

Prenatal testing for Down syndrome

Fact Sheet

You may find this information helpful if you are considering having prenatal testing or if you've had a test that shows your baby may have or does have Down syndrome.

Down Syndrome Australia respects the right of families to make decisions appropriate to their own particular beliefs and circumstances. This information was prepared to provide you with some basic information as a starting point as well as links to other resources and support to help you make informed decisions about prenatal testing.

Down syndrome is a genetic condition – it is not an illness or disease. It occurs at conception as a result of an extra chromosome.

Permission to test

Prenatal testing is offered routinely to most women in Australia, and it can sometimes feel that it is assumed you will agree to testing. However, it is your choice whether or not to have prenatal testing and it is important not to be rushed into it. You should only have testing if you have given your permission. It is important to understand what the tests measure and if there are any associated risks before you give your permission.

Information about prenatal testing

Prenatal tests are divided into two types: screening tests and diagnostic tests.

Screening tests

These are tests that can be carried out on the mother's blood in the first or second trimester of pregnancy. In the first trimester, you would have the blood test as well as an ultrasound that measures nuchal translucency (the fluid in the baby's neck) and also its nasal bone. Most pregnant women are also routinely offered

an ultrasound at 19-20 weeks of pregnancy, which can sometimes show other signs of Down syndrome.

The likelihood of your baby having Down syndrome is calculated and if the result shows there is an increased likelihood, you may be offered a diagnostic procedure such as chorionic villus sampling (CVS) or amniocentesis.

Diagnostic tests

Both CVS and amniocentesis are invasive procedures that involve obtaining a sample of the baby's genetic material for examination. Both tests increase the risk of miscarriage.

There is also a new non-invasive prenatal blood test (NIPT), which is available in the private health system in Australia. While this test is highly accurate, detecting 99% of pregnancies with a diagnosis of Down syndrome, women are usually advised to be sure by also having a CVS or amniocentesis. NIPT is costly and there is currently no Medicare rebate.

Prenatal genetic counsellors can help you with information about all the different types of prenatal testing, and you can find more detailed information about the different tests in "Your choice, screening and diagnostic tests in pregnancy." <http://www.mcric.edu.au/Downloads/PrenatalTestingdecisionAid.pdf>

Making a decision

If diagnostic testing reveals Down syndrome, parents are usually offered a choice about continuing or terminating their pregnancy. This can be a very difficult and emotional time and it is important to take some time to carefully consider all the available information about Down syndrome and your options.

What is Down syndrome?

Down syndrome is a genetic condition – it is not an illness or disease. It occurs at conception as a result of an extra chromosome.

In Australia, around 1 in 1,100 babies are born with Down syndrome. Down syndrome occurs across all ethnic and social groups and to parents of all ages and has nothing to do with anything the mother or father did before or during pregnancy. Down syndrome almost always occurs randomly.

People with Down syndrome have some characteristic physical features, some health and development challenges, and some level of intellectual disability. Because no two people are alike, each of these things will vary from one person to another. When a baby is born, there is no way to tell what level of intellectual disability the child may have. Nor can we predict the way in which this may affect a person's life. However, we do know that having Down syndrome will not be the most important influence on how that person develops and lives their life. Instead, family, environmental, cultural and social factors will shape their life, just like everyone else.

While some aspects of life may be more challenging than for a typical person, such as healthcare and education, people with Down syndrome now commonly take part in mainstream school and post-school education,

sports, performing and visual arts, community volunteering and the workplace. A growing number of people with Down syndrome live more independently and are choosing to form relationships and get married. Life for people with Down syndrome these days is very different from how it used to be, and looks even more hopeful for the future.

Many parents worry about how a child with Down syndrome will affect their family. Of course, every family is different, but personal stories and research show that most families that have a child with Down syndrome are stable, successful and happy and that siblings often have greater compassion and empathy. In fact, some studies have shown that families of children with Down syndrome have lower rates of divorce than the national average.

You can find more information about living with Down syndrome here:

http://www.downsyndrome.org.au/what_is_down_syndrome.html

Where to get information and support

You will undoubtedly have many other questions about Down syndrome, and how it will affect your baby and your family. The Down syndrome organisation in your state or territory is a good place to contact for non-directive information, ask questions and speak to, or arrange to speak to, a parent of a child with Down syndrome, if that's what you would like to do.

Contact 1300 881 935

This 1300 number will connect you to your state or territory Down syndrome support organisation. You will also find lots of useful information on their websites which you can reach via the Down Syndrome Australia website at www.downsyndrome.org.au