

Information for patients on amniocentesis

Key Points

- Amniocentesis is a procedure in which a small amount of the amniotic fluid surrounding the baby in the womb is removed by passing a fine needle through the mother's abdomen.
- Amniocentesis can detect if a baby has a chromosomal disorder, such as Down syndrome.
- Amniocentesis is performed after 15 weeks of pregnancy.
- About 1 in every 100 women (or 1%) miscarry as a result of the procedure.
- If a disorder is diagnosed, then you should be given information and the opportunity to discuss this with a specialist.

What is amniocentesis?

Amniocentesis is a diagnostic procedure carried out during pregnancy. It is most commonly used to check the baby's chromosomes for specific disorders, such as Down syndrome.

When and how is amniocentesis performed?

Amniocentesis is performed after 15 weeks. Ultrasound jelly is applied over your abdomen and a detailed ultrasound scan performed. The positions of both the baby and the placenta (afterbirth) determined (Fig. 1).

Your skin is then cleaned where the needle will be inserted. Under ultrasound guidance a fine needle is pushed through your skin and womb, to reach the amniotic fluid. A small sample of the fluid (about 15 mls) is removed using a syringe, this will reform within the womb within 2-4 hours. This fluid is usually yellow in colour but may be stained with blood.

The time taken to remove the fluid is about a minute. The needle is then taken out and the baby is checked on ultrasound. The amniotic fluid, which contains some of the baby's cells, is sent to the laboratory for testing (see section on 'What are the laboratory tests?').

Occasionally insufficient fluid is withdrawn and the procedure needs to be repeated. This may be due to the position of the baby or movement of the amniotic membrane.

If your blood group is Rh (rhesus) negative, you will be advised to have an injection of anti-D immunoglobulin after the procedure to prevent you from developing antibodies against the baby's blood cells. You can find more information about this in *Guidance on the routine use of anti-D prophylaxis for RhD negative women: information for patients*, by the National Institute for Health and Clinical Excellence (NICE).

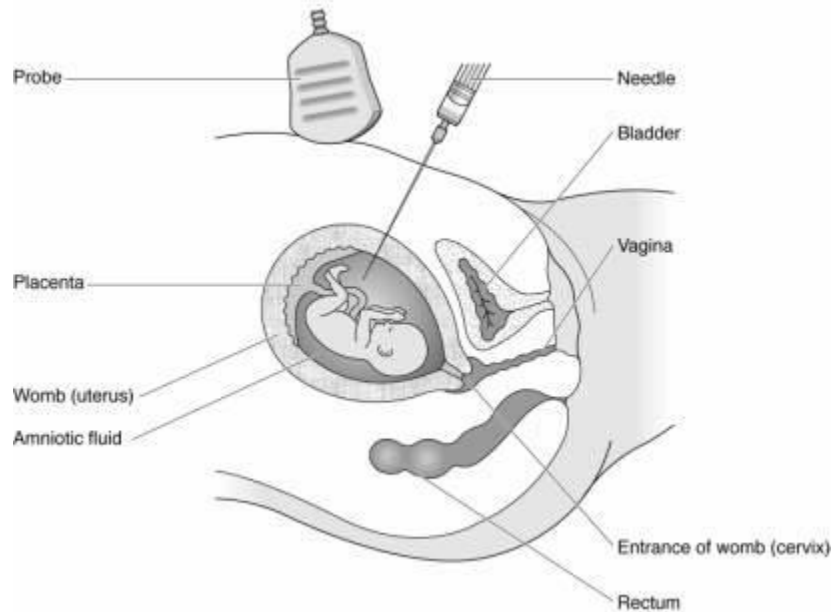


Fig.1 Schematic representation of the amniocentesis procedure

Is amniocentesis painful and what should I do afterwards?

Most women say that having amniocentesis is uncomfortable rather than painful, a bit like a period pain. Women often feel anxious but generally women say that the thought of it is worse than the actual test. After the procedure you should rest, if you wish to, for the remainder of the day. It is a good idea to take things easy for a couple of days after the test, avoiding any heavy lifting or strenuous exercise.

You may notice a spot of blood on your underwear and mild cramping afterwards for up to 24 hours. This is normal. It is perfectly safe to take 500 - 1000 mg of paracetamol to ease the pain.

However, if you experience any more significant or unusual symptoms following the test, such as feeling shivery (as if you have 'flu), fluid loss, bleeding or contractions you should seek advice immediately (see separate sheet "Advice for Women following Amniocentesis").

What are the risks of amniocentesis?

Every pregnancy carries a risk of miscarriage. As amniocentesis involves putting a needle through the wall of the womb, it may sometimes cause a miscarriage due to inflammation or infection in the womb. The additional overall risk of miscarriage from amniocentesis is approximately 1%. In other words, about 1 in every 100 women who have amniocentesis under ultrasound guidance after 15 weeks will miscarry.

There is a small risk, less than 1 in 1000 women who have amniocentesis, that the procedure will cause a serious infection that affects the mother's health. A maternal infection can be caused by the needle puncturing the bowel, or by skin contamination. These risks can be minimised by standard procedures to reduce infection.

Who should consider amniocentesis?

