

Amniocentesis test – information for parents

This booklet gives you some information about the amniocentesis diagnostic test so you can decide whether to have the test.

1 Introduction

In this leaflet, you will find information about the amniocentesis test.

If your doctor or midwife offers you an amniocentesis test, it can be helpful to talk through the test with them before you decide whether to have the test or not. The information in this leaflet might help you make this decision.

Your doctor or midwife will give you information on the benefits and risks of the test before you make a decision.

It is important that you understand you have a choice of whether or not to have the test – you do not have to have it if you do not want it.

If a test result suggests that your baby has a chromosomal abnormality, you will be able to talk to a consultant paediatrician, consultant geneticist or genetic counsellor.

2 What is amniocentesis?

Amniocentesis is a test carried out during pregnancy which involves using a fine needle to remove a small amount of the amniotic fluid around your unborn baby.

It is used to detect chromosomal abnormalities such as Down's syndrome or conditions such as sickle-cell diseases. It can also detect certain other genetic conditions if your doctor or midwife has asked for you to be tested for these.

Amniocentesis is known as a diagnostic test because it gives you a diagnosis. For example, it tells you that your baby does or does not have Down's syndrome.

3 When should the test be carried out?

If you choose to have an amniocentesis, it will usually be carried out between your 15th and 18th week of pregnancy. However, the test can be carried out later in pregnancy.

4 Why am I offered the test?

Your doctor or midwife will offer you an amniocentesis if you have received a higher-risk result (this may also be called a higher-chance result) from a screening test for Down's syndrome.

You may also be offered a test if:

- your doctor thinks you may have a higher risk of passing on an inherited abnormality to your baby
- you have had a previous pregnancy where your baby was born with a physical, genetic or chromosomal abnormality; or
- people in your family have been born with abnormalities.

You will also be offered this test if your scan has shown that your baby has an increased chance of having a chromosomal or genetic disorder.

Not all women are offered this test.

5 How do I decide whether I want to have the test?

It can be difficult to make a decision about having an invasive test such as an amniocentesis (an invasive test is where we take a sample of cells or fluid from inside your body). You may find it helpful to talk to your midwife, doctor and your friends and family. You may also find it helpful to talk to some of the support groups that we have included at the end of this leaflet. You should remember that only you can decide whether or not you want to have this test.

6 Is there a risk to my pregnancy if I have the test?

Because an amniocentesis involves inserting a needle into the fluid around your baby, it may increase the risk of you having a miscarriage.

About one in every 100 women who have an amniocentesis will miscarry. There is a small risk, less than one in 1000, that the test will cause a serious infection. This is a rare complication.

We do not know why some women miscarry after having an amniocentesis. Research has shown that if a miscarriage does happen, it is usually because the amniotic sac (or bag of fluid) which surrounds the baby breaks, bleeds or becomes infected.

It is also hard to tell when you are most likely to miscarry after you have had an amniocentesis. Most miscarriages happen up to two weeks after the test. After three weeks, your chances of miscarrying become much lower.

7 Can I do anything to prevent a miscarriage after I have had an amniocentesis?

There is nothing you can do to prevent a miscarriage after an amniocentesis. Some hospitals will recommend that you rest for the day after the procedure and that you should avoid sex, any heavy lifting or strenuous exercise. However, there is no evidence to show that this helps. You do not need to rest in bed.

8 Other screening test results your doctor might need to know about

If your blood group is rhesus negative you will be offered an anti-D injection before this procedure. Anti-D prevents your body producing a reaction to your unborn baby if its blood group is different to yours.

Because amniocentesis is an invasive test, the hospital will need to know whether or not you have HIV or hepatitis B. If you've not been screened for HIV or hepatitis B yet, you'll be offered a blood test to find out whether or not you have HIV or hepatitis B.

It is important to know whether you have HIV or hepatitis B so that we can make sure the baby has the lowest chance of catching the disease as a result of the test.

9 Bringing a partner or friend

Most hospitals will recommend that you bring a partner, friend, or a family member when you have the test because you might be anxious. We suggest that you ask somebody to drive you to and from the hospital. You should not bring children along to the test because childcare facilities are not usually available in most hospitals.

10 Eating and drinking

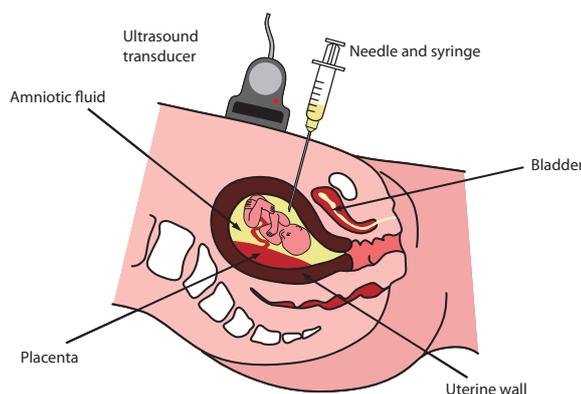
Most hospitals agree that it is safe to eat and drink as normal before and after the test. Your hospital will give you specific instructions.

