



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

Prenatal screening and diagnostic tests



Delivering a **Healthy WA**

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Introduction

This booklet describes the different tests you may be offered during pregnancy:

- tests for infections in the mother
- prenatal screening tests of the baby
- diagnostic tests of the baby.

A diagram at the back of the pamphlet outlines all of the tests and when they will be offered during your pregnancy.

First trimester routine tests in the mother

During your first trimester of pregnancy you may be offered tests¹ for:

- full blood count
- glucose challenges for diabetes
- ultrasound to check for dates, number of fetuses and development
- blood group and antibodies
- midstream urine
- random blood glucose
- syphilis
- rubella
- hepatitis B
- hepatitis C
- HIV antibodies
- chlamydia screening
- sickle cell and Thalassaemia (Haemoglobinopathy) screening for at risk women (Ethnic groups at high risk – Mediterranean, Middle Eastern, African, Asian, Pacific Islander, South American, New Zealand Maori)

¹ King Edward Memorial Hospital. Antenatal Shared Care

A number of these tests will be performed on one sample of your blood. It is best to have the tests as early as possible in your pregnancy.

Your doctor will explain the meaning of your test results.

Testing for health conditions in the baby

Prenatal tests are also available to check the health of your baby.

What conditions can be found?

- Chromosome conditions such as Down syndrome, Trisomy 13 and Trisomy 18.
- Neural tube defects such as spina bifida and anencephaly.
- Some birth defects such as congenital heart conditions and malformed kidneys.

About 5% of babies born in Western Australia have a birth defect. Prenatal tests cannot detect all possible conditions.

Down syndrome

Down syndrome is a condition that results in a range of physical and intellectual disabilities. It is caused by an extra copy of chromosome 21. Down syndrome is also known as Trisomy 21.

Down syndrome occurs in about one in 380 pregnancies. Women of **any age** can have a baby with Down syndrome; however this risk rises for every year over 35 years of age.

Trisomy 13 and Trisomy 18

Trisomy 18 is also a chromosome condition associated with intellectual disability and physical abnormalities in many parts of the body. It is known as Trisomy 18 because most babies born with this condition have three copies of chromosome 18 instead of the usual two copies. Trisomy 18 occurs in about one in 2,000 pregnancies.

Trisomy 13 is associated with intellectual disability and physical abnormalities in many parts of the body. It is known as Trisomy 13 because babies born with this condition have three copies of chromosome 13 instead of the usual two copies. Trisomy 13 occurs in about one in 4,800 pregnancies.

Babies with either Trisomy 13 or Trisomy 18 usually miscarry and if liveborn, rarely live longer than the first month.

Please ask your doctor if you have any questions about Down syndrome, Trisomy 13 or Trisomy 18.

Neural tube defect

A baby's brain and spine develop from the neural tube in the first four weeks of pregnancy. A neural tube defect occurs when the tube does not fully develop.

Spina bifida occurs when the tube does not completely close along the spine. Other neural tube defects include **anencephaly** and **encephalocele** in which the brain and skull do not develop properly.

About one in every 700 pregnancies is affected by a neural tube defect. This defect often causes the baby to be stillborn or die shortly after birth.

How to reduce the risk of neural tube defects

The vitamin folic acid (folate) is important for the healthy development of a baby. Most neural tube defects can be prevented by taking a supplement containing 0.5 mg of folic acid every day for at least one month before pregnancy and for the first three months of pregnancy.

A folate rich diet is also advised. Bread is now fortified with folic acid, but you should still take folate supplements to prevent neural tube defects.

Some women may also need to take a higher dose of folic acid if they have spina bifida or epilepsy, a previous pregnancy affected by a neural tube defect, or a family history of a neural tube defect.

Please ask your doctor or pharmacist for more information on folic acid during pregnancy.

Why would you have a prenatal test?

It is important you are aware of the choices available. Having a prenatal test is a decision for you and your family. It can be helpful to discuss these choices with your doctor or genetic counsellor.

Even if you would not terminate a pregnancy, knowing whether your baby has special needs could help you to prepare. You may require specialist antenatal care or to book into a tertiary hospital for the birth.

What are the prenatal tests?

