

## Your results

Your maternity care provider will have the results of your screening in about a week to 10 days. They will be able to tell you whether you have a low or increased chance of having a baby with Down syndrome or another condition. They will discuss what this might mean for you.

- A low risk result means there is very little chance that your baby has any of the conditions screened for. It does not entirely rule out the possibility.
- An increased risk result means there is a higher chance that your baby has Down syndrome or another condition. This does not mean your baby has the condition.

Further tests, including diagnostic testing, are available to you. Only diagnostic testing can confirm whether or not your baby has a condition before he or she is born. Your maternity care provider will recommend that you discuss your results, your options and possible diagnostic tests with a specialist obstetrician.

## Diagnostic tests

Diagnostic tests look at the chromosomes from a baby's cells to find out whether the baby has Down syndrome or another condition. The diagnostic tests that may be offered after first trimester combined screening are:

- before 14 weeks: chorionic villus sampling – a sample of the developing placenta (whenua) is taken and sent to the laboratory for analysis
- from 14 weeks: amniocentesis – a sample of amniotic fluid (waters around the baby) is taken and sent to the laboratory for analysis.

The laboratory analysis will take about two weeks. For every 200 diagnostic tests, one or two pregnancies will miscarry. For this reason, some women choose not to have the tests. Your decision must be the right one for you.

Some conditions require different diagnostic testing. Your maternity care provider will discuss this with you.

## Your rights

The Code of Health and Disability Services Consumers' Rights protects your rights, including your right to have information and support for making informed choices about your health care. Your decisions about screening and any further testing are personal choices and will be respected. For further information see [www.hdc.org.nz](http://www.hdc.org.nz)

## Confidentiality

Your screening results will be sent in confidence to the maternity care provider who ordered them.

The Ministry of Health collects information to enable monitoring and evaluation of screening activities.

Details that could be used to identify you will be carefully protected.

## Additional information

The blood test is available free. There may be a charge for the NT scan.

Please ask your maternity care provider for more information.

Other sources of information include:

- the National Screening Unit at [www.nsu.govt.nz](http://www.nsu.govt.nz)
- the New Zealand Down Syndrome Association at [www.nzdsa.org.nz](http://www.nzdsa.org.nz) 0800 NZDSAI (0800 693 724)
- the New Zealand Organisation for Rare Disorders at [www.nzord.org.nz](http://www.nzord.org.nz)



## Antenatal Screening

# First Trimester Combined Screening

Screening for Down Syndrome  
*and Other Conditions*

## Antenatal screening for Down syndrome and other conditions

Most pregnancies result in the birth of a healthy baby. Sometimes a baby is born with Down syndrome or another condition affecting the baby's physical or mental development.

Screening during the first trimester of pregnancy (first three months) can provide some information about the chance of your baby having Down syndrome or another condition. Screening does not give a definite diagnosis and does not identify all babies who have a condition.