Women may consider amniocentesis if they:

- have received a high-risk screening result from a blood test for Down syndrome.
- have received a result from a scan which shows certain features, such as fluid collection at the back of the baby's neck (nuchal translucency), or a heart defect which indicates the baby may have a chromosomal abnormality.
- have had a previous pregnancy affected with a disorder.
- have one or more relatives affected with a genetic disorder.
- want to know for certain whether the baby has a disorder.

What are the laboratory tests?

There are two stages of the laboratory results. These two stages are performed in different ways, evaluating the baby's chromosomes. These are:

- **a rapid test** (PCR or FISH test) which checks for specific chromosomes. Results from this test are usually available after three working days. The disorders that can be detected by the rapid test include Down syndrome (trisomy 21 - caused by an additional chromosome 21), Edward syndrome (trisomy 18 - caused by an additional chromosome 18) and Patau syndrome (trisomy 13 - caused by an additional chromosome 13) and if requested sex chromosome disorders.
- **a full karyotype** which checks all the baby's chromosomes. Results from this test are usually ready within two to three weeks.

We will arrange with you how you would like to receive your results (by phone at a pre-arranged time or by post)

How reliable are the laboratory tests?

For most women the laboratory test will give a definite 'yes' or 'no' answer. The result will let you know, one way or the other, whether the baby has the disorder the test was looking for.

However, with the full karyotype test there is small chance, about 1 in 100, that it will not give a clear result. This could be because it was not possible to analyse the sample in the laboratory (culture failure) or that the sample was analysed and the test gave an uncertain result. Occasionally there are problems with the rapid test and due to blood staining it is not feasible to give a "quick result".

Even if the rapid test does not find a problem, the full karyotype may show an abnormality (not detectable with the rapid test) when the full results become available about two weeks later. If the result is not clear, it may be necessary to offer you a repeat amniocentesis, or other tests

What will the results tell me?

The laboratory tests are able to detect a range of disorders, but not all.

- Most women who have amniocentesis will have a 'normal' result; in other words, their baby will be born without the disorder(s) the test was looking for.
- Some women will be informed that the baby has the disorder that the test was looking for.
- Very occasionally women have amniocentesis to detect Down syndrome and it is discovered that the baby has another chromosomal disorder.
- Very occasionally the baby who has a 'normal' result can be born with another chromosomal or genetic condition that had not been tested for. A normal karyotype result does not exclude every disorder.

What are my choices if the results are abnormal?

If the results are abnormal, these will be discussed fully by the Doctor and any appointments with geneticists or paediatricians arranged. For the majority of disorders, there is no treatment or cure. You will need to consider what is best for you and the baby. This might be to:

- continue with the pregnancy and use the information you have gained to help prepare for the birth and aftercare of your baby.
- terminate this pregnancy.

If you consider a termination of pregnancy following an abnormal result, you will be given full information about what this involves. It will depend upon how many

weeks pregnant you are when you make the decision. Ending the pregnancy may involve going into labour.

Is there anything else I should know?

- If you are HIV positive and you decide to have amniocentesis, this might increase the risk of passing HIV on to your baby. You may be offered treatment with HAART (highly active antiretroviral therapy) if you are not already taking it. This reduces the risk of the HIV virus infecting the baby.
- If you carry hepatitis B or hepatitis C viruses, there is in theory a possibility that amniocentesis might increase the risk that you pass this onto your baby. There is not enough information to be sure about whether this risk is real or not.

Sources and acknowledgements

This information is based on the Royal College of Obstetricians and Gynaecologists (RCOG) guideline **Amniocentesis and Chorionic Villus Sampling** (published by the RCOG in January 2005 and due for a review in January 2008). The guideline contains a full list of the sources of evidence we have used. You can find it online.

Patient agreement to the procedure of Amniocentesis

Statement of Doctor performing the amniocentesis

I have explained the procedure to the patient and she has had the opportunity to read our patient information leaflet on the test. In particular I have discussed

- the intended benefits of the procedure, which include testing for Down syndrome
- any serious or frequently occurring risks from the test and she is aware that a miscarriage can occur in approximately 1/100 women following the test

Signature of Doctor.....

Name (print)..... **Date**...../...../.....

Statement of Client

- I agree to the amniocentesis
- I am aware that the procedure involves the introduction of a thin needle through my abdomen into the amniotic fluid within the womb
- I understand that the procedure will not involve local anaesthesia
- I am aware that the risk of miscarriage from amniocentesis is approximately 1% above the background rate
- I am aware that there is a small risk of laboratory failure

Signature of

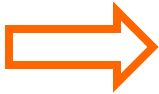
Client.....

Name (print)..... **Date**...../...../.....

Advice for Women following the Amniocentesis

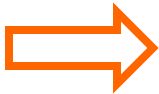
As explained in the information leaflet that you read prior to the amniocentesis, if you experience any significant or unusual symptoms following the test, such as

- feeling shivery, as if you have influenza
- lower abdominal pain
- fluid loss from the vagina
- bleeding from the vagina that is more than spotting
- uterine contractions (severe cramping pains)
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you should seek medical advice immediately

- Contact your Private Obstetric Consultant that referred you to the Women's Wellness Centre
- Contact your General Practitioner
- Contact your local hospital labour ward for advice
- Contact your local Hospital Accident and Emergency department for advice
- Contact the WWC between 09.00 and 19.00 Monday to Friday on 020 734 95200 for advice



Take a copy of the report with you and tell them that you have had an invasive test