Testing for Down’s syndrome in pregnancy

Choosing whether to have the tests is an important decision, for you and for your baby.

This booklet gives you some information about Down’s syndrome and about testing for it, so you can decide whether to have the tests.
Contents

This booklet is divided into sections which describe screening and diagnostic tests for Down's syndrome you may be offered during your pregnancy.

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What happens if I get a low risk result, so I am not offered more tests?

Diagnostic tests for Down's syndrome
What can you tell me about diagnostic tests?
What is an amniocentesis?
What is chorionic villus sampling (CVS)?
Are these procedures safe?
Are the tests painful?
How long does it take to get the results?
How would I get my results?

What are the possible results from diagnostic tests?
The baby does not have Down's syndrome
The baby does not have Down's syndrome but the tests have shown some other problem has been identified.
The baby has Down's syndrome.

Further information

Your notes

References
All pregnant women are now offered tests for Down's syndrome. This booklet gives you some information about Down's syndrome and about testing for it, so you can decide whether to have the tests.

Choosing whether to have the tests is an important decision, for you and for your baby. You need to make the decision that is right for you, so please read this booklet carefully.

Your midwife or your GP will talk to you about testing for Down's syndrome. They will be happy to answer your questions - so please do ask if there is anything you are not clear about.

What is Down's syndrome?

There is no such thing as a typical person with Down's syndrome. Like all people, they vary a lot in appearance, personality and ability. People with Down's syndrome have learning difficulties.

Some have more serious difficulties than others. It is hard to tell in babies how much they will be affected as children, or when they are grown up. Some adults with Down's syndrome are able to get jobs and live fairly independent lives. However, most people with Down's syndrome need long-term help and support.

A number of health problems are linked to Down's syndrome. But again, people vary, and some people with Down's syndrome enjoy good health.
Problems which are linked with Down’s syndrome include heart problems and reduced hearing and vision. Many of the problems can be treated, and frequent health checks can make sure that any problems are picked up as early as possible. Most people with Down’s syndrome live to be 50 years of age and some live to be over 70. Alzheimer’s disease (a form of senile dementia) may affect people with Down’s syndrome at an earlier age than other people.

How common is Down’s syndrome?

People do not usually expect to have a baby with Down’s syndrome. It does not usually run in families. Some people think that only older women can have a baby with Down’s syndrome, but this is not true. Anyone can have a baby with Down’s syndrome, but the risk does go up with age. The older a mother is, the more chance she has of having a baby with the condition. For example, the chance of having a baby with Down’s syndrome is one in 1500 for women who are 20 years old, one in 900 for women who are 30 years old, and one in 100 for women who are 40 years old. Some people prefer to think of the risk as a percentage. A risk of one in 1500 is the same as a 0.07% risk. A risk of one in 900 is the same as a 0.1% risk. A risk of one in 100 is the same as a 1% risk.

Anyone can have a baby with Down’s syndrome.
What causes Down's syndrome?

Inside all the cells of our bodies are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. Most people have 23 pairs of chromosomes in each of their cells. When our bodies produce the special cells needed to make babies, the chromosome pairs divide and rearrange themselves. Sometimes these pairs of chromosomes do not divide correctly, and this causes the baby's cells to have an extra copy of chromosome number 21. This causes Down's syndrome. (And it is the reason why one of the medical names for Down's syndrome is Trisomy 21.) The extra chromosome cannot be removed from cells, so there is no cure for the condition.

If the chromosomes divide incorrectly, this happens by accident. It is not caused by anything parents have done or have not done.

Testing for Down's syndrome during pregnancy

Should I have the tests for Down's syndrome?

Only you can decide that. Some women want to find out if their baby has Down's syndrome, and some do not. Information about the tests and how they work can help you make up your mind. This booklet gives the main facts, and tells you how you can get more information if you want to know more.
Will the tests tell me for certain if the baby has Down's syndrome?

We do not offer all women a test that will tell them for certain. This section explains why.

There are tests which give definite information. These are called **diagnostic tests**. The problem is that having a diagnostic test increases the risk of miscarriage. This is why we do not offer diagnostic tests to all women.

Instead, we offer tests in two stages. We begin by offering all women a test that carries no risk of miscarriage. This type of test is called a **screening test**. Screening tests do **not** give a definite answer, but they do tell us which babies have an increased risk of having Down’s syndrome. We then offer diagnostic tests to the women at increased risk. (There is more information on diagnostic tests later in the booklet.)

If your screening result falls between ‘one in two’ and ‘one in 250’, we will offer you a diagnostic test. (A risk of one in 250 is the same as a 0.4% risk.)

It is very important to understand that screening tests cannot tell you that your baby definitely does or definitely does not have Down’s syndrome.

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**Screening tests do not give a definite answer.**

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**For every 800 pregnant women who choose screening tests, 40 will be considered high risk.**

One will definitely have Down’s syndrome and may choose to either continue the pregnancy or end the pregnancy.