Amniocentesis

Introduction

Amniocentesis is a diagnostic test carried out during pregnancy.

It can assess whether the unborn baby (foetus) could develop, or has developed, an abnormality or serious health condition.

Things that increase the risk of an abnormality include:

- the mother's age
- the mother's medical history
- a family history of inherited genetic conditions

Why and when amniocentesis is used

Amniocentesis can be used to detect a number of conditions, such as:

- Down's syndrome – a genetic condition that affects a person's physical appearance and mental development
- spina bifida – a series of birth defects that affect the development of the spine and nervous system
- sickle cell anaemia – a genetic disorder that causes a person's red blood cells to develop abnormally

Read more about why amniocentesis is used.

Amniocentesis is usually carried out during weeks 15-20 of the pregnancy.

The procedure can be performed earlier than 15 weeks, but this is avoided if possible because it may increase the risk of causing complications or a miscarriage (loss of the pregnancy).

Read more about when amniocentesis is used.

What happens during amniocentesis

Before you have amniocentesis, a healthcare professional will explain the procedure to you, including why they think it's necessary and the benefits and risks.

They'll also tell you about any alternative tests that may be appropriate, such as chorionic villus sampling (CVS).

If you decide to have amniocentesis, you'll usually be asked to sign a consent form.

During the procedure, a needle will be used to extract a sample of amniotic fluid, the fluid that surrounds the foetus in the womb (uterus).

Amniotic fluid contains cells shed from the foetus that can be examined and tested for a number of conditions.

Read more about how amniocentesis is performed.

Possible complications

Diagnostic tests, such as amniocentesis, are usually only offered to women when there's a significant risk their baby will develop a serious condition or abnormality.

This is because the procedure is invasive (involves going into the body) and has a small associated risk of miscarriage, estimated to be about 1 in 100.

A bacterial infection is another, but rare, possible complication of amniocentesis. The risk of developing a serious infection from amniocentesis is estimated to be less than 1 in 1,000.

The symptoms of an infection include:

- a high temperature (fever) of 38ºC (100.4ºF) or above
- tenderness of your abdomen (tummy)
- contractions (when your abdomen tightens then relaxes)

Seek immediate medical attention if you've recently had amniocentesis and you experience any of these symptoms.

Read more about the possible complications of amniocentesis.

Your results

After you've had amniocentesis, the amniotic fluid sample taken during the procedure will be tested in a laboratory.

Most women's test results will be negative and their baby won't have any of the disorders that were tested for.

A positive test result means your baby has a disorder that was tested for. The implications of this will be fully discussed with you.

Read more about the results of amniocentesis.

Why you may need amniocentesis

Amniocentesis is a diagnostic test to detect a serious or potentially serious disorder in an unborn baby (foetus).

Amniotic fluid

Amniotic fluid surrounds the foetus in the womb. It contains cells shed from the skin of the developing baby and the baby's waste products.

Every cell in the amniotic fluid contains a complete set of the baby's DNA. This means that cell samples obtained during amniocentesis can be used to assess the health of the developing baby and diagnose potential problems.

Amniocentesis provides healthcare professionals with important information about the likelihood of a baby developing one or more inherited genetic conditions, or conditions that develop during the pregnancy.

If a serious abnormality is detected, amniocentesis enables parents to choose whether to continue with the pregnancy or terminate it at an early stage.

Conditions

Amniocentesis can be used to help diagnose many different conditions. Some are described below.

Chromosomal conditions

Chromosomal conditions affect the chromosomes (the threadlike structures inside cells that carry genetic instructions in the form of genes). For example:

- **Down's syndrome** – a syndrome that affects physical appearance, mental development and learning ability; it's caused by an extra chromosome 21, giving it it's other name, trisomy 21
- **Edward's syndrome** – a syndrome that causes severe physical and mental abnormalities; it's caused by an extra chromosome known as trisomy 18
- **Patau's syndrome** – a rare but serious syndrome where babies usually don't survive for more than a few days; it's caused by an extra chromosome known as trisomy 13

Blood disorders

Amniocentesis can also be used to check for inherited blood disorders such as:

- sickle cell anaemia – an inherited genetic condition that causes a person's red blood cells to develop abnormally
- thalassaemia – a group of inherited blood disorders where the part of the blood known as haemoglobin is abnormal; the abnormality affects the red blood cells' ability to function normally
- haemophilia – a condition that affects the blood's ability to clot

Neural tube defects

Amniocentesis can test for neural tube defects.

