The Royal Australian and New Zealand College of Obstetricians and Gynaecologists
Excellence in Women's Health

Prenatal Screening for Chromosomal and Genetic Conditions

Our genetic material, or DNA, is organised into 46 packages called chromosomes. Some people are born with too many or not enough chromosomes, which can affect their health and learning. Smaller changes in the DNA can also occur within a chromosome, resulting in a variety of genetic conditions.

Every baby has a small chance of being born with a chromosomal or genetic condition. Testing during pregnancy for some of these conditions is called prenatal screening. This is available to provide you with more information about your unborn baby. It is your choice whether to have any prenatal screening tests. You should only have testing if you understand what the test is for and what the results are able to tell you.

Screening tests

The principle of screening is to offer a safe, accessible test to identify women with an increased chance of having a baby affected by a chromosomal or genetic condition. These women with an increased chance are offered genetic counselling and follow up diagnostic testing (see below under 'Diagnostic tests'). It is very important to understand that a screening test does not tell you for certain if your baby has the condition, only if there is an increased chance. Screening tests may also miss some babies that have the condition. Your doctor or midwife should explain the results of your screening test to you and refer you to a genetic counsellor if required.

Which chromosomal conditions can be screened for?
The most common chromosomal cause of intellectual disability in children and adults is Down syndrome. This condition occurs when a baby has three copies of chromosome 21 (trisomy 21), instead of the usual two copies. The chance of a woman having a baby with Down syndrome increases with her age. Because of its frequency in the population (about 1 in 800) and its effects on health and learning, prenatal screening for Down Syndrome is made available to all Australian women. Other chromosomal conditions that may be screened for with the test are Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13). These are serious conditions associated with pregnancy loss, disability or death of the newborn.

Types of prenatal screening tests for Down syndrome and other chromosomal conditions

Name of test	What it involves
Combined first trimester screening	Ultrasound at 11–13 weeks plus a blood test at 10–13 weeks
Cell-free DNA screening or noninvasive prenatal testing (NIPT)	Blood test from 10 weeks
Second trimester serum screening	Blood test between 14–20 weeks

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 test for other chromosomal conditions. An 11–13 week ultrasound is not included with this test and has to be arranged separately if you decide to have one.

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.

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.

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
http://raisingchildren.net.au/articles/down_syndrome.html
http://raisingchildren.net.au/articles/trisomy_18_d.html
http://raisingchildren.net.au/articles/trisomy_13_d.html

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