordered for all in vitro fertilisation (IVF) singleton pregnancies, including those with egg donors. The NIPT comprehensive report includes the percentage of foetal DNA analysed for every sample, which is important in reducing the chance of false-negative results. The Harmony test is currently viewed as a screening test and invasive testing is always recommended to confirm a positive result.

Why Choose Harmony?

- Exceptional accuracy for any age or risk [1-12, 14]
  - Blinded studies in over 22,000 women of all ages [1,2,6,7,9]
  - Less than 0.1% false-positive rate for trisomies 21, 18, 13 [1-3, 5-9, 12, 14]
- Performed as early as 10 weeks
- Trusted by clinicians worldwide, over 500,000 pregnancies screened and available in over 100 countries [14]
- May minimise invasive procedures caused by false-positive results [13]

Dr Mirette Saad

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Dr Mirette Saad is a Chemical Pathologist and the National Clinical Director of the Molecular Genetic Pathology at Australian Clinical Labs. Dr Saad's current professional interests are continuing development in Molecular Genetics and Chemistry. In addition to her daily work to provide a comprehensive Chemical Pathology testing service, she currently medically supervises the Molecular Genetic testing for non-invasive prenatal testing (NIPT), hereditary disorders, personalised drug therapy and cancer. She is working closely with Health Practitioners and the laboratory team to employ the knowledge and identify opportunities to provide the best diagnostic services which include interpretation of results and the provision of important consultative advice to doctors in assisting with the management plans of patients.

How to fill out our request form

Patient Information

Please ensure all the patient information is filled in, as this is necessary for identification purposes.

Clinical Information

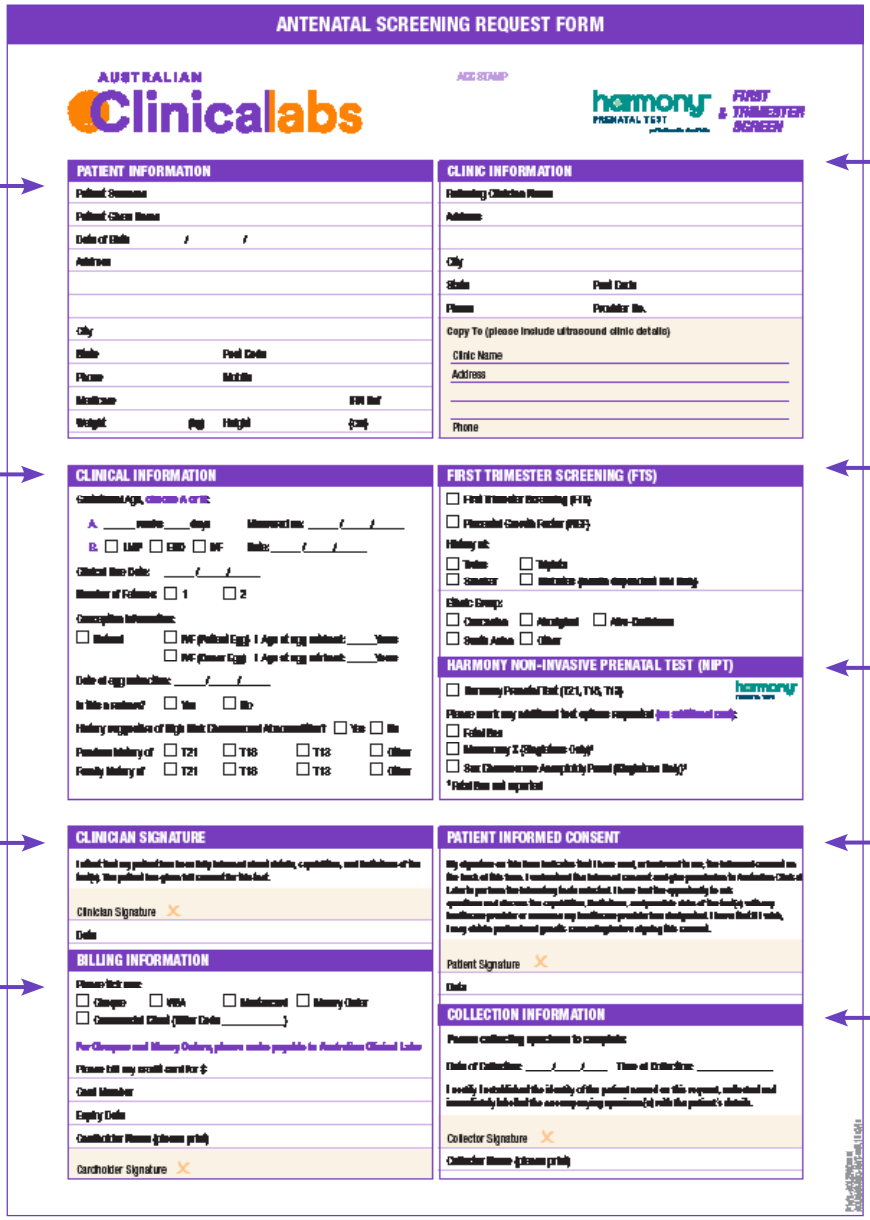
To produce an accurate result, all the clinical information here will need to be completed.

Clinician Signature

Please ensure this section has been signed.

Clinician Signature

This section is for the patient to complete. Please ensure all details have been completed and the cardholder signs.



Clinical Information

The clinic's information will need to be filled out.

Please include the ultrasound clinic details in the 'Copy To' section.

FTS Testing

If you would like a First Trimester Screening test to be completed, please ensure this section is completed.

Harmony Testing

If you would like the Harmony NIPT test to be completed, please ensure this menu selection has been completed.

Patient to Complete

Please ensure the patient has read and signed this section.

Sample Collection

This section is specifically for the person collecting the specimen to complete.

Please ensure it is signed.

How to import our template into Medical Director

1. Go to the 'Tools' drop down menu and select 'Letter Writer'.
2. From the 'File' drop down menu, please select 'New'.
3. Select 'Blank Template' and press 'ok'.
4. From the 'File' drop down menu, select 'Import'.
5. From here, you will need to locate the template, select it and press 'Open'.
6. The template will then insert into the blank document with the inbuilt template fields.
7. If they are not correct, you will need to insert them using the 'Template Field' panel to the right.
8. On completion, please click the 'File' drop down menu and select 'Save As Template'.
9. Type in the name of the template and click 'Save'. Your template has now been imported into medical director.

Please contact your BDM to receive your template.

Further information: **1300 453 688 | antenatal.clinicallabs.com.au/doctor**

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4. Sparks et al. Prenat Diagn. 2012 Jan;32(1):3-9.
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7. Nicolaides et al. Am J Obstet Gynecol. 2012 Nov;207(5):374.e1-6.
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11. Hooks et al. Prenat Diagn. 2014 May;34(5):496-9.
12. Juneau et al. Fetal Diagn Ther. 2014;36(4):282-6.
13. Wax et al. J Clin Ultrasound. 2015 Jan;43(1):1-6.
14. Data on file.

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