Different types of tests are available during pregnancy. A **screening test** shows if a pregnancy is at 'increased risk' of a birth defect. Different screening tests are available in the first or the second trimester of pregnancy. These results indicate the risk of the baby having Down syndrome.

A screening test does not give a definite answer, but it does tell us which babies have an increased risk of having Down syndrome. The results may then help you decide if you want to have a diagnostic test.

A **diagnostic test** can identify a condition, and is very accurate. Diagnostic **invasive** tests (e.g. Chorionic Villus Sampling and Amniocentesis) however, increase the risk of miscarriage. This is why diagnostic tests are not routinely offered to all women. Instead, tests are offered in two stages. All women should be offered a screening test which carries no risk of miscarriage or harm to the baby. These tests identify most babies that have an increased risk of having Down syndrome. Diagnostic tests are then offered to women at increased risk.

Ultrasounds are also diagnostic tests but they are not invasive. This means there is no risk to you or your baby.

You may be offered an ultrasound to diagnose conditions, such as cardiac problems in your baby.

It is your choice to decide whether or not to have the screening and/or diagnostic tests. If you decide to have a screening test and you are later offered a diagnostic test, it is your choice whether or not to have the diagnostic test.

What are the screening tests?

There are two different screening options:

- First trimester screening – 9 weeks to 13 weeks six days of pregnancy.
- Second trimester screening – 14 to 18 weeks of pregnancy.

Many women choose to have a first trimester screening test to find out early in their pregnancy if there could be a problem. Second trimester screening is valuable for women who are too late for the first trimester screening test or if the first trimester screening test is not available where you live. A first or second trimester screening test can help you decide if you want a diagnostic test. Having both a first and second trimester screening test is not recommended.

What is first trimester screening?

9 weeks to 13 weeks 6 days

A first trimester screening test combines results from a blood test and an ultrasound to provide information on the risk of having a baby with Down syndrome. This test can

detect some other abnormalities. It may also tell you if you have a multiple pregnancy (e.g. twins). The screen does not usually detect spina bifida.

When is it done?

The first trimester test involves two measurements:

- Blood can be taken for testing between 9 weeks to 13 weeks 6 days of pregnancy, ideally 9-12 weeks.
- The ultrasound is done between 11 weeks to 13 weeks 6 days of pregnancy, ideally 12-13 weeks.

How is it done?

The blood test: The mother has a sample of blood taken at any Pathology collection centre. The blood is tested for the concentration of two hormones (free B-hCG and PAPP-A) that change during pregnancy. The amounts of these hormones are often changed when the baby has a serious chromosome problem.

The ultrasound: An ultrasound should be performed by an operator credentialled by the Fetal Medicine Foundation or the Nuchal Translucency Education and Monitoring Programme of The Royal Australian and New Zealand College of Obstetricians and Gynaecologists. 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 nuchal

translucency (NT) is often larger in babies with Down syndrome. The ultrasonographer will also take some measurements of the crown-rump length or biparietal diameter of the baby to calculate the gestational age.

A computer program then uses the results of the blood test and ultrasound NT measurement, together with the mother's exact age, weight and correct gestation of pregnancy, to provide risk assessments for Down syndrome and Trisomy 13 and Trisomy 18. The results of the risk assessment will be given to you by your doctor. Your doctor will also tell you if a physical abnormality has been found during the ultrasound examination.

Results should be available within five days.

What do the results mean?

Results are given as a risk or chance that the baby will be affected.

'Not at increased risk' means the risk of having a baby with Down syndrome is very low (the risk is less than one in 300).

A different birth defect could still be present but this risk is also low.

'At increased risk' means the risk of having a baby with Down syndrome is greater than 1 in 300 (the risk lies between 1 in 2 and 1 in 300). If a pregnancy is at increased risk, a diagnostic test will be recommended to confirm whether or not the baby has Down syndrome. Please discuss these results with your doctor.

What are the limitations?

One in every 25 women tested will be told they are at increased risk. This does not mean there is definitely something wrong but you might consider further diagnostic tests.

What are the costs for screening tests?

There may be a cost for the screening tests. You may be able to claim part of this cost from Medicare. Please ask when you book your appointment, for the costs involved.

What is an ultrasound?

An ultrasound provides an image of the baby in the womb. It can be done at any stage in your pregnancy.

