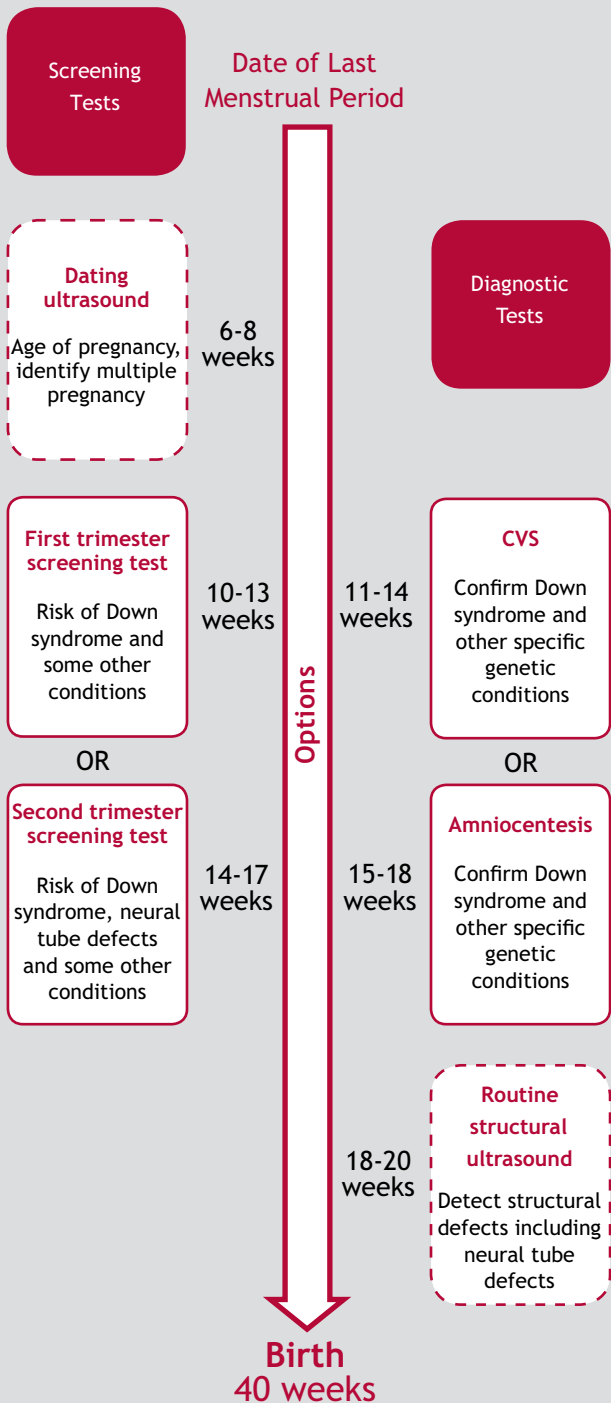




# Prenatal screening and diagnostic tests





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## Introduction

Tests are available during pregnancy to check the health of a baby.

### What conditions can be found?

- Down syndrome.
- Neural tube defects such as spina bifida and anencephaly.
- Birth defects such as congenital heart conditions, malformed kidneys, missing limbs, trisomy 13 and trisomy 18.

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 in every cell (trisomy 21). Down syndrome occurs in about one in 400 pregnancies. Women of **any age** can have a Down syndrome pregnancy, however this risk is higher for women over 35 years (see FAQ section). Other types of chromosome conditions are trisomy 13 and trisomy 18.

### Neural tube defect

A baby's brain and spine develop from a 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.

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 0.4mg 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. Some women may 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 have a prenatal test?

It is important to be 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 interrupt a pregnancy, knowing whether the baby has special needs could help you to prepare. A specialist may need to be present at the birth. Some structural abnormalities can even be treated before the baby is born.

### Types of 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 tests tell us if Down syndrome is **likely or unlikely** to be present. A screening test cannot definitely tell if the baby has Down syndrome but the result may help you decide if you want to have a diagnostic test.

A **diagnostic test** can identify a condition, and is very accurate.

## Screening tests

There are two different screening options:

- First trimester screening - 10 weeks to 13 weeks 6 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. 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.

### First Trimester Screening 10 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). This first trimester screen does not detect neural tube defects.

#### When is it done?

The first trimester test involves two measurements. Blood can be taken for testing between 10 weeks and 13 weeks 6 days of pregnancy, but is ideally done between 10 and 12 weeks. The ultrasound is done between 11 weeks and 13 weeks 6 days of pregnancy.

#### How is it done?

The mother has a sample of blood taken and an ultrasound performed by an operator accredited by the Fetal Medicine Foundation or the Nuchal Translucency Education and Monitoring Program.

