Government of **Western Australia**
Department of **Health**

Prenatal screening for genetic conditions

Information about the tests available during pregnancy to check the health and development of your baby before birth

Contents

Introduction

This booklet describes the different tests you may be offered during your pregnancy to identify the risk of your baby having a genetic condition.

These include:

- genetic carrier screening tests for you and your partner
- screening tests for your baby
- diagnostic tests for your baby.

It is important to find out if there are any conditions which run in your family that may affect the health of your baby. It is best to do this before you get pregnant but genetic carrier screening is also offered in early pregnancy.

If you are concerned about a particular genetic condition in your family, please talk to your doctor or contact Genetic Services of Western Australia (see page 22).

Why have prenatal screening?

Prenatal screening provides you with important information about the health of your baby.

Knowing whether your baby has special needs could help you prepare for their arrival. You may also require specialist antenatal care and support during your pregnancy. You may need to be admitted to a tertiary hospital for the birth where immediate life-saving care for your newborn baby can be provided.

If you decide to end your pregnancy, you will be supplied with information about this and how you can seek additional support.

Whether you proceed with prenatal testing is a decision for you and your family, but it is important for you to be aware of the choices available. It can be helpful to discuss your options with a doctor, midwife or genetic counsellor.

It is important to note prenatal tests cannot detect all conditions.

What is the difference between the tests?

There are different types of tests available to you during pregnancy.

A **genetic carrier test** is a blood test which screens the woman or couple for their carrier status for common genetic conditions.

A **screening test** will show whether your pregnancy is at 'increased risk' of having a baby with a condition and should be offered to all women. Different screening tests are available in the first and second trimesters of pregnancy.

Prenatal screening tests will not harm your baby or increase your risk of miscarriage.

A **diagnostic test** will accurately identify the condition affecting your baby.

Some diagnostic tests are more invasive and may carry a very low chance of miscarriage.

What is genetic carrier screening?

Genetic carrier screening is a blood or saliva test to test the woman or couple for their carrier status for common genetic conditions.

Genetic carrier screening for an individual only needs to be done once in a lifetime and ideally before conception to identify if you and your partner are carriers of a genetic condition.

If you have a family history of a specific condition you may be eligible for additional testing.

If you are already in the first trimester of pregnancy, you and your partner may be tested at the same time for timely assessment. If both of you are carriers, the baby will have a high risk of having the condition.

Your doctor or genetic counsellor will explain your results to you and how this may affect a pregnancy.

A diagnostic test can confirm if the baby **definitely** has the condition.

What genetic conditions may be identified?

The conditions that may be identified differ depending on the screening test.

Genetic carrier screening may be offered for three common conditions **cystic fibrosis** (CF), **spinal muscular atrophy** (SMA) and **fragile X syndrome**.

Cystic fibrosis is a life limiting disease affecting the lungs and digestive systems. It is estimated that 1 in 25 Australians are genetic carriers (but clinically unaffected).

Information and support for families affected by CF can be found at Cystic Fibrosis Western Australia at www.cfwa.org.au

Spinal muscular atrophy is a group of inherited diseases that affects motor neurons that control muscle activity and progressively weaken with time. It is estimated that about 1 in 40 to 1 in 60 people are carriers of SMA.

Information and support for families affected by SMA can be found at Muscular Dystrophy WA at www.mdwa.org.au

Fragile X syndrome is a genetic disorder that results in a wide range of intellectual, physical and behavioural symptoms. It is estimated that one in 250 women and one in 800 men are Fragile X carriers.

Information and support for families affected by **fragile X syndrome** can be found at Fragile X Association of Australia at www.fragilex.org.au

There are many conditions that can be identified by genetic carrier screening if there is a family history or for certain populations.

Haemoglobin disorders such as thalassaemia and sickle cell disease are conditions that affect the ability of blood cells to function well and occur more frequently in certain populations. Screening may be offered to women of Mediterranean, Middle Eastern, African, Asian, Pacific Islander, South American and New Zealand Maori descent.

Information and support for families affected by **haemoglobin disorders** can be found at **Thalassaemia and Sickle Cell Australia** at info@tasca.org.au

What chromosomal conditions may be identified?

First trimester and second trimester screening tests can detect certain **chromosomal conditions**.

A chromosomal condition is caused by an alteration in the number or structure of chromosomes. There are 3 common trisomy conditions and several sex chromosome disorders that can be identified through screening.

Trisomy conditions arise from the affected baby having 3 copies of a chromosome instead of the usual 2. Screening currently tests for the following conditions:

- **Down syndrome** is caused by an extra copy of chromosome 21 which is why it is also known as trisomy 21.
- **Edwards syndrome** is known as trisomy 18 as babies born with the condition have 3 copies of chromosome 18.
- **Patau syndrome** is known as trisomy 13 as babies with this condition have 3 copies of chromosome 13.

