

Amniocentesis:

what you need to know



Royal College of
Obstetricians and
Gynaecologists

Setting standards to improve women's health

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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.
- One of the main reasons amniocentesis is used is to detect whether or not a baby has a chromosomal disorder, such as Down syndrome.
- The safest time to have amniocentesis is after 15 weeks of pregnancy.
- About 1 in every 100 women (or 1%) who have amniocentesis from 15 weeks of pregnancy under ultrasound guidance miscarry as a result of the procedure.

- Amniocentesis is usually offered to women who have an increased chance of having a baby with a disorder. It is not usually offered on a routine basis.
- If a disorder is diagnosed, then you should be given information and the opportunity to discuss this with a specialist.
- With an experienced clinician, the risk of blood stained amniotic fluid is less than 1% (that is, less than 1 in 100 tests).
- You will be asked to sign a consent form before having amniocentesis.

About this information

This information is intended for you if you are pregnant and you have an increased chance of the baby having a disorder. You may have already been offered amniocentesis. The information is based on the Royal College of Obstetricians and Gynaecologists (RCOG) guideline [Amniocentesis and Chorionic Villus Sampling](#) (published in January 2005). To find out more about the guideline, see the [Sources and acknowledgements](#) section.

This information tells you about amniocentesis and what you need know if you are considering whether or not to have this procedure during pregnancy.

This information aims to help you and your healthcare team make the best decisions about your care. It is not meant to replace discussions with an obstetrician, midwife or genetic counsellor about your situation.

This information does **not** tell you about:

- amniocentesis in late pregnancy (after 24 weeks).
- amniocentesis and multiple pregnancy.
- chorion villus sampling (CVS) - see RCOG patient information, [CVS: What You Need to Know \(www.rcog.org.uk\)](#).

If you would like further information about these topics, then please ask your healthcare professional.

- Some of the recommendations here may not apply to you. This could be because of some other illness you have, your general health, or some or all of these things. If you think the treatment or care you get does not match what we describe here, talk about this with your obstetrician, midwife or genetic counsellor.

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 Edward syndrome or Down syndrome.

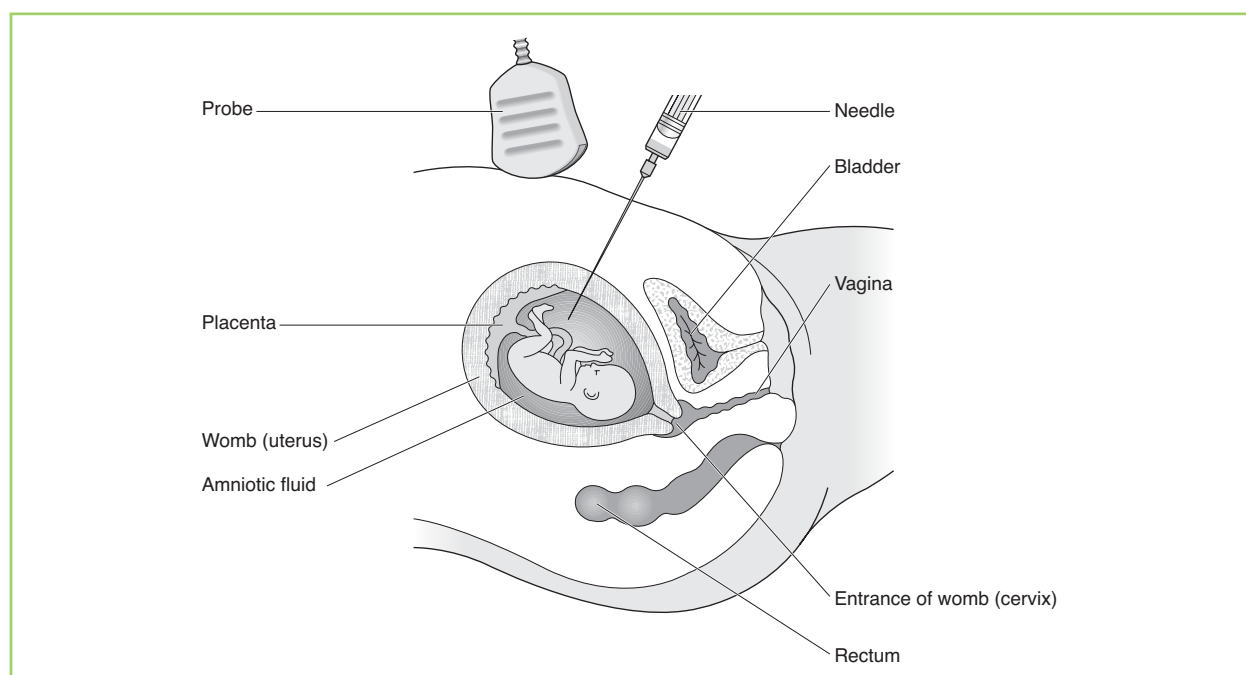
When and how is amniocentesis performed?

