Prenatal testing for Down syndrome

Fact Sheet

Down Syndrome Australia

Prenatal testing for Down syndrome

You may find this information helpful if you are considering having prenatal testing or if you have 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.

Permission to test

Prenatal testing is offered routinely to most women in Australia, but it is your choice whether or not to have prenatal testing and it is important to take time to ensure that you have understood all the correct information about the tests, their risks and limitations and the conditions that are being tested for. You should only have testing if you have given your permission.

Information about prenatal testing

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

Screening tests

These are tests that are used to identify the likelihood of your baby having Down syndrome. They do not provide a definitive diagnosis. There are two types of screening tests - maternal blood screening and ultrasound examination of the baby.

Blood screening tests

These are tests that are used to identify the likelihood of your baby having Down syndrome. They do not provide a definitive diagnosis.

The relevance (sensitivity and specificity) of any screening test is dependent upon a number of variables including the type of material that is being tested ie. Biochemical analytes vs DNA, the brand/performance characteristic of the particular test being used and other pregnancy related factors such as maternal age. It is important to ask about the specific performance of any screening test you are offered and to be aware that sometimes a test can suggest a "high chance" result when the baby does not have Down syndrome; or a "low chance" result when the baby does have Down syndrome.

The **1st trimester combined screening test** is available between 11-13 weeks of pregnancy. It costs approximately $350 after the Medicare rebate.

It combines a measurement of a number of analytes that are present in maternal blood with:

- information from an ultrasound examination including a measurement of nuchal translucency (the fluid visualised behind the baby's neck)
- the absence or presence of a nasal bone
- maternal age
- other relevant pregnancy information

If the resulting calculation indicates an increased chance of your baby having Down syndrome, you may be offered a confirmatory diagnostic procedure such as chorionic villus sampling (CVS) or amniocentesis. (See next section.)

Second trimester screening is available from 14-22 weeks of pregnancy.

It uses the measurement of several substances in the blood, gestation of the pregnancy, maternal age and weight to provide an estimate of the chance of your baby having Down syndrome. If the resulting calculation indicates an increased chance of your baby having Down syndrome, you may be offered a confirmatory diagnostic test (amniocentesis).

Non-invasive prenatal testing (NIPT) is a blood test available from the 10th week of pregnancy. NIPT costs approximately $450 and there is currently no Medicare rebate.

This test analyses both maternal and foetal cell-free DNA circulating in maternal blood to provide either a high probability or low probability of your baby having Down syndrome. If the resulting calculation indicates an increased likelihood of your baby having Down syndrome, you will usually be offered a confirmatory diagnostic procedure such as chorionic villus sampling (CVS) or amniocentesis.

Ultrasound screening

Most pregnant women are also routinely offered an ultrasound examination at 19-20 weeks of pregnancy, which can sometimes show other subtle signs of Down syndrome. If this happens you may be offered an amniocentesis as a confirmatory diagnostic procedure.

Diagnostic tests

Both CVS and amniocentesis are invasive procedures that involve obtaining a sample of the baby's genetic material for examination. They are the only way to get a definitive diagnosis of Down syndrome prenatally. These invasive tests increase the risk of miscarriage (above the usual background risk).

Your obstetrician, GP, midwife or a prenatal genetic counsellor 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." www.mcri.edu.au/sites/default/files/media/documents/prenatal-screening-decision-aid.pdf. 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.

Regardless of the results of your screening tests you are under no obligation to undertake further diagnostic testing. This choice is entirely up to you. People report lower levels of regret when these decisions are made with access to high quality, up-to-date information about the condition, a lack of time pressure, and access to unbiased and nondirective counselling and support by health care providers.

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. It can be helpful to talk with other families who have a child with Down syndrome to learn more about their experiences. This can be arranged by contacting the Down Syndrome Association in your State. You may also want to get more information about other options that are available such as placing the baby for adoption after birth.

There are also online support groups available where you can connect with other people with a prenatal diagnosis or high probability screening result and parents of young children with Down syndrome.

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 typically developing 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: 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 find lots of useful information on their websites which you can reach via the Down Syndrome Australia website at www.downsyndrome.org.au.

Useful Links

Information resources

- Down Syndrome Australia website - www.downsyndrome.org.au
- Down Syndrome Diagnosis Network - www.dsdiagnosisnetwork.org
- Raising Kids Network - raisingchildren.net.au
- Lettercase - lettercase.org
- National Down Syndrome Congress Information for new and expectant parents - www.ndsccenter.org/programs-resources/new-and-expectant-parents

Support groups

- State based Down syndrome organisations – contact your local organisation on 1300 881 935.

Online Groups

- Some state organisations have online support groups- contact your local organisation on 1300 881 935.
- T21 Mum Australia - www.facebook.com/t21mum

Video interviews and talks of people with Down syndrome

- ABC You can't ask that – Down syndrome - iview.abc.net.au/programs/you-cant-ask-that/LE1617H002S00#pageloaded
- Karen Gaffney – All lives matter - www.youtube.com/watch?v=HwxjoBQdn0s
- Dear Future Mom - www.youtube.com/watch?v=Ju-q4OnBtNU
- BBC Three – interviews – "Things people with Down's syndrome are tired of hearing" - www.youtube.com/watch?v=AAPmGW-GDHA

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