The neural tube is a collection of cells that form very early in the development of an embryo. The top of the neural tube eventually forms the brain and the remainder becomes the spinal cord.

If the neural tube isn't completely closed, it can cause a defect to develop, such as a hole in the spinal column. These defects often occur within the first month of pregnancy.

The most common neural tube defect is spina bifida, which can cause learning difficulties and paralysis (weakness) of the lower limbs.

Musculoskeletal disorders

Amniocentesis can also be used to diagnose conditions that affect the musculoskeletal system (your bones and muscles), such as muscular dystrophy.

Muscular dystrophy is an inherited condition that causes muscles to gradually weaken, resulting in an increasing level of disability.

Other genetic conditions

As well as helping identify chromosomal conditions, blood disorders, neural tube defects and musculoskeletal disorders, amniocentesis can also help diagnose a number of genetic conditions, such as Marfan syndrome. Marfan syndrome affects the tissues that provide support and structure in the body.

If there's a risk of an inherited condition being passed to your baby – for example, if there's a family history of the condition – your GP, midwife or genetic counsellor will explain the risk to you and your partner.

Genetic counsellors are healthcare professionals who help people understand and deal with genetic conditions.

Read more about genetic testing and counselling.

When amniocentesis is carried out

Amniocentesis is usually carried out during weeks 15 to 20 of the pregnancy.

The procedure can be performed earlier than 15 weeks, but this is avoided as it may increase the risk of a miscarriage (loss of the pregnancy) or club foot in the foetus.

See complications of amniocentesis for more information about the risks of amniocentesis.

Occasionally, amniocentesis is carried out later in the pregnancy to test for an infection. For example, in rare cases it may be used to test a pregnant woman who has been exposed to the parvovirus B19, which causes slapped cheek syndrome (a childhood infection characterised by a bright red rash that develops on the cheeks).

Amniocentesis may also be recommended when there's a high risk of the unborn baby having a serious inherited condition.

This could be because:

- **you've had a previous pregnancy with a foetal problem** – for example, a baby born with a chromosome abnormality
- **you have a family history of a condition** – for example, muscular dystrophy (an inherited condition that causes increasing muscle weakness)
- **you're over 35 years of age** – the risk of your child developing Down's syndrome increases with age
- **an earlier antenatal screening test has suggested there may be a problem** – for example, a high level

of alpha-fetoprotein (a protein found in the blood) may indicate neural tube defects (birth defects that affect the spine and nervous system), such as spina bifida

Amniocentesis or CVS?

Another diagnostic test known as chorionic villus sampling (CVS) can be carried out slightly earlier than amniocentesis (from about week 10 of the pregnancy).

CVS tests a sample of cells taken from the placenta (the organ that links the mother's blood supply with her unborn baby's).

With CVS, the risk of miscarriage is about 1-2%, which is slightly higher than the risk of miscarriage for amniocentesis. However, the test can be carried out earlier, so you'll have more time to consider the results.

Genetic counselling

If there's a risk you could pass a genetic condition on to your child, your GP or midwife can discuss appropriate tests with you and explain why they might be necessary.

In some cases, you may be referred to a genetic counsellor. Genetic counsellors are healthcare professionals who help people understand and deal with genetic conditions.

They'll discuss the likelihood of you passing certain genetic conditions on to your children. They'll also be able to give you advice about what to do when you get the results.

Find out more about genetic testing and counselling, or read What is genetic counselling and do I need a genetic counsellor? for more information.

How amniocentesis is performed

Amniocentesis involves removing a small sample of amniotic fluid from the womb so the cells it contains can be tested.

Amniotic fluid surrounds the foetus (unborn baby) in the womb (uterus). The fluid contains cells shed by the foetus that can be analysed for information about the health of your baby.

Before you have amniocentesis, a healthcare professional will explain the procedure to you, including why they

think it's necessary and the benefits and risks.

They'll also tell you about alternative tests that may be appropriate. If you decide to have amniocentesis, you'll usually be asked to sign a consent form.