Women of **any age** can have a baby with a chromosome condition however the risk increases with every year over the age of 35.

Information and support for families affected by **Down syndrome** can be found at www.downsyndrome.org.au

Information and support for families affected by **Edwards and Patau syndrome** can be found at the Support Organisation of Trisomy Syndrome **www.soft.org.au**

Sex chromosome conditions can be identified through some screening tests. These conditions arise due to variation in the number of X and Y chromosomes. Most people with these conditions lead normal lives with increased support and medical care. The most common **sex chromosome conditions** include:

- **Jacobs syndrome** affecting males with one extra Y chromosome.
- **Klinefelter syndrome** affecting males with extra X or Y chromosomes instead of the usual XY.
- **Trisomy (or triple) X syndrome** affecting females with 3 X chromosomes.
- **Turner syndrome** affecting females with only one X chromosome instead of the usual XX.

Information and support for families affected by **Klinefelter**, **Jacobs** and **Triple X syndrome** can be found at the Australian A and Y Spectrum Support **axys.org.au**

Information and support for families affected by **Turner syndrome** can be found at the Turner Syndrome Association of Australia **www.turnersyndrome.org.au**

When are the screening tests done?

The timing of the screening tests is different for each test.

The prenatal screening tests can be done during your:

- first trimester (10 – 14 weeks) or
- second trimester (14 – 20 weeks).

First trimester screening will enable you to find out early in your pregnancy of potential problems or concerns with your pregnancy.

Second trimester screening is valuable if you missed the first trimester screening test.

A first or second trimester screening test can help you decide whether you will have a diagnostic test. You only need to have the screening blood tests once each pregnancy.

What is first trimester screening?

A **first trimester screening test** will provide information on your risk of having a baby with one of the tested conditions.

The available screening tests include:

- First trimester blood test and
- Ultrasound (12–13 weeks).

Together, these 2 tests form the Combined First Trimester Screen.

or

- Non-invasive Prenatal Test (NIPT) and
- Ultrasound (13 weeks).

First trimester blood test

The first trimester blood test is taken between 9 weeks to 13 weeks and 6 days of pregnancy.

A sample of blood can be taken at any pathology collection centre. The blood is tested for the concentration of 2 hormones (free ßhCG and PAPPA) that change during pregnancy.

In a woman whose baby has a serious chromosome condition, the levels of these hormones will differ from normal levels.

Ultrasound

An ultrasound provides an image of your baby in the uterus. The ultrasound is performed between 12 weeks and 13 weeks of pregnancy.

The ultrasound allows a measurement to be taken of the thickness of fluid in an area behind the baby's neck. This area, known as the nuchal translucency is often larger in babies with health conditions.

The ultrasonographer will measure the crown to rump length to check for growth and gestational age. This measurement is from the head (crown) to the bottom of the buttocks (rump) of the fetus.

In addition to genetic conditions, the ultrasound can also detect some physical abnormalities.

Combined First Trimester Screen

A computer program uses the results of the first trimester blood test and ultrasound nuchal translucency measurement – together with the mother's age, weight and gestation of pregnancy – to provide risk assessments for chromosomal conditions.

This screening test is mostly covered by Medicare, the results should be available within 5 days and given to you by a doctor.

Non-invasive Prenatal Test (NIPT)

The NIPT is a blood test which can be done from 10 weeks of pregnancy.

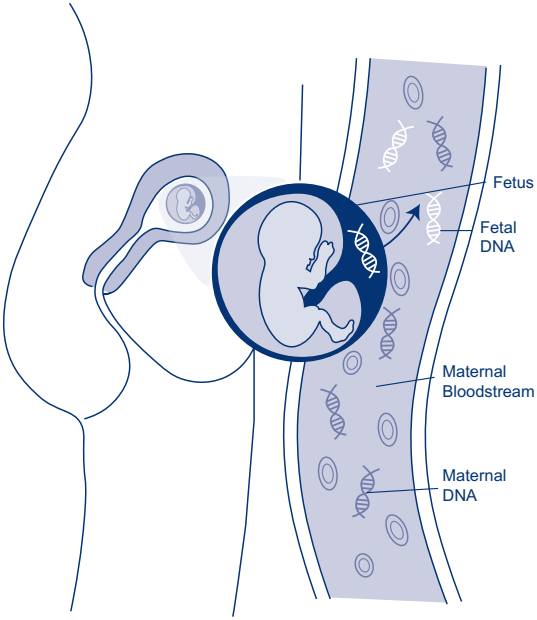
During pregnancy some of the DNA from the placenta crosses into the mother's bloodstream. This DNA carries the baby's genetic information, and this is what is analysed and measured in the mother's blood.

