Antenatal screening and testing for Down syndrome and other conditions

– in pregnancy

Optional screening – your choice – your decision

All pregnant people are advised of screening to assess the chance of their baby having Down syndrome or another condition. These conditions may affect a baby's development. This screening is optional.

Antenatal screening for Down syndrome and other conditions

During your pregnancy, you will be advised of screening for a number of conditions that may affect you or your baby. This screening includes a blood test from you and measurements of your baby taken from an ultrasound scan, or a blood test from you only. Screening can be done up until you are 20 weeks pregnant.

All pregnant people are advised of screening to assess the chance of their baby having one of the screened conditions. These conditions can delay the baby's learning and physical development.

Depending on the screening results, you may be offered diagnostic tests to confirm if your baby has a condition. Some, but not all, conditions that may affect you and your baby can be detected during pregnancy.

Screening and testing are your decisions

Choosing whether to have this screening is an important decision. You need to have enough information to make the decision that is right for you. Some people will want to know if their baby has one of the conditions being screened for before the baby is born. Other people will decide not to have this screening.

Your midwife or doctor will discuss with you:

- the conditions that are screened for
- the differences between screening and diagnostic testing
- the risks and benefits of screening and testing
- what it may mean to have a child with one of the conditions being screened for
- where and when to get the blood test and scan, and any charges that may apply
- what the results may mean and the next steps.

You do not have to have screening and testing if you do not want to.

What is Down syndrome (also called trisomy 21)?

Down syndrome is a lifelong condition present from conception. People with Down syndrome have an extra copy of chromosome 21 that results in learning difficulties and may cause health problems.

People with Down syndrome have different abilities and personalities just like everyone else. They have some physical features in common, but they also resemble their parents and family.

People of any age and ethnicity can have a baby with Down syndrome. It usually happens by chance. Down syndrome occurs in approximately 1 in 1000 births.

Some health problems can be linked with Down syndrome. These problems may range from minor to severe and include:

- reduced muscle tone
- sight and hearing problems
- heart and bowel problems.

Many of these problems can be treated. Help and support is available for families who have a child with Down syndrome. People with Down syndrome attend school and take part in family/whānau and community life.

What other conditions may be indicated by this screening?

Down syndrome is the most common condition indicated by this screening. Other, more rare conditions include Patau syndrome (also called trisomy 13) and Edwards syndrome (also called trisomy 18). These conditions (caused by extra copies of chromosomes) can lead to life-threatening complications that severely affect the brain, heart, and kidneys. Most babies only live for a few months and need full-time care. As well as Patau and Edwards syndromes, other rare conditions may be identified by screening. Your midwife or doctor will be able to provide you with information about these conditions.

A scan when you are around 18 weeks pregnant can provide extra information about your baby. For example, this scan may be the best way to find out if your baby has spina bifida or a heart condition.

Screening

All pregnant people are advised about this screening. This screening indicates whether your baby has a low or increased chance of having Down syndrome or another condition. ***Screening alone cannot tell you for sure whether your baby has a condition.***

First trimester combined screening is offered if you are less than 14 weeks pregnant.

- This screening includes a blood test from you and measurements of your baby taken from an ultrasound scan. The blood test is free – you may be charged for the scan.
- The best time for the blood test is when you are 9–10 weeks pregnant, but it can be done up to 13 weeks and 6 days pregnant (see image 1).
- An ultrasound scan of your baby is ideally taken when you are 12 weeks pregnant, but it may be done between 11 weeks and 2 days and 13 weeks and 6 days (see image 2). The nuchal translucency or NT is a measurement of the fluid-filled space at the back of your baby's neck (see image 3).
- The laboratory combines the blood test and scan results with other information, such as your age and weight, to work out the chance of your baby having one of the screened conditions.

Second trimester maternal serum screening is offered if you are 14–20 weeks pregnant.

- This screening involves a blood test from you. This blood test is free.
- The best time to test your blood is when you are 14–18 weeks pregnant, but it can be done up until you are 20 weeks pregnant (see image 1).
- The laboratory combines this blood test with other information, such as your age and weight, to work out the chance of your baby having one of the screened conditions.

Image 1. The blood test.

Image 2. Having the ultrasound scan.

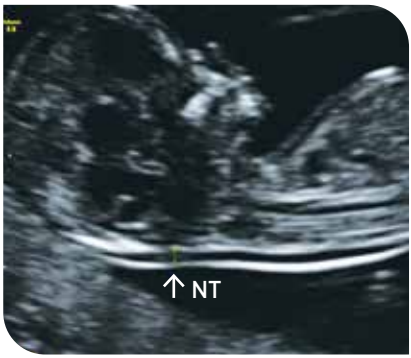


Image 3. Nuchal translucency or NT scan showing the fluid-filled space at the back of the baby's neck.