Amniocentesis is performed after 15 weeks.

You do not usually need a local anaesthetic. Before the procedure, gel is applied over your abdomen. You are scanned to check the positions of both the baby and the placenta (afterbirth). Your skin is then cleaned where the needle will be inserted.

Using an ultrasound probe for accurate guidance and to ensure a safe distance from the baby, a fine needle is pushed into your skin, through your abdomen and womb. Usually the person doing the procedure will avoid the placenta. Sometimes the needle will go through the placenta as this might be the only way to get the fluid. This is unlikely to cause you or your baby any harm.

A small sample of the fluid surrounding the baby is removed using a syringe. This fluid should be an amber/yellow colour but may be stained with blood. This is not harmful but may affect the accuracy of the result. The needle is then taken out and the baby is checked on ultrasound.



Amniocentesis

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?](#)).

For about 8 in every 100 women having amniocentesis, not enough fluid can be taken and the needle has to be re-inserted. This is usually due to the position of the baby. If a second attempt fails, you will be offered an appointment to have the procedure again on another day.

If your blood group is Rh (sometimes called 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), at www.nice.org.uk/pdf/Anti_d_patient_leaflet.pdf.

Is amniocentesis painful?

Most women say that having amniocentesis is uncomfortable rather than painful, a bit like a period pain. Women describe a sharp stinging feeling when the needle goes in and a feeling of pressure when the needle comes out. Women also say they feel anxious.

After the procedure you should rest, if you wish to, for the remainder of the day.

You may notice some 'spotting' of blood and cramping for a few hours afterwards. This is normal. If you experience any unusual symptoms immediately after the test, such as feeling shivery (as if you have flu), fluid loss, bleeding or contractions you should seek advice immediately.

What are the risks?

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 injury 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. Your healthcare professional will discuss the risk at your hospital.

There is a small risk, less than 1 in 1000 women who have amniocentesis, that the procedure will cause a serious infection. Infection can be caused by the needle puncturing the bowel, or skin contamination, but neither of these should happen if standard practices for amniocentesis are followed. Infection can also be caused if the

needle is contaminated by the ultrasound gel or the ultrasound probe. These risks can be minimised by standard procedures to reduce infection, for example by using sterile gel.

With an experienced clinician, the risk of blood stained amniotic fluid is less than 1% (that is, less than 1 in 100 tests).

Clinicians who do amniocentesis often, rather than occasionally, seem to be slightly better at getting enough fluid at the first insertion of the needle, and may also have a lower risk of miscarriage. The RCOG recommends that to maintain their skills, clinicians doing amniocentesis should do at least 10 each year.

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 missed screening for Down syndrome and they are over a certain age (typically over 35 years). This is because the risk of having a baby with Down syndrome increases as a woman gets older.
- 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. This may be found on a scan at 20 weeks.
- 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 types of laboratory test that can be used to look at the baby's chromosomes. These are:

- a full karyotype which checks all the baby's chromosomes. Results from this test are usually ready within 2-3 weeks.
- a rapid test which checks for specific chromosomes. The disorders that can be detected by the rapid test include Down syndrome (known as 'trisomy 21' caused by an additional chromosome 21), Edward syndrome (known as 'trisomy 18' caused by an additional chromosome 18) and Patau syndrome (known as 'trisomy 13' caused by an additional chromosome 13) and, if requested, sex

chromosome disorders. Results from this test are usually available after 3 working days. There may be a charge for this test.

The unit where you had the amniocentesis will make arrangements with you as to how you will receive your results.

How reliable are the laboratory tests?

The laboratory tests use different techniques to analyse the baby's chromosomes.

- With the full karyotype test there is a 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.
- Problems with the rapid test are that some samples may not be suitable for the test.
- 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 2 weeks later.

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.