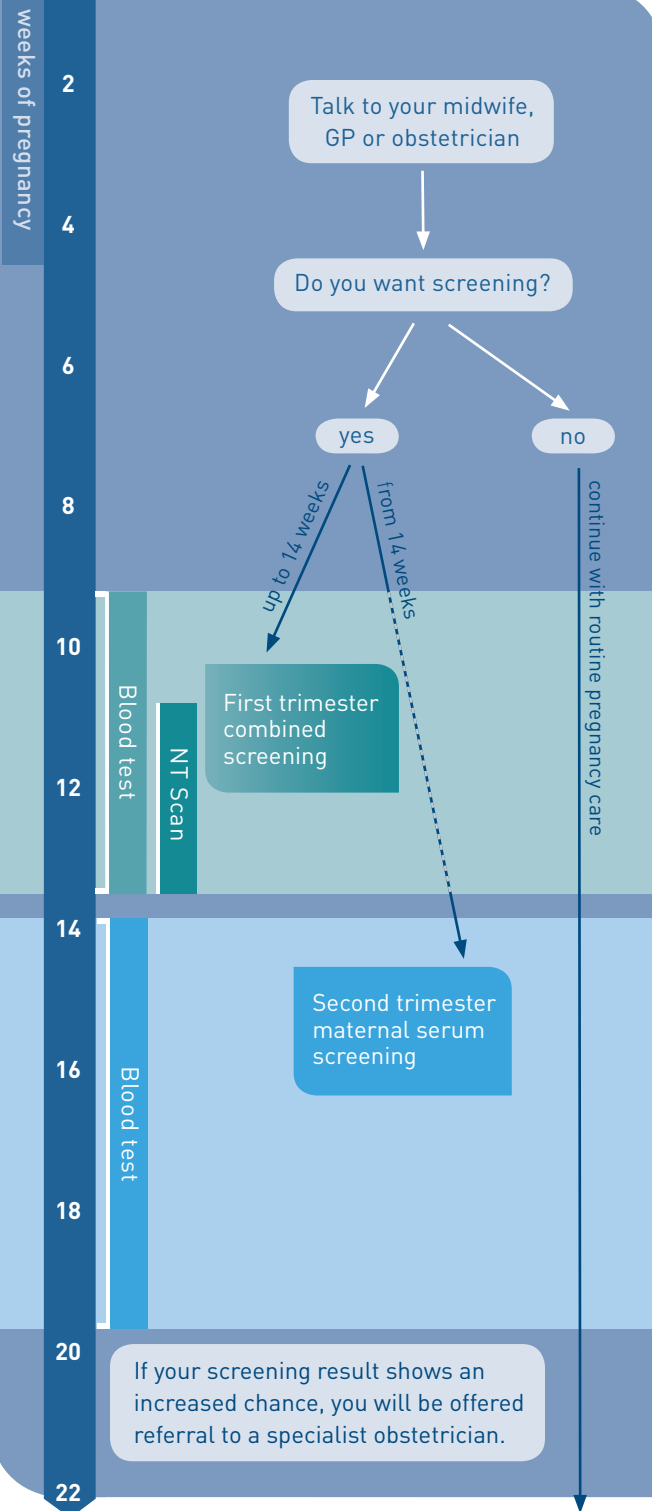
Pregnant women in New Zealand are offered antenatal screening for Down syndrome and other conditions. You may or may not want to find out if your baby has a condition. Information about the screening and how it works can help you make up your mind.

## Down syndrome

Down syndrome is a lifelong condition that causes varying degrees of delay in learning and development. It is a genetic condition that usually happens by chance, but may be inherited in some instances. Babies with Down syndrome may have many or only some of the features associated with the condition. They tend to have reduced muscle tone and may have heart, bowel or other medical issues.

While most people have 23 pairs of chromosomes in each cell of their body, people with Down syndrome have an extra copy of chromosome 21. This is why Down syndrome is sometimes called trisomy 21.

Down syndrome is the most common condition indicated by screening. Other conditions that may be indicated include trisomy 13, trisomy 18, neural tube defects or some rare metabolic or genetic disorders. These may be more or less severe than Down syndrome.



## First trimester combined screening

First trimester screening combines the results from a blood test and a nuchal translucency (NT) scan. Other information, such as your age, weight, and how far through your pregnancy you are, is also included in the calculation of your result.

## The blood test

The blood test is taken between nine weeks and 13 weeks 6 days of pregnancy. It measures the pregnancy-associated plasma protein-A (PAPP-A) and beta human chorionic gonadotrophin ( $\beta$ hCG). The levels of these tend to be different if you are carrying a baby with Down syndrome or another condition.

## Nuchal translucency scan

The NT scan is an ultrasound scan that is done between 11 weeks and 13 weeks 6 days of pregnancy. It measures the thickness of the fluid-filled space at the base of the baby's neck. Babies with Down syndrome and some other conditions tend to have a larger NT measurement.

Obtaining an NT scan result can sometimes be difficult. If an NT scan result cannot be obtained, a first trimester combined screening result cannot be calculated. If this happens, you will be offered second trimester maternal serum screening.

## Informed consent

You may choose not to have screening or not to have any further tests after screening. Choosing whether to have screening is an important decision for you and your baby. You need to make the decision that is right for you.

Talk to your maternity care provider if you have previously been pregnant with (or have had a child with) a significant physical or learning disability, or if you or your partner have a family history of a genetic condition. A referral to Genetic Services or a specialist obstetrician may be recommended before you are offered screening.