Getting your screening results

Your midwife or doctor will receive your results in 7–10 days and will discuss what this may mean for you.

- A **low chance** result means there is very little chance that your baby has one of the screened conditions. It does not completely rule out the possibility. A few people who have a low chance result may still have a baby with a screened condition, or another health condition.
- An **increased chance** result means there is a higher chance that your baby has one of the screened conditions. It does not mean your baby has the condition. Very few people who have an increased chance result will have a baby with a screened condition.

Receiving an increased chance result

If you receive an increased chance result, your midwife or doctor will discuss the results with you, answer your questions and offer a free appointment with a specialist.

You can accept or decline this referral.

The specialist will discuss what your results mean and your options, including diagnostic testing, which is the only way to confirm if your baby has one of the screened conditions.

Talking to a specialist can help you to decide whether to have diagnostic testing. Your decision can be made after this discussion.

Diagnostic testing

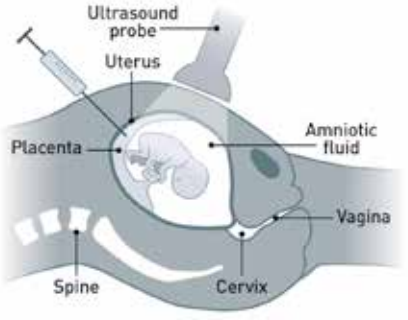
Diagnostic testing will tell you definitely whether your baby has Down syndrome or another screened condition. Most people who have further testing find out that their baby does not have a condition. A very small number of people will find out that their baby does have a condition.

Some people have further testing to find out if their baby has one of the screened conditions. This can help them to prepare for the birth and family life. Others may consider terminating the pregnancy (also called abortion). Adoption may also be considered.

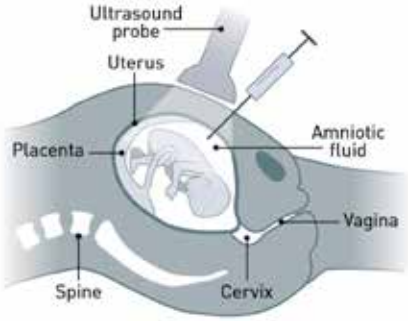
- Take time to talk to your midwife or doctor, your specialist and your family/whānau.
- Ask as many questions as you need to before deciding whether to have the diagnostic test.

Diagnostic tests look at a sample of your baby's cells. To collect the sample, a thin needle is inserted through the skin on your abdomen (belly) and into the placenta or the fluid around the baby. A scan is done at the same time to guide the position of the needle. The type of sample collected depends on how many weeks pregnant you are (see the diagrams).

Diagnostic testing is your choice. For every 200 people who have diagnostic tests, one or two will have a miscarriage. For this reason, some people choose not to have diagnostic testing.



Chorionic villus sampling – before 14 weeks, a sample of the developing placenta (whenua) is taken.