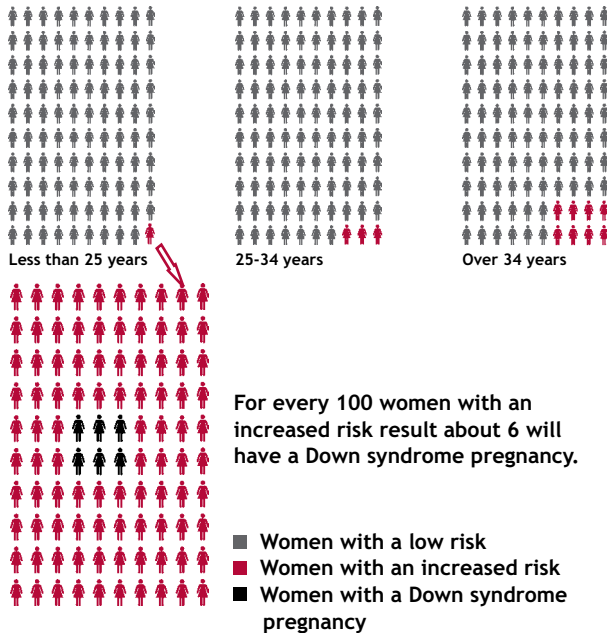
**The blood test:** The mother's blood is tested in a laboratory for the level of two hormones (free  $\beta$ -hCG and PAPP-A) that change during pregnancy.

**The ultrasound:** 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. (See page 9 for information about ultrasounds).



The information about the two hormones in the mother's blood is combined with the information from the ultrasound to provide a risk assessment. Your doctor will also tell you if a physical abnormality has been found during the ultrasound examination.

The charts show the proportion of women at increased risk of having a Down syndrome pregnancy, identified through first trimester screening. Women at increased risk have a screening result between 1 in 2 and 1 in 300.



### Your results

**‘Not at increased risk’** means...

The chance of having a baby with Down syndrome is very low. A different birth defect could still be present but this risk is also low.

**‘At increased risk’** means...

The chance of having a baby with Down syndrome is between 1 in 2 and 1 in 300. 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 twenty 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 (see page 11).

## 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 wand into the uterus.
- The ultrasound wand is moved over your abdomen and an ultrasound image is produced by the reflection of the sound waves off the baby.
- The 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. Your doctor will advise which procedure is appropriate for you.

### Why would you have this test?

Ultrasound is an important part of first trimester screening. An ultrasound is also recommended at 18-20 weeks.

Some women may be offered an ultrasound at 6-8 weeks of pregnancy to confirm the baby’s gestational age and due date, show if you are carrying twins, show the position of the placenta and baby and check the baby’s growth.

Ultrasound may also be used to investigate unusual symptoms. These include bleeding, fluid retention and lack of movement by the baby.

### What are the risks?

An ultrasound is safe for you and the baby.

## Second Trimester Screening

### 14 to 18 weeks

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

#### When is it done?

Blood for the second trimester screening test can be taken for testing between 14 weeks and 17 weeks 6 days of pregnancy, but is ideally done between 15 and 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  $\beta$ -hCG and alpha fetoprotein).

#### Your results

**'Not at increased risk'** means...

The chance of the baby having Down syndrome or a neural tube defect is very low. 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 a chromosome abnormality is between 1 in 10 and 1 in 300. The risk of the baby having a neural tube defect is between 1 in 12 and 1 in 128. A diagnostic test will be recommended to confirm whether or not the baby has Down syndrome or spina bifida. 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.

### Diagnostic tests

A diagnostic test is used to confirm a chromosome abnormality such as Down syndrome or an inherited condition in the baby. There are two different diagnostic test procedures:

- **Chorionic Villus Sampling (CVS)** - 11 to 14 weeks of pregnancy  
A small sample of **placenta** is taken for testing in a laboratory.
- **Amniocentesis** - 15 to 18 weeks of pregnancy  
A small sample of **amniotic fluid** is taken for testing in a laboratory.

You may choose to have a diagnostic test if:

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

These diagnostic tests have a small risk of miscarriage, which occurs in less than 1 in 200 procedures.

### How are they done?

A needle is inserted through the abdomen (transabdominal) into the uterus with the guidance of an ultrasound. A small sample of fluid or tissue is removed for testing in a laboratory. This sample is tested for missing, extra or altered chromosomes.

CVS may also be done through the cervix (transcervical). A fine cannula (tube) is inserted through the vagina and into the placenta with the guidance of an ultrasound.

### Your results

Results should be available within 12-14 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.

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

**These tests are 99% accurate.**

## 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.
- Detect spina bifida and other neural tube defects.
- Detect malformed kidneys.
- Detect some congenital heart defects and short or missing limbs.

### How is it done?

See 'Ultrasound' on page 9 for this information.

### Your 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.

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.

## 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 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 determined 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?

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 (or 9340 2705)

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

Phone: (08) 9358 3544  
[www.dsawa.asn.au](http://www.dsawa.asn.au)

#### The Spina Bifida Association of WA (Inc)

Phone: (08) 9346 7520  
[www.sbawa.asn.au](http://www.sbawa.asn.au)

#### Genetic Support Council WA

Phone: (08) 9389 6722  
[www.geneticsupportcouncil.org.au](http://www.geneticsupportcouncil.org.au)