

FIRST TRIMESTER SCREENING (FTS) Parent Information on the 12-week scan

The 12-week scan is the first detailed exam. How it is used will depend on whether you had an earlier ultrasound and what other early tests you are having. It can be a screening test for chromosome problems such as Down syndrome and for major fetal physical defects. The detail achievable at this early stage varies, is often surprisingly good, and is constantly improving. It is a check to see that the pregnancy is progressing normally in many ways. A due date can be provided if required.

All women have a risk of delivering a baby with physical and or mental disability. A common cause is chromosome abnormality, such as Down syndrome (trisomy 21). Because Down's is a serious condition which can be diagnosed during pregnancy, **screening** tests are available to help guide whether a **diagnostic** test should be offered – traditionally CVS or amniocentesis, and now the newer non-invasive **near-diagnostic** blood tests (NIPT).

The **chance of having a baby with Down syndrome increases with the mother's age**. This information is free and real. It determines the background risk for any pregnancy. The **thickness of fluid in the fetal neck (nuchal translucency, NT)** is a strong indicator of risk, especially at 11 to 13 weeks, and other ultrasound features may be helpful. Thirdly, **hormone levels measured in the mother's blood** can also help assess the risk. The **FTS blood test is most accurate at 10 weeks**, whereas the ultrasound is best at 12 ½ weeks. The three ways of assessing risk (age, ultrasound and blood test) are independent of each other, so they can be used together to make screening more accurate.

This 12 week ultrasound provides the **best ultrasound opportunity** for assessing the risk of Down syndrome in the fetus, because around 80% of Down's fetuses have an increased NT at this stage of pregnancy. International not-for-profit UK-based research in millions of pregnancies, led to a computer program combining the fetal NT thickness, the mother's age and screening blood test, which detects up to 90% of chromosome anomalies. The computer program is only released to authorised users to maintain accuracy and standards.

If you are in a high-risk age group (eg over 35 years old and not having non-invasive prenatal diagnostic tests regardless of FTS) aim for 12 weeks, otherwise, the ultrasound is more detailed if you can leave it the extra few days until 12 ½ weeks.

FTS Ultrasound – what to expect

Usually the views and measurements are obtained by ultrasound on your abdomen. Rarely a vaginal scan is required. Depending on multiple factors including fetal position, the ultrasound may be quick, or may take considerable patience. If the blood test was performed at the correct time, the estimate of risk is provided to the parents at the ultrasound appointment. We also let you know if there are other important findings.

If the risk is high, further tests will be offered to clarify whether there is a chromosome abnormality. The level of risk as well as your own preference, helps guide which test is best.

If you are expecting to deliver your baby outside Western Australia, please tell us so we can provide you with a follow-up form for audit accuracy.

DO NOT LAMINATE YOUR THERMAL PICTURES – YOU LOSE THEM.

We also provide computer images, usually email.

ALTERNATIVE SCREENING OPTIONS to the FTS ultrasound and blood test combo

NIPT

*The near-diagnostic **non-invasive prenatal test (NIPT)** is an alternative test for common chromosome anomalies, including Down syndrome. It does not assess structural problems. There is no Medicare rebate at present. It is an option in addition to the 12-week scan, picking up the 80% commonest chromosome problems, with the labs quoting > 99% accuracy for Down's. It can be performed from 10 weeks, and is often used when the risk is high enough to warrant testing but not so high as to require an urgent result. Even if NIPT is requested, a 12-week structural check is still important, as many of the findings detected at this stage are not chromosomal. There is a Park Ultrasound handout on NIPT for more info.*

Routine anatomy scan at around 19+ weeks

*This is still the most thorough ultrasound of the pregnancy to check the structures and development of the fetus, and is **advised for every pregnant woman**.*

DIAGNOSTIC TESTS – CVS OR AMNIOCENTESIS

- *Used when risk is too high for NIPT and for specific individual situations or detailed tests.*
- *All chromosomes are checked, not just the common ones.*
- *Separate more detailed written information sheets are given where relevant.*
- *Documentation of blood group is required.*
- *Rapid versions are available for both tests (usually results within 24 hours).*

Chorionic Villus Sampling (CVS) at 11 to 14 weeks

The fetus and placenta develop from the same origin, so the placenta can be used to check the chromosomes of the fetus. A fine needle is passed into the uterus through the mother's abdomen and a small sample of the placenta is taken. Local anaesthetic is used for CVS. The advantage of CVS over amnio for chromosome tests is rapidity. The risk of miscarriage due to CVS is around 1 %. It is also used for DNA tests.

Amniocentesis at 15+ weeks

A very thin needle is passed through the mother's abdomen into the uterus to take a small amount of fluid from around the fetus. The risk of miscarriage from amniocentesis is about ½ %. It is considered the gold standard diagnostic test. However with very high-risk first trimester results, the delays waiting for amniocentesis may not justify the anxiety.