11 How is the amniocentesis done?

Immediately before the test, your abdomen is cleaned to make sure that the test can take place in the most sterile conditions possible.

During the amniocentesis, a sonographer puts gel on your abdomen. You will then have an ultrasound scan to check the position of your baby. A fine needle is then inserted through your skin, through your abdomen and into your womb. The needle is used to remove a small sample of the amniotic fluid surrounding your baby.

You may need to have a full bladder when you come for the appointment. The doctor and midwife looking after you will let you know before you come. If you are not sure, you can contact them to ask.



Amniocentesis

12 Is an amniocentesis painful?

Most women say that having an amniocentesis is uncomfortable rather than painful. Some say it feels like period pain.

Women who have had an amniocentesis describe a sharp stinging feeling when the needle goes in and a feeling of pressure when the needle comes out. They also say they feel anxious before and after the test. You may notice some cramping for a few hours afterwards. This is normal.

You can take paracetamol for any discomfort (remember, you can only take a maximum of eight tablets in 24 hours).

If you experience any unusual symptoms immediately after the test or over the next few days (for example, if you have been feeling shivery (as if you have flu), have lost any of the fluid that surrounds your unborn baby (called amniotic fluid), have been bleeding or have contractions), you should call your midwife or doctor, and the hospital where you had the test, straight away.

13 What happens after the amniocentesis?

After the test, the hospital will send the sample of amniotic fluid to a laboratory for testing. The sample contains some of your baby's cells (cells contain a person's genetic information).

Two types of laboratory test are used to look at your baby's chromosomes – 'PCR' (polymerase chain reaction) and a 'full karyotype'.

PCR checks for the three most common chromosomal abnormalities. These are Down's syndrome (trisomy 21, caused by an extra chromosome 21), Edwards' syndrome (trisomy 18, caused by an extra chromosome 18) and Patau's syndrome (trisomy 13, caused by an extra chromosome 13).

Your doctor may also ask the laboratory to check for sex chromosome abnormalities (abnormalities affecting the X and Y chromosomes). They will discuss this with you in detail when you have your amniocentesis test.

The results from PCR laboratory tests are usually ready after three working days.

A full karyotype checks all your baby's chromosomes. Results from this test are usually ready within two to three weeks.

Your doctor or midwife will usually only ask for a PCR test. They will only request a full karyotype if there is a family history of chromosomal abnormalities or if your scan has shown that your baby has an increased chance of having a chromosomal or genetic disorder.

14 What about the results?

Before you leave the hospital, someone will ask you how you would like to receive the results of the test. You can usually choose whether to receive the results by phone or come into the hospital again and receive your results face-to-face. In some cases, a community midwife may be able to come to your home to give you the results. They will also give you written confirmation of your results.

For most women, the laboratory test will give a definite 'yes' or 'no' answer. These results will let you know whether your baby has the chromosomal abnormality the test was looking for.

Most women who have an amniocentesis will have a 'normal' result (in other words, their baby won't have the chromosomal abnormality the test was looking for).

Occasionally, either PCR or a full karyotype does not give a clear result. This is rare but if it happens, your doctor or midwife may offer you another amniocentesis.

Some women will be told their baby has the chromosomal abnormality the test was looking for. If your baby is diagnosed with a chromosomal abnormality, your doctor or midwife will give you information on the condition your baby has and they will give you the opportunity to discuss this with a specialist.

Very occasionally, women who have an amniocentesis to detect Down's syndrome find out their baby has a different chromosomal abnormality. If this happens to you, your doctor or midwife will give you information on the condition your baby has and they will give you the opportunity to discuss this with a specialist.

A normal amniocentesis result does not guarantee your baby will not have any abnormalities. Not all abnormalities can be detected by the amniocentesis test.

15 What if there is something wrong with my baby?

If the results show that the baby has a chromosomal abnormality, your doctor or midwife will talk to you in detail about this.

When you are deciding what to do, you need to consider what is best for you and your family.

These decisions are often very difficult to make and you might want to talk about your feelings with a midwife, doctor or a support organisation for parents such as Antenatal Results and Choices (ARC). ARC's details are at the back of this leaflet.

You might choose to:

- continue with your pregnancy and use the information you have gained from the test results to help prepare for the birth and care of your baby; or
- end the pregnancy (have an abortion).

If you decide to continue with your pregnancy, you can talk to your doctor or midwife and contact support organisations about how you can learn more about your baby's condition and how best to care for him or her.