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 that look at the baby's chromosomes 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 another disorder is detected by the test.
- A very few women will have a 'normal' result and yet, in spite of this, they will have a baby born with the disorder tested for, or another chromosomal or genetic condition. A normal 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 with you. 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.
- end this pregnancy.

If you decide to end the pregnancy, 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.

It is helpful to talk through all the options with your doctor or midwife before you make a decision about having amniocentesis. You should also be able to talk through your options with a paediatrician and consultant geneticist or genetic nurse counsellor.

Some women who made an informed decision to end their pregnancy find it helpful to talk with a counsellor afterwards about their experience.

Making a decision about amniocentesis

Making a decision about having a diagnostic procedure during pregnancy, such as amniocentesis, can be difficult. You may be making this decision alone or with your partner.

To help make the decision which is best for you, your healthcare professional should discuss the following information with you:

- the types of laboratory tests available and what the results will tell you
- the reliability of the laboratory test(s)
- the risk of having an uncertain result and being offered a repeat procedure
- the risk of miscarriage from amniocentesis, including the risk in your own unit at this time
- how long the results will take
- how you will get the results
- your options if the baby is found to have a disorder.

You may want to know more about the disorder or disorders which can be detected by amniocentesis. You may also want to know what is involved in ending a pregnancy and how you may feel afterwards.

In making a decision about amniocentesis, it is important you have enough time and that you feel supported in your final decision. You should be given time to talk through your options and be able to request any further information.

The final decision is yours. Only you can weigh up how much you want information about a disorder versus the slight risk that the procedure may lead to miscarrying a baby who may, or may not, have a disorder.

If you decide to have amniocentesis, then you will be asked to sign a consent form before the procedure is carried out.

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.
- Amniocentesis is occasionally performed to test for other disorders in pregnancy such as infection. Although the procedure is the same, it may be done later and the amniotic fluid is analysed slightly differently. The tests look for substances in the fluid itself, rather than at the baby's chromosomes.
- You have the right to be fully informed about your health care and to share in making decisions about it. Your healthcare team should respect and take your wishes into account.

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 at:

<http://www.rcog.org.uk/guidelines.asp?PageID=106&GuidelineID=2>.

Clinical guidelines are intended to improve patient care. They are drawn up by teams of medical professionals and consumers' representatives, who look at the best research evidence there is about care for a particular disorder or treatment. The guidelines make recommendations based on this evidence.

This information has been developed by the Patient Information Subgroup of the RCOG Guidelines and Audit Committee, with input from the Consumers' Forum and the authors of the clinical guideline. It is being reviewed until the end of August 2006 by women attending clinics in Bristol and London. All comments will be considered and the information may be amended. The final version is the responsibility of the Guidelines and Audit Committee of the RCOG.

Other organisations

ARC (Antenatal Results and Choices)

73 Charlotte Street

London W1T 4PN

Helpline: 0207 631 0285 (Monday to Friday, 10am to 5.30pm)

Email: info@arc-uk.org

Website: www.arc-uk.org

Contact a Family

209-211 City Road

London EC1V 1JN

Helpline: 0808 808 3555 (Monday to Friday, 10am to 4pm)

Email: info@cafamily.org.uk

Website: www.cafamily.org.uk

Down's Syndrome Association

Langdon Down Centre

2a Langdon Park

Teddington TW11 9PS

Helpline : 0845 230 0372 (Monday to Friday, 10am to 4pm)

Email: info@downs-syndrome.org.uk

Website: <http://www.downs-syndrome.org.uk>

Genetic Interest Group

Unit 4D, Leroy House
436 Essex Road
London N1 3QP
Tel: 0207 704 3141
Email: mail@gig.org.uk
Website: www.gig.org.uk/index.html

Sickle Cell Society

54 Station Road
London NW10 4UA
Tel: 0208 961 7795
Email: info@sicklecellsociety.org
Website: <http://www.sicklecellsociety.org>

SOFT UK (Patau's and Edward syndrome)

48 Froggatts Ride
Walmley
Sutton Coldfield B76 2TQ
Tel: 0121 351 3122
Email: enquiries@soft.org.uk
Website: <http://www.soft.org.uk>

The Miscarriage Association

c/o Clayton Hospital
Northgate
Wakefield
West Yorkshire WF1 3JS
Helpline: 01924 200799 (Monday to Friday, 9am to 4pm)
Email: info@miscarriageassociation.org.uk
Website: <http://www.miscarriageassociation.org.uk/main4.htm>

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