

3. You or your doctor considers your age is a risk.
4. You have a screening test that shows a high chance of your baby having a problem.

There are two diagnostic tests. In both tests a needle is put into your uterus. Ultrasound is used to guide the needle. These tests involve a small risk of complications such as miscarriage.

### 1. Chorionic Villus Sampling (CVS)

A CVS is performed between 11 - 14 weeks of pregnancy. The needle is guided into the placenta and a small sample is taken for testing.

### 2. Amniocentesis

An amniocentesis is performed after 15 weeks of pregnancy. The needle is guided into the sac around the baby and fluid is taken for testing.

### Results

The test looks at all your baby's chromosomes and the result is usually ready in 2 - 3 weeks.

Although diagnostic tests cannot rule out all chromosomal or genetic problems or syndromes, a normal result is reassuring.

**Your GP or specialist will talk to you about your screening or diagnostic test results and help you to understand your choices and options, or refer you for more information.**

### KEY POINTS TO REMEMBER

- Every woman should think about having the first trimester combined test – this is the best screening test available for Down syndrome. Other valuable information can also be gained from this test.
- If you cannot get the first trimester combined test, you should ask about the second trimester Triple test.
- Screening and diagnostic tests cannot find all problems – every test has its limits.
- Speak to your GP, obstetrician, midwife, Genetic Health Queensland or phone 13HEALTH (13432584) for more information.



**Queensland**  
Government

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# Screening for Down Syndrome in Pregnancy



### Did you know that .....

Most pregnant women can expect to have a healthy pregnancy and baby.

However, not every pregnancy is trouble free.



**Queensland** Government

## **Congenital Abnormalities**

**One in twenty-five babies are born with a congenital problem.**

A lot of congenital problems can be identified by ultrasound or screening tests, but some cannot. Things like blindness, deafness, cerebral palsy and autism cannot be found before birth.

Some problems like spina bifida may be diagnosed by an ultrasound at 18 to 20 weeks of pregnancy (the Second Trimester Ultrasound).

Down syndrome cannot be diagnosed by ultrasound, but there are screening tests available.

## **Screening Tests**

**A screening test tells you if you have a high or low chance (risk) of having a baby with certain conditions like Down syndrome in this pregnancy.**

**It will not tell you if the baby definitely has a problem. Screening tests may help you decide if you would like to have further testing (a diagnostic test).**

There are two screening tests available for Down syndrome in pregnancy. The 18 to 20 week ultrasound is a screening test for problems like spina bifida.

## **1. Down Syndrome Screening**

### **a. First Trimester Combined Test:**

**Nuchal Translucency Ultrasound (Scan) and First Trimester Blood Test.**

**This is the best test available to screen for Down syndrome.**

The test is performed between 11 to 13 weeks and 6 days of pregnancy and needs a special blood test combined with the nuchal translucency scan.

The blood test should be done about 3 days before your scan. If you have your blood test on the day of your scan your test results will not be ready for a few days.

Your blood test, scan and age are combined to calculate the risk of your baby having Down syndrome.

Even if you don't want to know this information, a nuchal translucency scan can give you other important information about your pregnancy such as whether you are having twins.

### **b. Second Trimester Blood Test / Triple Test**

This is a specific blood test that can be done between 15 and 20 weeks of pregnancy. This test can be helpful if you were not able to have the first trimester test.

## **2. Second Trimester Ultrasound/ Morphology Scan**

This is an ultrasound usually done between 18 and 20 weeks of pregnancy. During the scan your baby is carefully checked to see how it is growing.

This scan is a good screening test for congenital problems like spina bifida, but is not very good at screening for Down Syndrome.

## **What if a Problem is Found?**

If your screening test is high risk it means the *chance* of your baby having Down syndrome is increased. To find out if your baby has Down syndrome you would need to have a diagnostic test.

## **Diagnostic Tests**

Diagnostic tests (also known as "invasive" tests) diagnose chromosome problems like Down syndrome.

You may be offered a diagnostic test if

- 1.** A relative or family member has a history of chromosome or genetic problems.
- 2.** You have had a previous baby with Down syndrome or other chromosomal problems.