- A gel is applied to your abdomen to allow sound waves to pass from the ultrasound probe into the uterus.
- The ultrasound probe is moved over your abdomen and an ultrasound image is produced by the reflection of the sound waves off the baby.
- Your bladder should be comfortably full to get a clear image.
- Sometimes, in the first trimester of pregnancy, a vaginal ultrasound is performed rather than an abdominal ultrasound.

The person performing the ultrasound will advise which procedure is appropriate for you.

Why would you have an ultrasound?

An ultrasound is an important part of first trimester screening. An ultrasound is also recommended at 18–20 weeks. (see page 18)

Some women may be offered an ultrasound at 6–8 weeks of pregnancy to confirm the baby's gestational age, show if you are carrying twins, and check the baby's growth.

An ultrasound may also be done if there are unusual symptoms, such as bleeding.

What are the risks?

An ultrasound is safe for you and the baby.

What is second trimester screening? 14 to 18 weeks

A second trimester screening test is a blood test of the mother that provides information on the risk of having a baby with Down syndrome or a neural tube defect like spina bifida. This test is sometimes called a maternal serum screen (MSS) or the triple test.

When is it done?

Blood for the second trimester screening test can be taken for testing between 14 weeks to 18 weeks of pregnancy, but is ideally done between 15 to 17 weeks.

How is it done?

A blood sample is taken from the mother and tested in a laboratory. The mother's blood is tested for three hormones (estriol, free B-hCG and alpha fetoprotein).

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

Results should be available within five days.

What do the results means?

‘Not at increased risk’ means... The chance of the baby having Down syndrome or a neural tube defect is very low (less than one in 300). A different birth defect may still be present but this risk is also low.

‘At increased risk’ means... the chance of the baby having Down syndrome or another chromosome abnormality is greater than 1 in 300 (between 1 in 10 and 1 in 300). Alternatively, the risk of the baby having a neural tube defect lies between one in 12, and 1 in 128. A diagnostic test will be recommended to confirm whether or not the baby has Down syndrome (chorionic villus sampling or amniocentesis) or spina bifida (ultrasound). Please discuss these results with your doctor.

What are the limitations?

One in every twenty women tested will be told they are at increased risk. This does not mean there is definitely something wrong but you might consider having further diagnostic tests.

What are the costs?

There may be costs for having the screening tests. You may be able to claim part of this cost from Medicare. Please ask when you book your appointment, for the costs and any rebates available for the screening tests.

What is a diagnostic test?

A diagnostic test is used to confirm a chromosome abnormality such as Down syndrome or an inherited condition in the baby.

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

- had a previous pregnancy with Down syndrome or other birth defect.
- been given an 'at increased risk' result from a first or second trimester screening test.
- a family history of a genetic condition.

What are the different diagnostic tests?

The types of diagnostic tests are Chorionic Villus Sampling, Amniocentesis and ultrasound.

Chorionic Villus Sampling (CVS) 11 to 14 weeks of pregnancy

A needle, guided by ultrasound to avoid damage to the fetus, is inserted through the abdomen to take a sample of chorionic villus cells from the placenta. This sample is tested for missing, extra or abnormal chromosomes.

CVS is an outpatient procedure that takes about 20 minutes. You will be awake during the procedure but should experience little or no pain. It is suggested you rest for about 20 minutes after the procedure.

The risk of miscarriage is less than 1 in 100 (less than 1%).