The NIPT is more accurate than other screening blood tests for identifying pregnancies at risk for chromosome conditions. It can also determine the sex of the baby and babies with sex chromosome abnormalities.

There are extra conditions that can be detected by NIPT, and the cost of the test may differ depending on the number of screened conditions. Discuss with your doctor or midwife if it is appropriate for you to test for the extra conditions. The accuracy of results for these extra conditions is lower than the common chromosomal conditions. The ability to accurately detect conditions is reduced in multiple pregnancies.

NIPT is **not** covered by Medicare so there will be an out of pocket cost for it.

Figure 1: Non-invasive prenatal testing of placental DNA



Ultrasound

The ultrasound is performed at 13 weeks of pregnancy (see page 11), **after** you have received your NIPT test results.

What is second trimester screening?

The available **second trimester screening tests** include:

- second trimester blood test (MSS)
- NIPT
- second trimester fetal anatomy scan.

Second trimester screening provides information on the risk of your baby having Down syndrome or a neural tube defect such as spina bifida.

Second Trimester Blood Test

A second trimester blood test is usually offered if the first trimester screen was missed. Testing can be done from 14 weeks to 18 weeks of pregnancy.

A sample of your blood is taken and tested in a laboratory for 3 hormones – estriol, free ßhCG and alpha fetoprotein. This test is sometimes called a maternal serum screen (MSS) or the triple test.

The results of the second trimester screening should be available within 5 days. Please discuss this with your doctor or midwife.

Non-Invasive Prenatal Test (NIPT)

NIPT is a blood test that can accurately identify pregnancies at risk for chromosomal conditions (see page 13).

This test can be done in either first or second trimester, but it is not needed in both.

Second Trimester Fetal Anatomy Scan

A 19–20 week ultrasound is recommended for **all** pregnancies to:

- check the position of the placenta
- check cervical length
- check the amount of amniotic fluid
- check the baby's growth
- screen for structural abnormalities in the fetus. This includes neural tube defects such as spina bifida, heart defects, kidney structure and limb abnormalities.

Ultrasound results may be available immediately or they may be sent to your doctor.

If a physical abnormality is found your doctor will explain what this means and, where appropriate, refer you to specialists.

What are the limitations of ultrasound?

The accuracy of the ultrasound depends on the equipment used, the mother's weight, the developmental stage of the baby and its position in the uterus, the visibility of the abnormality and other factors.

What do the screening results mean?

The results of the screening tests will be given to you by your doctor.

'**Not at increased risk**' means the risk of your baby having a condition that has been screened for is very low.

'**At increased risk**' means the risk of your baby of having a condition that has been screened for is higher than average.

Being told you are 'at increased risk' does not mean that your baby definitely has a condition.

If your pregnancy is 'at increased risk', a diagnostic test will be recommended and, with your consent, can confirm whether your baby has a condition.

Please discuss your results with your doctor, you may be eligible for genetic counselling.

What is a diagnostic test?

A diagnostic test is used to confirm if your baby has an abnormality.

You may decide to have a diagnostic test if you have:

- had a previous pregnancy with a genetic condition or abnormality
- been given an 'at increased risk' result from a first or second trimester screening test
- the ultrasound results show a possible abnormality
- a family history of a genetic condition.

What different diagnostic tests are available?

There are 2 invasive diagnostic tests for genetic conditions available to you during your pregnancy. These are:

- chorionic villus sampling (CVS)
- amniocentesis.

CVS and amniocentesis are both short outpatient procedures where you will be awake. Many women find the diagnostic tests uncomfortable, but you should experience little or no pain as they are often managed by local anaesthetic.

It is suggested that you rest for a short time after the procedure and take things easy for one to 2 days.

There may be costs for having the diagnostic tests, some of which are claimable through Medicare. Enquire about out of pocket costs when you book your appointments.

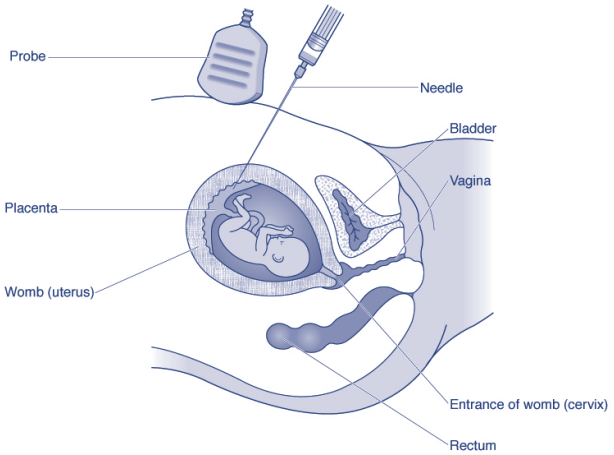
Chorionic villus sampling

This test is available from 12 to 14 weeks of pregnancy.

