Combined first trimester screening

This screening test involves an ultrasound at 11 to 13 weeks and a blood test between 10 and 13 weeks. The ultrasound measurement of the back of the baby’s neck (nuchal translucency) is combined with the results of the blood test and your age to estimate the chance of the baby having Down syndrome. It can identify approximately 85–90% of babies with Down syndrome, and may detect an increased risk of a range of other less-common chromosomal conditions. The ultrasound can also detect major structural conditions in the fetus.

Cell-free DNA screening or noninvasive prenatal testing

Cell-free DNA screening, or noninvasive prenatal testing (NIPT), uses a sample of your blood to estimate the chance of your baby having a chromosomal condition such as Down syndrome. It can identify about 99% of babies with Down syndrome, and can also be offered for other chromosomal conditions. An 11–13 week ultrasound is not included with this test and must be arranged separately if you decide to have one.
Who can I talk to?

Your doctor or midwife should be able to discuss the available screening and diagnostic tests with you. You may be referred to a genetic counsellor if you require additional support and information.

Women who have previously had a child with a chromosomal or genetic condition should receive individualised counselling from a specialist clinical genetics service, preferably before getting pregnant.

Second trimester serum screening

This blood test can be performed between 15 and 20 weeks of pregnancy. It can detect approximately 75% of pregnancies with Down syndrome.

Diagnostic tests

Only a diagnostic test, such as an amniocentesis or chorionic villus sampling (CVS), can definitely tell you whether your baby has a genetic or chromosomal condition. As diagnostic tests carry a small risk of miscarriage, most women have a screening test first before deciding if they wish to have diagnostic testing.

It is important to remember that even a normal amniocentesis or CVS result cannot guarantee a ‘perfect’ baby. There are many conditions that cannot be detected before a baby is born. More information about amniocentesis and CVS can be found on the RANZCOG website under Patient Information.

Useful resources

For further information on prenatal screening tests can be found at www.mcri.edu.au/prenatal-screening. It will be useful for you if you:

- Want to learn more about prenatal screening
- Are undecided whether or not to have screening or
- Are unsure about which test to have

For further online information about Down syndrome and other chromosomal conditions, go to the Raising Children Network: