Non-Invasive Prenatal Screening

We are pleased to offer a new blood test to detect Down's Syndrome and other chromosomal abnormalities, called GeneSyte/GeneSyte PLUS (also known by other providers as Harmony, verify etc) The blood test looks at fetal DNA in the maternal blood that crosses the placenta. It provides screening for chromosomes 13, 18, 21 and if requested sex chromosomes.

This is a simple blood test from the mother which is safe for both for the mother and the baby, avoiding the risks of miscarriage associated with invasive prenatal testing such as amniocentesis and Chorionic Villous sampling (CVS). It is suitable for both single and twin pregnancies as well as IVF pregnancies
The test is performed as early as 10 weeks gestation up to 20 weeks gestation. It is carried out at our Women's imaging centre following a quick viability scan beforehand.

We offer a one-stop service on the day

- Viability scan
- Blood collection

We also arrange Genetic counselling if required and communicate the results to the patient and referring doctor. We also provide the 13 week structural scan.

This is a screening test and not a definitive diagnostic test. It identifies Trisomy 21 (Down's Syndrome) in greater than 99 % of cases, Trisomy 18 (Edwards Syndrome) and Trisomy 13 (Patau Syndrome) in greater than 98% of cases.
It is essential that an ultrasound is also performed at 13 weeks (in addition to the 18 week morphology scan) to assess for structural abnormalities that would not be detected with the blood test (and that may not be evident on earlier scans) This is offered at Spectrum Women's Imaging as part of our NIPS package.

Do I still need the first trimester screening bloods/PAPP-A and free BHCG (those taken as part of the nuchal translucency scan) if having the NIPS?

The recommendations are that if a NIPS blood test is being performed that these blood tests are no longer necessary. If your obstetrician or GP requests these for a specific reason then we can perform these at the same time as your NIPS at a small additional cost.

How long do the results take?

Your results will be available within 5 working days. One of our staff members will contact you with the results.
Is there a chance that the test won't work?

Sometimes the initial blood sample may be inadequate. This may be due to technical

problems. If this occurs we will contact you and arrange for you to have an additional sample collected at no extra charge.

How do I get the test?

We offer the blood test at our conveniently located Spectrum women's Imaging centre in Randwick. Your doctor will request this test along with a viability scan (to check your baby has a hear beat and that your dates are correct) performed beforehand on the same day. Alternatively, after the viability scan you can attend a Genea collection site for your blood test. We can provide more information on this.

What does it cost?

Medicare does not currently offer a rebate on this test. The payment for this test is directly to Genea and a payment slip will be provided when you have the blood test. Spectrum Women's Imaging offers a very competitive package for $700. This includes:

- Viability scan
- NIPS bloods ($445 paid directly to Genea)
- 11-13 week structural scan
- Genetic counselling if required
- Communication of results to patient

What will the results say?

A positive result indicates that the lab has detected one of the chromosomal abnormalities tested (13,18 or 21) This does not definitely mean that your baby has this abnormality. Further definitive testing is required. You will be contacted by a genetic counsellor regarding the results. The counsellors we use are highly trained and experienced in speaking with people about genetic conditions and testing. You may be advised to consider further testing such as chorionic villous sampling (CVS) or amniocentesis to confirm or disprove the result. You will be assisted to arrange this if required.

A negative result means that none of the above listed chromosomal conditions have been detected by this test. Therefore, no further invasive testing is required, however, it is important to talk to your Doctor about follow up scans at 13 weeks and 18 weeks to check for any other fetal anomalies and structural defects. These scans are essential for further assessment of the health of your baby. We offer both at Spectrum Women's Imaging.
What is the 13-week structural scan?

Even if your NIPS result comes back as negative it is essential that a structural scan is carried out at 13 weeks. This is because the NIPS blood test does not give information on other chromosomal abnormalities or conditions that your baby may have. This scan assesses the baby's head, abdomen, bladder, spine and limbs looking for early signs of abnormalities within these structures. The fluid sac behind the baby's neck (nuchal translucency) is also measured (like in a dedicated nuchal translucency scan) as this can be

increased in other chromosomal abnormalities and medical conditions such as heart defects. We offer this scan as part of our NIPS package.

It is recommended that in addition to your NIPS/GeneSyte that you have ultrasounds of your baby at 13 weeks and 18-20 weeks.