A needle is inserted through the abdomen into the uterus to take a sample of chorionic villus cells from the placenta. This procedure is done under a guided ultrasound to avoid harming the baby. Genetic testing is done on the sample which contains the placental DNA.

The risk of miscarriage from CVS is very low (less than 0.3%).

Figure 2: Chorionic villus sampling



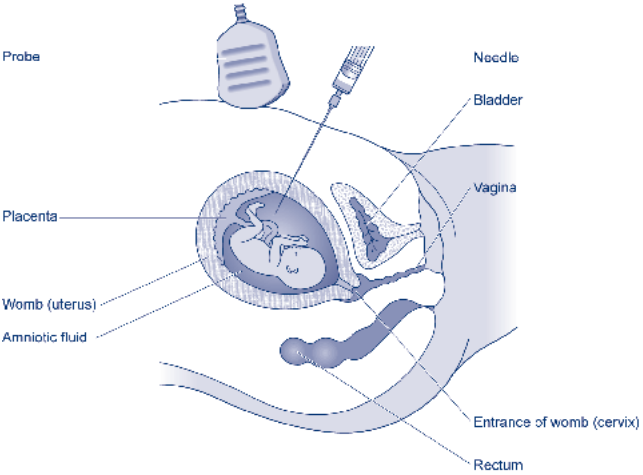
Amniocentesis

This test is usually done between 15 to 20 weeks of pregnancy.

A needle is inserted through the abdomen to take a small sample of amniotic fluid around the baby. This procedure is done under a guided ultrasound to avoid harming the baby. Genetic testing is done on the sample which contains the fetal DNA.

The risk of miscarriage from amniocentesis is very low (0.1 – 0.3%).

Figure 3: Amniocentesis



When should I receive the results of the diagnostic test?

The samples collected by CVS and amniocentesis are tested in a laboratory. While some results may be available within 24 to 48 hours, others may take up to 14 days, depending on the test.

Your doctor will explain your test results to you.

If testing confirms that your baby has a genetic condition, your doctor and/or genetic counsellor will discuss your options and support you. It will be up to you to make the decision that is right for you.

A normal result means your baby does not have the conditions tested for, but it does not rule out all genetic conditions or abnormalities.

Should I have these tests?

If you are still unsure about whether you should have prenatal screening tests you may like to consider the following questions:

- Do I want to know if my baby has a condition before they are born?
- What would I do if my diagnostic test showed my baby had one of these conditions? Would I want to know so I could prepare for a child with special needs? Would I end the pregnancy?
- How will this information affect my feelings – and my partner's feelings – throughout the pregnancy?
- Where can I get more information and support?

Talk to your doctor before you decide which of these tests are appropriate for you.

Where can I go for more information?

- **Your doctor or midwife**
- **Healthy WA website**
  www.healthywa.wa.gov.au
- **Maternal Fetal Medicine Service**
  King Edward Memorial Hospital
  374 Bagot Road, Subiaco WA 6008
  Phone: (08) 6458 2843
  www.kemh.health.wa.gov.au
- **Genetic Services of Western Australia**
  King Edward Memorial Hospital
  374 Bagot Road, Subiaco WA 6008
  Phone: (08) 6458 1242
  www.kemh.health.wa.gov.au

Support groups and associations

- **Cystic Fibrosis Western Australia**
  Phone: (08) 6457 7333
  www.cfwa.org.au
- **Down Syndrome Western Australia**
  Phone: (08) 6253 4752
  Toll free: 1800 881 935
  www.downsyndrome.org.au
- **Fragile X Association of Australia**
  Phone: 1300 394 636
  www.fragilex.org.au
- **Muscular Dystrophy WA** (includes SMA)
  Phone: (08) 9380 3400
  www.mdwa.org.au

- **Support Organisation of Trisomy Syndrome**
  Phone: 0407 820 560
  www.soft.org.au
- **Thalassaemia and Sickle Cell Australia**
  Phone: (03) 7015 5637
  info@tasca.org.au
- **The Spina Bifida and Hydrocephalus Association of WA (Inc)**
  Phone: (08) 9346 7520
  www.sbhawa.com.au

To improve the accuracy of the screening program, results and outcomes of pregnancies will be monitored.

Your privacy will be respected, and your personal details will remain confidential.

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