Other conditions

Each of the other conditions that can be indicated by this screening has a different potential to impact on your baby's growth and development. You should discuss this with your maternity provider and a specialist obstetrician/paediatrician or genetic counsellor so that you can make a fully informed choice about diagnostic testing.

How will I receive the diagnostic test results?

Your specialist or maternity care provider will give you the results of your diagnostic test and discuss what this will mean for you and your family. To enable you to make a decision about continuing or terminating your pregnancy, you will be offered information on the availability of counselling, support and other services.

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

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

Please ask your maternity care provider or specialist for more information. Other sources of information include:

- Northern Regional Genetic Service
  Toll free 0800 476 123
  www.healthpoint.co.nz/default,49655.sm
- Central and Southern Regional Genetic Service
  Toll free 0508 364 436
  www.healthpoint.co.nz/default,73079.sm
- The New Zealand Down Syndrome Association
  0800 NZDSAI (0800 693 724)
  www.nzdsa.org.nz
- The New Zealand Organisation for Rare Disorders
  www.nzord.org.nz
- The National Screening Unit
  www.nsu.govt.nz

MINISTRY OF HEALTH
MANATŪ HAUORA

New Zealand Government

This resource is available from **www.healthed.govt.nz** or the authorised provider at your local DHB.

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Antenatal Screening

Increased Chance

Screening for Down Syndrome *and Other Conditions*

You have an increased chance result

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. Your screening result indicates that there is an *increased chance* that your baby has Down syndrome or another condition. This does not necessarily mean that your baby has one of these conditions.

What happens next?

Your maternity care provider will talk with you about your options and will recommend that you see a specialist obstetrician to discuss your results, and whether you wish to have diagnostic testing. You are welcome to take support people with you to this appointment.

What is diagnostic testing?

Diagnostic testing is a way to find out whether your baby has Down syndrome or another chromosomal condition before he or she is born. The results will show whether or not a condition is present; they will not show the degree to which the baby may be affected.

Diagnostic tests look at the chromosomes from a baby's cells to find out whether the baby has Down syndrome or another condition. The two test options are:

- before 14 weeks – chorionic villus sampling, in which a sample of the developing placenta (whenua) is taken and sent to the laboratory for analysis
- after 14–15 weeks – amniocentesis, in which 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 a test. Your decision must be the right one for you.

Screening results may indicate other conditions, such as trisomy 13, trisomy 18, neural tube defects or other rare metabolic or genetic disorders. Some of these may require different diagnostic testing. Your specialist obstetrician will discuss this with you.

What do I need to think about before making my choice?

Before you consider diagnostic testing, it is important to think about what you plan to do if you find you are carrying a baby with Down syndrome or another condition. To help you with your choices:

- find out as much as you can about diagnostic testing
- find out as much as you can about the condition your baby may have
- talk to your maternity care provider, specialist and family/whānau
- take your time so that you are sure of the decision you make.

There are a number of reasons why you might choose to accept the offer of diagnostic testing. Some people wish to know if they are having a baby with Down syndrome or another condition so they can prepare for the birth and for family life. Others may wish to consider adoption or a termination of the pregnancy. You may choose to do nothing more and wait until the baby is born.

Your choices are your own, so please ask as many questions as you need to before making a decision.

If I choose not to have diagnostic testing, do I need to be referred to a specialist obstetrician?

It is your choice. If you accept the offer of a referral, this does not mean you have chosen to have diagnostic testing. It means you are taking the opportunity to talk with a specialist, who can give you further information to help you to make a fully informed choice.

What are Down syndrome and other conditions?

Down Syndrome

Down syndrome was first described in detail by an English doctor, John Langdon Down, in 1866. It is a lifelong, genetic condition that causes varying degrees of delay in learning and development. It usually happens by chance, but may be inherited in some instances.

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. The condition most commonly occurs as the cells divide at or soon after conception, resulting in an extra copy of chromosome 21:

- in all cells (95 percent of cases),
- in some cells (1 percent of cases, known as mosaic Down syndrome), or
- attached to another chromosome (4 percent of cases, known as translocation Down syndrome).

Down syndrome is usually recognisable at birth by the presence of certain physical characteristics, and diagnosis is confirmed by chromosome analysis.

The impact of Down syndrome varies from person to person. People with Down syndrome may have varying degrees of:

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

Many children with Down syndrome learn and develop, attend school and join in family/whānau and community life.