



Information for women with a higher chance of having a baby with **Down's syndrome**

This leaflet is for women who have had a screening test for Down's syndrome and have been told that they have a higher chance of having a baby with Down's syndrome.

The leaflet explains the result of the screening test and gives information about what will happen next.

The leaflet also gives information about people with Down's syndrome.



Screening for Down's syndrome

The result of the screening test for Down's syndrome tells you what your chance is of having a baby with Down's syndrome.

Remember that the lower the number, the higher the chance. Here are some examples.

- **One in 50** means there is one chance that your baby has Down's syndrome and 49 chances that your baby does not have Down's syndrome.
- **One in 450** means that you have less chance of having a baby with Down's syndrome. That is, one chance that your baby has Down's syndrome and 449 chances that your baby does not have Down's syndrome.

All women have a chance of having a baby with Down's syndrome, including women whose result shows they have a lower chance (less than one chance in 150).

If you have a higher chance of having a baby with Down's syndrome, you can discuss the result and you will be offered chromosome testing.

The screening test you had for Down's syndrome shows your chance of having a baby with Down's syndrome. To work out the chance of your baby having Down's syndrome, a computer programme used:

- your age;
- the results of the blood test;
- your ethnic background;
- information about whether you smoke;
- the results from the early pregnancy ultrasound scan test; and
- your weight.

The midwife will check this information with you. If it is not correct, the computer will work out your result again.

The chance of having a baby with Down's syndrome increases as you get older.

The screening test result takes into account your age. The screening test for Down's syndrome is more accurate in estimating your chance of having a baby with Down's syndrome than using your age alone.

What the Down's syndrome screening result means

The result does **not** mean that your baby has Down's syndrome. The screening test only shows which women have a higher chance of having a baby with Down's syndrome.

Most women whose result shows they have a higher chance do not have a baby with Down's syndrome. A result showing a higher chance can be caused by other reasons, and the result does not mean there is definitely a problem.

The result means that you are in the group of women being offered a

chromosome test. About 3% of women (that is, three out of 100) who have the screening test for Down's syndrome are offered chromosome tests.

No test is 100% accurate, but the chromosome tests for Down's syndrome are very reliable. They usually give you a definite answer.

To do the diagnostic test, a sample of the fluid from around your baby is needed. The procedure to get this fluid is called an amniocentesis.

Only you can decide if you want to have this procedure. Your midwife or obstetrician can explain the test to you.

What is Down's syndrome?

Down's syndrome is a lifelong condition and will result in some degree of learning disability.

It is a genetic condition which is caused by having an extra chromosome. Instead of 46 chromosomes there are 47.

The extra chromosome appears when or shortly after the sperm fertilises the egg. It is not known what makes this happen. The extra chromosome cannot be removed from cells and so there is no cure even if the condition is diagnosed before the baby is born.

Babies with Down's syndrome

These are the signs that health professionals look for if they suspect

Down's syndrome. A baby with Down's syndrome can have all or some of them.

- They often have eyes that slant upward and outward. Their eyelids can have an extra fold of skin (epicanthic fold) which appears to make the slant more noticeable.
- They may have looser muscles and joints than other babies.
- They may have a birth weight that is lower than average and put on weight at a slower rate than other babies.
- The back of the baby's head may be flatter than average.
- Many babies with Down's syndrome have a single crease which runs right across the palm of their hand.
- Babies with Down's syndrome still look more like the rest of their family than anyone else.

Children with Down's syndrome

usually develop and learn more slowly than other children.

There are no tests that can tell you what difficulties your baby may have. Most babies with Down's syndrome will learn to walk and talk. Most children with Down's syndrome go to mainstream schools and learn to read and write.

They may have some medical problems that need special attention and treatments. About half of children with Down's syndrome will have a heart problem, which may need an operation. Some of the heart problems are life-threatening. About

20% of children (one child in five) with Down's syndrome will die before the age of five.

With appropriate medical care, most children and adults with Down's syndrome can lead healthy lives.

Adults with Down's syndrome can live partly-independent lives, choosing their friends and partners and working or contributing to society in other ways. They may need help with caring for themselves.

The average life expectancy of people with Down's syndrome is 55 years. Many live into their 60s.

Alzheimer's disease (a form of dementia) may affect people with Down's syndrome at an earlier age than other people.

There are support services available for families who have a child with Down's syndrome. These include the Down's Syndrome Association (their address is at the end of this leaflet).

Your hospital appointment

You may already have been offered an appointment with the hospital midwife who specialises in antenatal screening, or with a doctor (or both). This appointment is to explain the result of the screening test for Down's syndrome to you in more detail and to ask you if you want to have an amniocentesis procedure so that chromosome tests can be done.

The hospital midwife and doctor will explain more about the amniocentesis procedure and how it

is done. You do not have to have this procedure. Some women decide not to have this procedure. The midwife and doctor will support whatever choice you make.

You may want more time to think before you decide what to do next.

You may have a number of questions to ask the midwife or doctor and may want to have another appointment. You may want your partner or a friend to come with you.

If you decide to have an amniocentesis, you may be offered a separate appointment as soon as possible.

This procedure is not available in every hospital and you may be offered an appointment at another hospital where it can be done.

Amniocentesis

Amniocentesis is necessary if you want to know if your baby has a chromosome problem before it is born.

Chromosome tests are a very accurate way of telling whether your baby does or does not have Down's syndrome.

An amniocentesis is a procedure to remove a small amount of amniotic fluid (about 15 to 20 millilitres) from around your baby in your womb.

The cells from your baby that are floating in this fluid can be tested in the laboratory to look at the chromosomes.

In Wales, two tests are done on the amniotic fluid to look at your baby's chromosomes. These are called the polymerase chain reaction (**PCR**) test and the **karyotype** test. The result of the PCR will be available within three working days and the result of the karyotype will be available in about two weeks.

Amniocentesis has a chance of causing a miscarriage. If you have an amniocentesis, there is a 1% chance that you could have a miscarriage (that means one in every 100 women could lose their baby). The miscarriage can be the loss of a baby that was affected or unaffected by Down's syndrome.

You can find more information about this test in a leaflet called 'Amniocentesis', which your midwife can give you or it is available from www.antenatalscreening.org.

You may want to think about what you might do if the test result shows there is an abnormality. Some women will want to prepare themselves for the birth knowing that their baby has a problem. Other women may decide to end the pregnancy. Some women decide to continue with the pregnancy and consider having their baby adopted.

You can get more information about Down's syndrome and antenatal tests from your midwife or local hospital antenatal clinic.

More information

You can get more information from:

Antenatal Results and Choices (ARC)

ARC is an independent charity providing specialist information and support throughout antenatal screening and testing and its consequences.

Antenatal Results and Choices (ARC)

73 Charlotte Street

London

W1T 4PN

Helpline: 0207 631 0285

Website: www.arc-uk.org

E-mail: info@arc-uk.org

Down's Syndrome Association (DSA)

The Down's Syndrome Association support services include advisers who are available to talk to families at every stage in the testing process, throughout childhood and adulthood.

Down's Syndrome Association (DSA)

Langdon Down Centre

2a Langdon Park

Teddington

TW11 9PS

Phone: 0845 230 0372

Fax: 0845 230 0373

Website:

www.downs-syndrome.org.uk

E-mail:

info@downs-syndrome.org

Local contact details