

Is the amniocentesis test painful?

Most women say afterwards that the test is uncomfortable rather than painful and feels similar to period pain. Women say that the thought of it is worse than the actual test.

What happens after the amniocentesis test?

You will probably be at the hospital for 45 minutes, the test itself only takes a few minutes. We encourage you to bring a companion with you for support. It is a good idea to take things easy for a couple of days, avoiding any heavy lifting or strenuous exercise. The "period pain" feeling may persist for 24-48 hours. This is not unusual and should settle with Paracetamol, which is safe to take in pregnancy. If you have excessive pain, are leaking any fluid or bleeding or develop a high temperature, please contact your midwife, GP or the local hospital.

When do I get the results?

A rapid test that can detect trisomy 21 (Down Syndrome) and trisomy 13 and 18 (two other less common chromosome disorders) will be carried out on all samples. The results of this test will be reported to you within 72 hours by your Fetal Medicine midwife. The sex chromosome pattern can also be analysed using this test, but this will only be done if there is an indication to do so.

In addition to the rapid test, a more detailed test (known as a karyotype) will be performed on the sample. This test enables us to look at all the chromosomes in greater detail and usually takes about 2 weeks because it involves growing the cells in the laboratory.

We will telephone you with the early result and then send you a written result by post, once the chromosome test is complete. If a problem is discovered, you will be given the opportunity to discuss this with us. We will also notify your own hospital and GP of the results.

What if the results are abnormal?

If the results show anything abnormal, you will be told what the abnormality is and how this could affect your baby. You will have the chance to discuss the results fully before making any decisions.

HIV infection

At the moment our existing lab facilities and current Health and Safety law limit the tests we can perform on an amniocentesis sample from a woman who is known to be HIV positive. There may be a very small risk that an amniocentesis test could cause the HIV virus to be passed to the developing baby. We would encourage women who are HIV positive to discuss this with their specialist midwife or doctor to obtain further information.

And finally:

When you come to your amniocentesis appointment, a Fetal Medicine midwife will be available for you to talk to. You will be given an opportunity to discuss the amniocentesis test fully before making a final decision on whether to go ahead. We recognise that everyone will have his or her own particular questions and concerns to discuss. The midwife will be able to answer any questions you may have before you have the test.

For more information:

If you need more advice about any aspect of The Amniocentesis Test, you are welcome to contact:

Clinical Genetics Department

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 Fax: 01224 559390

Tayside (main base Dundee)
Tel: 01382 632035 Fax: 01382 645731

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West of Scotland (main base Glasgow)
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Seen in clinic.....

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This leaflet was written by Guys & St Thomas' Clinical Genetics. Updated by Genetic Interest Group Scotland

The Amniocentesis Test



Some general information

Introduction

This test is not offered as routine. It is used often to test for Down syndrome, the commonest chromosome disorder.

Usually we have 46 chromosomes (23 pairs) in each cell within our bodies. Chromosomes are made up of the vital genes that make us grow and develop into healthy individuals. A small number of babies, children or adults may have a chromosome disorder, which usually means they have too many or too few chromosomes in their cells. In most cases, Down syndrome is caused by the presence of an extra chromosome 21, so instead of having the usual two copies of chromosome 21 there are three copies (also known as trisomy 21).

Less commonly Amniocentesis may be used to test for other specific genetic disorders.

Who should consider amniocentesis?

The final decision about any test in pregnancy is yours the following women may consider amniocentesis:

1. Women who have had a high-risk result from a nuchal translucency scan or a blood test designed to find out the risk of having a baby with Down syndrome.
2. Women who have a potential problem found on ultrasound scan, which may suggest the pregnancy has a chromosomal abnormality.
3. Women who have had a previous pregnancy terminated for a genetic problem.
4. Couples who have had a baby with Down syndrome or those who have a family history of genetic disorders, including rare inherited diseases.
5. Women 36 years and over who have missed the opportunity to have Down screening. The risk of having a baby with Down syndrome gets higher as the mother gets older, as shown in the table.

Your Age Risk of Having a Down Syndrome Baby

25	1 in 1500
30	1 in 800
35	1 in 300
38	1 in 180
40	1 in 100
45	1 in 30

What are the risks of amniocentesis and is it reliable?

Amniocentesis has now been available for a number of years and we know that the test can sometimes cause a miscarriage. Every year in Britain over 25,000 women have the test performed, so we know a lot about its safety and accuracy.

We know that the test can sometimes cause a miscarriage. Approximately 1 in every 100 (1%) women who have the test will miscarry as a result of the procedure and we do not really know why this happens.

Other than this, there is no evidence that amniocentesis is harmful to your baby as the test is carried out under ultrasound guidance.

No test is absolutely perfect, but the chromosome test for Down syndrome is very reliable. It fails to give a clear result in less than 1 in 1000 cases. If you are having the amniocentesis for other genetic disorders, you should discuss the accuracy of the test with your genetic doctor or counsellor.

What are the benefits of having an amniocentesis?

The amniocentesis test can help to reassure you that your baby does not have a chromosome disorder which will affect growth and development. If the result shows that your baby does have Down syndrome, you will be able to prepare for the arrival of a baby with Down syndrome or make another decision which may include termination of pregnancy

Will the test tell me about anything other than Down syndrome?

Although you may be having this test to detect Down syndrome, which involves chromosome 21, chromosomes 13 and 18 will also quickly be checked. This means that even if your baby does not have Down syndrome, the test may occasionally detect problems with the other chromosomes instead. If the results show anything abnormal you will be told what the abnormality is and how this will affect your baby.

When is the amniocentesis performed?

The test is most safely performed from 16 weeks of pregnancy onwards.

How is the amniocentesis performed?

The test involves taking a small amount of the amniotic fluid (water) that surrounds the baby in the womb. This contains cells from the baby that are tested in the laboratory.

Before the test is performed, an ultrasound scan is carried out to check your dates and the position of both the baby and the placenta (afterbirth). You will not need a full bladder for this scan.

The skin over the womb area is cleaned with antiseptic solution. A fine needle is then passed into the womb and a sample of the fluid that surrounds the baby is removed with a syringe and sent to the laboratory. The position of the baby and the needle are monitored throughout the procedure by ultrasound. Very occasionally the doctor is unable to get enough fluid at the first attempt and may need to re-insert the needle.

