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Screening tests in First trimester of pregnancy

A screening test can help tell you the likelihood of your baby being affected by a chromosomal abnormality like Downs Syndrome. However these tests are not perfect - they do not detect all abnormalities. They also do not detect all babies affected so a low risk result does not mean a normal baby - just a low risk of an affected baby.

Combined First Trimester Screen

- Blood test at 10 wks combined with an 12-13 week ultrasound (done at either Kalgoorlie Medical Imaging or Kalgoorlie Hospital Xray department) that measures a fold behind the babies neck (the nuchal translucency)
- Calculates a risk based on those results and maternal age
- Detects 82-87% of Downs syndrome affected pregnancies
- Can also detect some other chromosomal abnormalities such as t18/13
- Ultrasound costs
 - Free at Kalgoorlie Hospital (can be difficult to get an appointment)
 - Kalgoorlie Medical Imaging - approximately \$250 (out of pocket ~\$180)
 Fees may vary for Health Care and Pensioner card holders
- 1 in 25 women will get a high risk result but most of these will not be affected babies
- High risk results can be further evaluated using the NIPT test or an CVS/amniocentesis
- For more information go to
https://www.healthywa.wa.gov.au/Articles/S_T/Screening_tests_in_the_first_3_months_of_pregnancy

Non invasive Prenatal Test (NIPT) also known as as Harmony test (Pathwest), VeriSeq (Sonic/Clinipath) or Generation NIPT (Western Diagnostics)

- Blood test from 10 wks that detects the babies free DNA in the mothers blood
- Can be used to asses risk of Downs Syndrome (t21), T13, T18 and some other chromosomal abnormalities
- This test can, in most cases, tell the sex of the baby
- Detects 99% of Downs syndrome affected pregnancies, 97% tr 18, 92% Tr 13
- Can also be used to screen for sex chromosome abnormalities however the accuracy of this is significantly lower and the majority of babies with abnormal results are in fact normal.
- Current cost is \$425 - payable online BEFORE test
- It is still recommended that women who have a NIPT test still have a 12-13 wk scan as the ultrasound can detect other abnormalities that the NIPT test does not.

For further information go to <https://www.sonicgenetics.com.au/patient/test-information/nipt/>