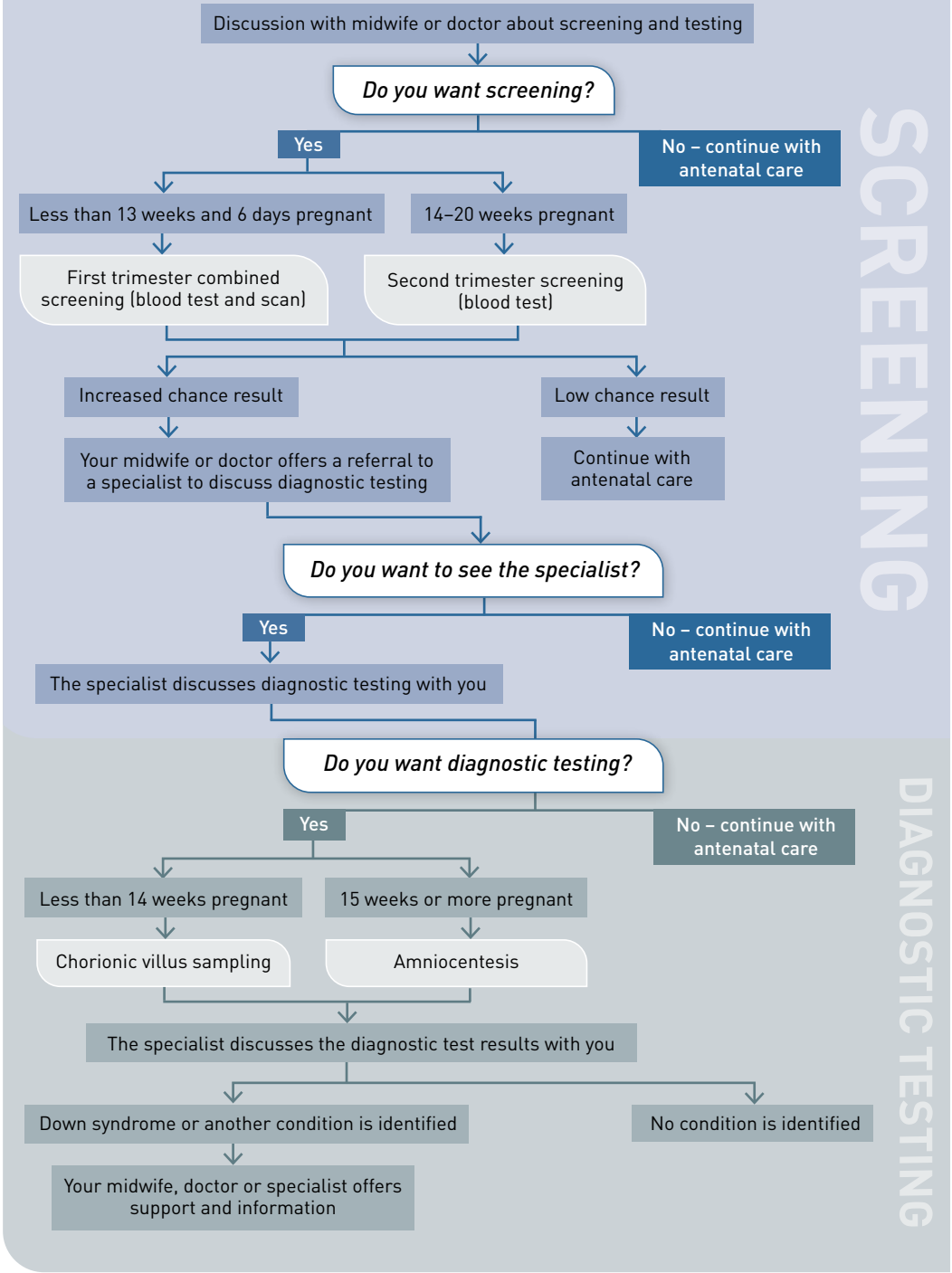
Amniocentesis – from 15 weeks, a sample of amniotic fluid (the waters around the baby) is taken.

Receiving the results of diagnostic testing

Your specialist will receive the test results in about two weeks. If your baby is found to have Down syndrome or another condition, your midwife, doctor or specialist will provide you with information and support.

You will be offered appointments with genetic services, a social worker or other services. You will be offered or given support to help you with any decisions that you may need to make. Diagnostic tests cannot tell you the range of abilities or challenges your baby may have.

The Screening and Testing Pathway



More information

It's important that you have enough information to help you decide about screening and testing for Down syndrome and other conditions. If you would like more information, ask your midwife, doctor or specialist. You can also visit the National Screening Unit's website www.nsu.govt.nz. Information on non-invasive prenatal testing (NIPT) is available from www.nsu.govt.nz and the New Zealand Maternal Fetal Medicine Network website https://www.healthpoint.co.nz/public/wahi-rua-new-zealand-maternal-fetal-medicine/.

For local support groups in your area contact:

- The New Zealand Down Syndrome Association 0800 NZDSAI (0800 693 724) www.nzdsa.org.nz
- The New Zealand Organisation for Rare Disorders NZ www.raredisorders.org.nz

Your rights

The code of Health and Disability Services Consumers' Rights protects your rights. For more information, see www.hdc.org.nz

The Health Information Privacy Code protects your personal details and gives you rights of access and correction. For more information, see www.privacy.org.nz

What information is collected and how is it used?

As part of screening, information about you is collected and stored by the laboratories. This includes your name, address, ethnicity, weight and other information that helps to give accurate results. This information is held securely and confidentially.

This information is used to:

- interpret screening results
- make sure that results can be given to your midwife or doctor
- monitor and evaluate this screening.

Antenatal screening for Down syndrome and other conditions is overseen by Te Whatu Ora. To maintain the quality of this screening, it is closely monitored. Te Whatu Ora collects information for monitoring and evaluation. The information may be used in published reports. These reports do not contain any information that identifies you in any way.

Te Whatu Ora
Health New Zealand

Te Kāwanatanga o Aotearoa
New Zealand Government

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