If you decide to end your pregnancy, you will be given information about what this involves. The type of abortion you will be able to have will depend on how many weeks pregnant you are when you make the decision to end your pregnancy.

It can be helpful to think about all of your possible options and talk these through with your doctor or midwife before you decide whether to have an amniocentesis. If an amniocentesis result tells you that your baby has an abnormality, you will also be able to talk to a consultant paediatrician, consultant geneticist or genetic counsellor.

At the end of this leaflet, you will also find details of many support groups who will be able to talk things through with you. Their websites often have useful information about the various conditions and stories of women who have had to make some of the same choices as you.

16 Where can I get more information about amniocentesis tests, the different conditions they might detect and the choices I have?

The following organisations can offer you support.

Antenatal Results and Choices (ARC)
73 Charlotte Street
London
W1T 4PN

Phone: 0207 631 0285
Email: info@arc-uk.org
Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides impartial information and individual support to parents going through antenatal screening or whose unborn baby has been diagnosed with an abnormality.

Contact a Family (CAFAMILY)
209 to 211 City Road
London
EC1V 1JN

Phone: 0808 808 3555
Email: info@cafamily.org.uk
Website: www.cafamily.org.uk

Contact a Family is a charity which provides support, advice and information for families with disabled children, no matter what their condition or disability.

DIPEX
PO Box 428
Witney
Oxon
OX28 9EU

Email: info@healthtalkonline.org
Website: www.healthtalkonline.org &
www.youthhealthtalk.org

DIPEX is a database of research articles based on the experiences of people who have had over 40 different illnesses and health conditions. The results of their research are published on two websites which are aimed at parents and their carers, family and friends, doctors, nurses and other health professionals. Their target is to have research available of at least 100 conditions within the next five to 10 years.

Down's Syndrome Association (DSA)
Langdon Down Centre
2a Langdon Park
Teddington
TW11 9PS

Phone: 0845 230 0372
Email: info@downs-syndrome.org.uk
Website: www.downs-syndrome.org.uk

The Down's Syndrome Association (DSA) aim to help people with Down's syndrome lead full and rewarding lives.

Genetic Interest Group (GIG)
Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Phone: 0207 704 3141
Email: mail@gig.org.uk
Website: www.gig.org.uk

The Genetic Interest Group (GIG) is a national network of patient organisations and charities which support children, families and people affected by genetic disorders.

The Miscarriage Association
c/o Clayton Hospital
Northgate
Wakefield
West Yorkshire
WF1 3JS

Phone: 01924 200799
Email: info@miscarriageassociation.org.uk
Website: www.miscarriageassociation.org.uk

Miscarriage can be a very distressing, frightening and lonely experience. The Miscarriage Association provides support and information for women experiencing miscarriage and their families.

Stillbirth and Neonatal Death Society (SANDS)
28 Portland Place
London
W1B 1LY

Phone: 0207 436 5881
Email: helpline@uk-sands.org
Website: www.uk-sands.org

SANDS support anyone affected by the death of a baby and promotes research to reduce the number of stillbirth and neonatal deaths.

Sickle Cell Society
54 Station Road
London
NW10 4UA

Phone: 0208 961 7795
Email: info@sicklecellsociety.org
Website: www.sicklecellsociety.org

The Sickle Cell Society provides information, counselling and care for people with sickle-cell disorders and their families.

S.O.F.T UK
48 Froggatts Ride, Walmley
Sutton Coldfield
West Midlands
B76 2TQ

Phone: 0121 351 3122
Email: enquiries@soft.org.uk
Website: www.soft.org.uk

S.O.F.T UK provides support for families affected by Patau's syndrome (trisomy 13), Edwards' syndrome (trisomy 18), partial trisomy, mosaicism, rings, translocation, deletion, and related disorders.

References

- 1 Royal College of Obstetricians and Gynaecologists. Amniocentesis and Chorionic Villus Sampling. London: RCOG, 2005
- 2 Antenatal Screening Wales and NHS Fetal Anomaly Screening Programme. Amniocentesis and Chorionic Villus Sampling: Policy, Standards and Protocols. April 2008
- 3 RCOG (2006) Amniocentesis; what you need to know. Available from www.rcog.org.uk
- 4 RCOG (2006) Chorionic Villus sampling (CVS): what you need to know. Available from www.rcog.org.uk
- 5 HIV Medicine (2008), chapter 9, pages 452 to 502, British HIV Association Guidelines, British HIV Association and Children's HIV Association guidelines for the management of HIV infection in pregnant women 2008.

This information has been produced on behalf of the NHS Fetal Anomaly Screening Programme and is based on text originally developed by Antenatal Screening Wales.

All of our publications can be found online at www.fetalanomaly.screening.nhs.uk

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If you have any comments on this booklet or questions about our screening programme, please contact us using the details below.

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