



Antenatal screening

What is it?

A fetal anomaly, sometimes called a birth defect, is when there is a change to part of the body of the foetus. Such a change may increase the chance of stillbirth. It may also cause problems with your child's health, development and quality of life.

Many foetal anomalies can be tested or 'screened' for in early pregnancy. Tests can show if there is an anomaly. Common anomalies include risk of trisomy 21 (Down syndrome), trisomy 18 or a heart defect.

Antenatal screening is optional and there is often a cost involved. You don't have to have it. You can choose based on your personal preferences and risk factors.

These tests do not diagnose these conditions. They only show if there is an increased risk of an anomaly. If your test results show that there may be a risk of an anomaly, you may need more tests.

What will my GP do now?

Your GP will discuss the options, costs and risks of each screening test with you. Your GP may discuss the reasons why you might have the tests. You will also get resources to help you make a decision.

If you have a family history of genetic conditions like Down syndrome or heart defects, your GP may refer you to a specialist.

If you choose to have antenatal screening done, your GP will discuss the results with you. If you have an increased chance of your baby having an anomaly, you may consider whether you want to have more tests.

Some examples of further testing include a more accurate screening test for some chromosome conditions or a diagnostic test (Chorionic villus sampling (CVS) or Amniocentesis).

Your GP will give you more information about these tests. They may refer you to a clinic to have these tests done. Your GP may also discuss your options if your baby has a foetal anomaly.

What can I do?

Tell your GP if you know about any foetal anomalies in your family history, such as foetal heart defects or spina bifida. Read through the information provided by your GP.

If you are worried or have questions, speak to your GP or antenatal clinic. Waiting for test results can be difficult, but try not to feel anxious. Remember, screening tests do not diagnose anomalies. They can only tell you if there is a low or high risk or an anomaly.

Screening test options

The most common antenatal screening tests are:

Nuchal Translucency
A combined blood test and ultrasound

Non-Invasive Prenatal Test (NIPT)
A blood test that picks up the baby's DNA in your bloodstream and looks for certain genetic birth defects

If you choose this test, it is important to still have an ultrasound to see how your baby is developing

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What supports are available?

The **Liverpool Hospital Fetal Maternal Unit** provides ultrasounds for women with possible pregnancy-related complications such as fetal abnormality and multiple pregnancy. They also provide invasive prenatal diagnostic testing and counselling if a fetal anomaly is detected and you are unsure how to proceed.



What questions could I ask my doctor?

How much will each test cost?

What are the risks of having these tests?

What if there is an anomaly?

How soon do I get results?

How accurate are the tests?

Is there a deadline for the tests?



Where can I learn more?

If you'd like to learn more about foetal anomalies, the following resources can help:

- **NSW Health** – prenatal testing: genetics.edu.au
- **healthdirect** – prenatal screening and testing: healthdirect.gov.au

This information is to be viewed by someone who has received a diagnosis from their doctor. It is not designed to be used to diagnose a condition or as a substitute for ongoing medical care

Health Resource Directory factsheets are endorsed by South Western Sydney PHN's Community Advisory Committee and local GPs

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