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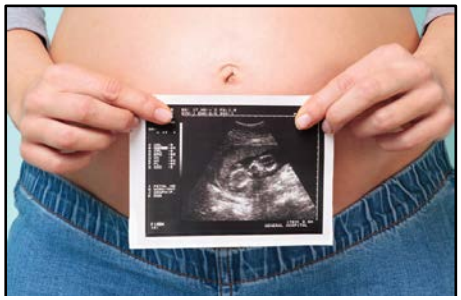
FACT SHEET

Combined First-Trimester Screening (CFTS)

The Combined First Trimester Screen has two components. Firstly, a maternal blood test is done looking at **free βHCG** and **PAPP-A** and then there is a **nuchal translucency scan**. The scan is scheduled sometime between 11 and 14 weeks' gestation. Ideally it is done close to 13 completed weeks so that the sonographer has a better look at the fetal anatomy. The scan is usually done through the lower abdomen but a vaginal scan may also be needed. It may last 20 to 60 minutes depending on how difficult it is to get good images.

Most people think that this test is to rule out Down syndrome in the fetus. No test can rule out this diagnosis 100%. The test looks at more than just the risk of Down syndrome (see table).



Nuchal Translucency Scan can detect:
Down syndrome (Trisomy 21)
Edward syndrome (Trisomy 18)
Patau syndrome (Trisomy13)
Spina bifida (defect in lower spine)
Anencephaly (missing brain)
Major heart defects
Other major defects

The blood test is best done 4 – 5 days prior to the scan. The background risk for a fetus with a chromosomal anomaly (extra chromosome) is determined by considering maternal age and gestational age. The risk is adjusted by using the results of the blood tests and the nuchal fold thickness (skin thickness at the back of the neck). If the risk for aneuploidy (chromosome abnormality) is less than 1 in 300, the test is **low risk**. If the risk is more than 1 in 300, the test is **high risk** and further testing will be recommended.

Possible further tests include:

1. **Non-invasive prenatal test**
   This is a maternal blood test that for the moment needs to be sent overseas. The cost is high at about $600 to $1000. Results return in 2 weeks.
2. **Chorionic villous sampling**
   This requires a needle to take a biopsy from the placenta under ultrasound guidance. It is normally done at about 15 weeks' gestation and has a risk of miscarriage of 1 in 200. Preliminary results take 48 hours but final results may take up to 4 weeks.
3. **Amniocentesis**
   This also requires a needle to be inserted under ultrasound guidance and draw out fluid from around the baby. It is normally done at about 16 weeks' gestation and has a miscarriage rate 1 in 300. Preliminary results take 48 hours but final results may take up to 4 weeks.

The nuchal translucency scan is performed by highly trained sonographers. Scans can be done at regular radiology practices or in specialised women's imaging practices. It makes no difference where you have the scan; it will be done to a high standard. The difference will be in the counselling you will receive following the scan. Specialised women's imaging practices will have a specialist obstetrician or radiologist explain the results to you, whereas, general radiology practices will ask you to book in with your obstetrician for the results. Hence, there is a difference in the cost for these 2 types of services.

The CFTS is not a routine antenatal screening test in Queensland hospitals but I believe it should be. I find that the test gives valuable information about the pregnancy and peace-of-mind to the pregnant couple. In a first world country like Australia, where these scans are easily accessible and, more often than not, affordable, every women planning to deliver in Australia should have this test.

Occasionally women present who were wanting to have a CFTS but for some reason have "missed the boat". For example, women have been overseas at the time the scan was due with no access to scanning or the scan was not offered to them as an option. At these times I normally recommend a Quadruple Test which is a blood test done at about 16 weeks' gestation. This test, similar to the CFTS, will give a risk assessment for chromosomal abnormalities. It does not assess early fetal anatomy. If you have any queries regarding the CFTS, please talk to your obstetrician.

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