

Amniocentesis test

Information for parents



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.

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 or genetic abnormality, you will be able to talk to a consultant paediatrician, consultant geneticist or genetic counsellor.

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.

Amniocentesis is used to detect chromosomal abnormalities such as Down's syndrome. It can also detect certain other genetic conditions, such as sickle cell disease or thalassaemia major, 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.

When should the test be carried out?

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

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 or your baby's father may have a higher risk of passing on an inherited abnormality to your baby, for example cystic fibrosis, sickle cell disease or thalassaemia major;
- you have had a previous pregnancy where your baby was born with a genetic or chromosomal abnormality; or
- you 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.

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.

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

Because an amniocentesis is an invasive test it may increase the risk of you having a miscarriage.

The overall risk of you having a miscarriage after amniocentesis is about 1%. In other words, 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, 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. Some problems, such as miscarriages, that happen after an amniocentesis will happen anyway, whether the test is done or not, especially when they happen more than three weeks after the procedure.

Can I do anything to prevent a miscarriage after I have had 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.

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 after this procedure. Anti-D prevents your body producing a reaction to your unborn baby's blood if its blood group is different to yours. If your blood group is not rhesus negative you won't need this injection. Your doctor will discuss this with you.

Because amniocentesis is an invasive test, the hospital will want 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 try to keep the chance of the baby catching the disease during the amniocentesis test as low as possible.

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. You should not bring children along to the test because there are often no childcare facilities in hospitals.

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.

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 you will 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. Because the needle is so fine, local anaesthetic is not usually used.

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.

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 are worried about taking painkillers or have any questions, you should talk to your doctor or midwife.

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, or the hospital where you had the test (or both) straight away.

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 often only ask for a PCR test. They will ask for a full karyotype if there is a history of chromosomal abnormalities or if your scan has shown that your baby has an increased chance of having a chromosomal or genetic disorder. As the full karyotype test is more detailed, it can sometimes take more time to get the results.

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 test.

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.

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
- continue the pregnancy and consider adoption; or
- end the pregnancy (have a termination)

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 procedure 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 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.

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

The following organisations can offer you support.

Antenatal Results and Choices (ARC)

Website: www.arc-uk.org

73 Charlotte Street

London

W1T 4PN

Helpline: 0207 631 0285

Email: info@arc-uk.org

Antenatal Results and Choices (ARC) provides information and support to parents before, during and after antenatal screening and diagnostic tests, especially those making difficult decisions about testing, or about continuing or ending a pregnancy after a diagnosis. ARC offers continuing support whatever decisions you make.

Down's Syndrome Association (DSA)

Website: www.downs-syndrome.org.uk

Langdon Down Centre

2a Langdon Park

Teddington

TW11 9PS

Phone: 0845 230 0372

Email: info@downs-syndrome.org.uk

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

Healthtalkonline

Website: www.healthtalkonline.org

PO Box 428

Witney

Oxfordshire

OX28 9EU

Email: info@healthtalkonline.org

The Health Experience Research Group has created a unique database of personal and patient experiences through in-depth research into over 60 different illnesses and health conditions. The results of their research are published on two websites – www.healthtalkonline.org and www.youthhealthtalk.org which are aimed at patients, their carers, family and friends, doctors, nurses and other health professionals. Their target is to complete at least 100 conditions within the next 5 to 10 years.

They have also recently started two social networking sites for people to add their own experiences of health and illness at www.myhealthtalk.org and www.myyouthhealthtalk.org

The websites are run by the DIPEX charity.

Sickle Cell Society

Website: www.sicklecellsociety.org

54 Station Road

London

NW10 4UA

Phone: 0208 961 7795

Email: info@sicklecellsociety.org

The Sickle Cell Society provides information, counselling and care for people with sickle cell disease or thalassaemia major and their families.

The Miscarriage Association

Website: www.miscarriageassociation.org.uk

17 Wentworth Terrace
Wakefield
WF1 3QW

Phone: 01924 200799

Email: info@miscarriageassociation.org.uk

The Miscarriage Association was founded in 1982 by a group of people who had experienced miscarriage and they continue to offer support and information to anyone affected by the loss of a baby in pregnancy, to raise awareness and to promote good practice in medical care.

UK Thalassaemia Society

Website: www.ukts.org

19 The Broadway
Southgate Circus
London
N14 6PH

Phone: 020 8882 0011

Email: office@kuts.org

The UK Thalassaemia Society improves the health education of the communities of the UK that are at risk of thalassaemia.

References

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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

To order copies of this leaflet please email:
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