Figure 1: Chorionic Villus Sampling - through the abdomen

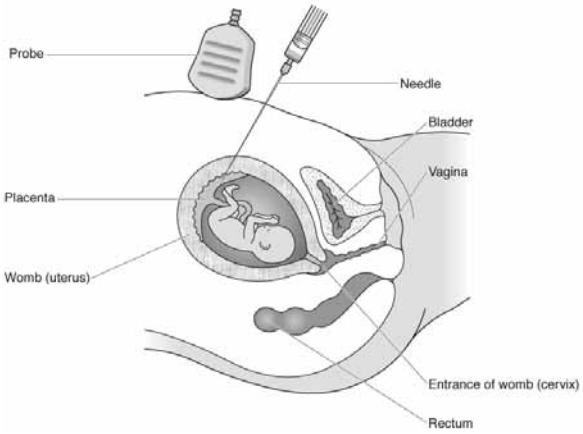
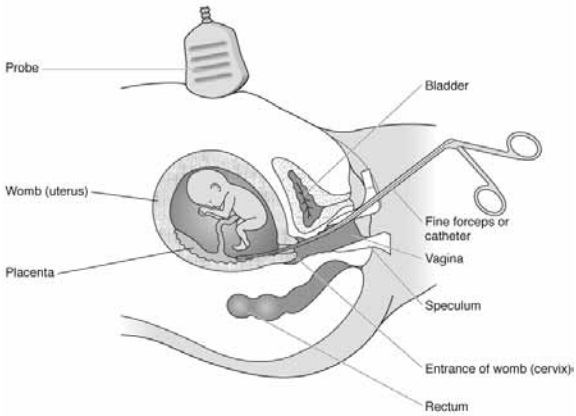


Figure 2: Chorionic Villus Sampling - through the cervix (entrance to the womb)



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Amniocentesis

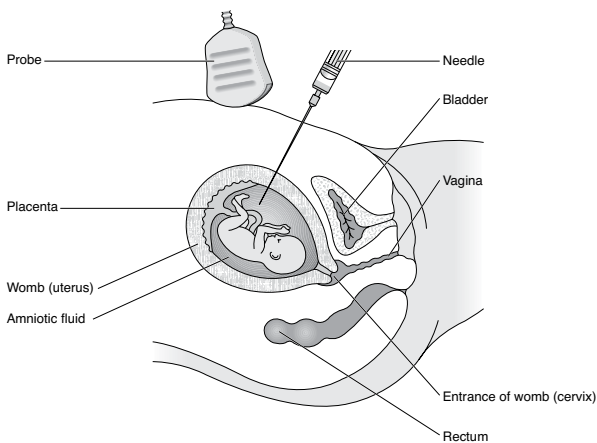
15 to 18 weeks of pregnancy

A needle, guided by ultrasound to avoid damage to the fetus, is inserted through the abdomen to take a small sample of amniotic fluid around the baby. This sample is tested for missing, extra or abnormal chromosomes.

Amniocentesis is an outpatient procedure that takes about 20 minutes. You will be awake during the procedure but should experience little or no pain. It is suggested you rest for about 20 minutes after the procedure.

The risk of miscarriage is less than 1 in 100 (less than 1%).

Figure 3: Amniocentesis



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Are the tests painful?

Many women find the diagnostic tests uncomfortable, and they are often managed by local anaesthetic. You should take things easy for one to two days after the tests.

When should I receive the results?

The samples collected by chorionic villus sampling or amniocentesis are tested in a laboratory. Depending on the test, results may be available within 24 hours, but it may take up to 14 days.

Your doctor will explain the test result and any implications. If a condition is found, counselling with Genetic Services of Western Australia may be recommended.

If the testing confirms your baby has Down syndrome, Trisomy 13 or Trisomy 18, your doctor and/or genetic counsellor will discuss your choices with you, but allow you to make a decision that is right for you. Your choices include ending the pregnancy, continuing the pregnancy, or placing the baby for adoption.

What are the limitations?

These diagnostic tests will detect practically all chromosomal abnormalities associated with Down syndrome, Trisomy 13 and Trisomy 18.

A normal result means the baby does not have Down syndrome or other common chromosomal conditions but does not rule out all birth defects.

What are the costs?

There may be costs for having the diagnostic tests. You may be able to claim part of this cost from Medicare. Please ask when you book your appointment, for the costs and any rebates available for the diagnostic tests.

What is a routine structural ultrasound? 18-20 weeks

An 18 to 20 week ultrasound is recommended to:

- Check the position of the placenta.
- Check the amount of amniotic fluid.
- Check the baby's growth.
- To detect structural abnormalities in the fetus – such as heart, limbs, abdomen, bones, brain, spine and kidneys.

How is it done?

See 'What is an ultrasound' on page 11 for this information.

When should I receive the results

Ultrasound results may be available immediately or may be sent to your doctor. If a physical abnormality is found your doctor will explain what this means and refer you to specialists where appropriate.

What are the limitations?

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 is the cost?

There may be a charge for having a routine structural ultrasound. You may be able to claim part of this cost from Medicare. Please ask when you book your appointment, for the costs and any rebates available for the ultrasound.

Frequently asked questions

What does prenatal mean?

Prenatal refers to any time during pregnancy.

Prenatal tests check the health of the baby during pregnancy. Some conditions cannot be detected with a prenatal test. Please discuss this with your doctor.

What does antenatal mean?

Antenatal also refers to any time during the pregnancy.

What does maternal mean?

Maternal is another word for mother. A maternal serum screen means a test of the mother's blood.

How do I know if I have a family history of a condition?

It is important to find out if there are any conditions that run in your family that may affect the health of the baby. You may have to ask your family members if they know of any conditions. It is best to do this before you get pregnant.

If you are concerned about a particular genetic disorder in your family please talk to your doctor or contact Genetic Services of Western Australia.

How is my due date estimated (EDD)?

An ultrasound can be used to estimate your due date. You can also estimate it based on your last menstrual period. Your due date is calculated by adding 40 weeks (280 days) to the first day of your last menstrual cycle.

Who should I ask for more information about these tests?

You may be unsure whether or not to have prenatal screening. Some questions which you may consider to help you decide include:

- Do I want to know if my baby has Down syndrome, Trisomy 13, Trisomy 18 or a neural tube defect before he/she is born?
- What would I do if my diagnostic test showed my baby has one of these conditions? Would I end the pregnancy? Would I want to know so I could prepare for a child with special needs?
- How will this information affect my feelings and the father of the baby's feelings throughout the pregnancy?

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

For more information, please contact:

Your doctor

or

Genetic Services of Western Australia

King Edward Memorial Hospital

374 Bagot Road, SUBIACO WA 6008

Phone: (08) 9340 1525

Fetal Medicine Service

King Edward Memorial Hospital

374 Bagot Road, SUBIACO WA 6008

Phone: (08) 9340 2700

**The Down Syndrome Association
of WA (Inc)**

Phone: (08) 9368 4002

Web: www.dsawa.asn.au

The Spina Bifida Association of WA (Inc)

Phone: (08) 9346 7520

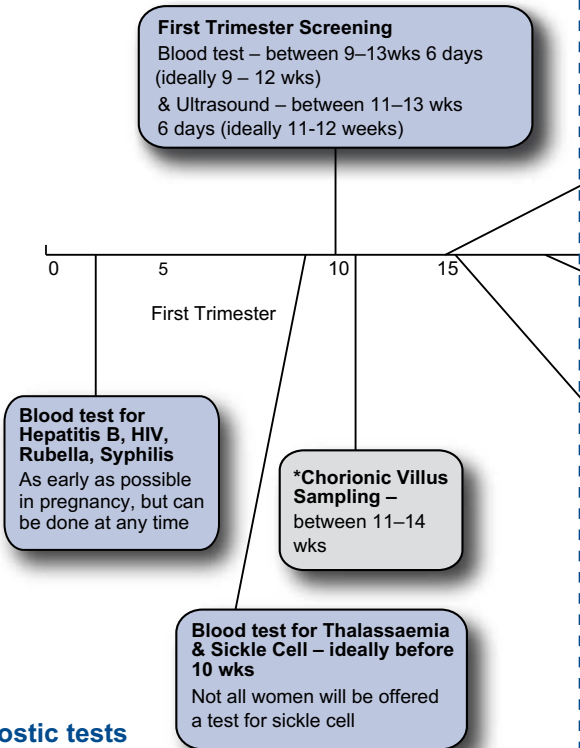
Web: www.sbawa.asn.au

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.

Screening and diagnostic test options

Screening tests

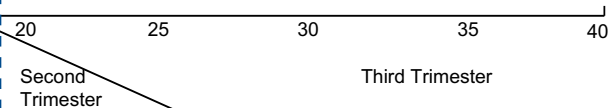


Diagnostic tests

Second Trimester Screening

Blood test (Maternal Serum Screen) –
between 14-18 wks
(ideally 15–17 wks)

You may choose whether or not to have the screening
and/or diagnostic tests.



***Amniocentesis –**
between 15–18 wks

**Routine structural
ultrasound – 18–20 wks**

*In consultation with health professionals, you may choose whether or not to have diagnostic testing. Diagnostic testing involves either chorionic villus sampling or amniocentesis.

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