Read about the possible complications of amniocentesis for more information about the risks.

Ultrasound scan

You'll have an ultrasound scan before and during amniocentesis. An ultrasound scan uses high-frequency sound waves to produce an image of your womb that's relayed to a monitor.

The ultrasound scan allows healthcare professionals to:

- check the position of the foetus
- find the best place to remove some amniotic fluid
- ensure the needle can pass safely through the walls of your abdomen (belly) and womb

Anaesthetic

Before the needle is inserted into your abdomen, the area may be numbed with anaesthetic. This involves having a small injection into your belly that may sting slightly.

However, anaesthetic isn't usually necessary because research suggests that it doesn't have much effect in most cases.

The procedure

An antiseptic solution will be used to clean your abdomen to minimise infection. A long, thin needle will be inserted through your abdominal wall. It may cause a sharp, stinging sensation.

Using the ultrasound image as a guide, the needle will be passed into the amniotic sac that surrounds the foetus. A syringe will then be used to remove a small sample of the amniotic fluid, which will be sent to a laboratory for analysis.

In about 8 out of every 100 women who have amniocentesis, not enough fluid is removed the first time the needle is inserted. If this happens, the needle will be inserted again.

Amniocentesis usually takes around 10 minutes. However, it can take slightly longer if the position or movement of the foetus makes it difficult to take a sample.

Amniocentesis isn't usually painful, but you may feel uncomfortable during the procedure. Some women describe experiencing a pain similar to period pain or feeling pressure when the needle is taken out.

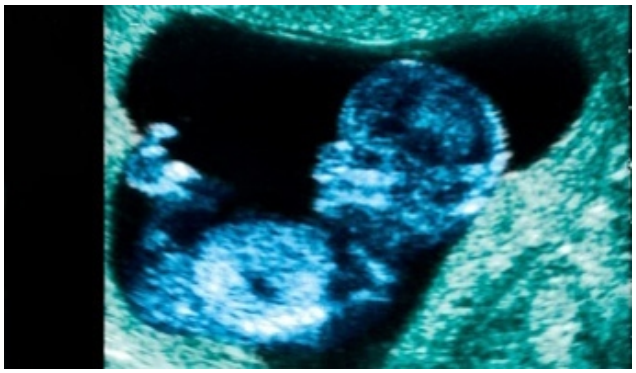
Recovery

After having amniocentesis, you'll usually be advised to rest for 24 hours. You may have cramps (similar to menstrual cramps) for a few hours and "spotting" (drops of blood from your cervix, located at the neck of the womb).

You should seek urgent medical attention if you:

- feel shivery
- have a high temperature (fever) of 38ºC (100.4ºF) or above
- are bleeding from your vagina
- have persistent lower back pain or abdominal pain
- have clear watery fluid coming from your vagina (that's not urine)
- have contractions (when your abdomen tightens then relaxes)

An ultrasound scan is carried out during amniocentesis to check the position of the foetus and locate the best place to remove the amniotic fluid.



Complications of amniocentesis

Amniocentesis is a fairly common and safe procedure.

For most women, the benefits of having amniocentesis – in terms of diagnosing problems with the developing baby – significantly outweigh the risks.

However, you should be aware of the possible complications during or after amniocentesis so you can make an informed decision. These are outlined below.

Injury from the needle

During amniocentesis, the placenta (the organ that links the mother's blood supply to her unborn baby's) may be punctured by the needle.

It's sometimes necessary for the needle to enter the placenta to access the amniotic fluid. If this happens, the puncture wound usually heals without any more problems developing.

An ultrasound scanner is now commonly used to guide the needle, significantly reducing the risk of injury.

Infection

In very rare cases, an infection may develop if the procedure introduces bacteria into your amniotic sac (the sac surrounding the foetus, which contains amniotic fluid). This can cause:

- a high temperature (fever) of 38ºC (100.4ºF) or above
- tenderness of your abdomen (tummy)
- contractions (when your abdomen tightens then relaxes)

You should seek immediate medical attention if you have any of these symptoms. The risk of developing a serious infection from amniocentesis is estimated to be less than 1 in 1,000.

Rhesus disease

Rhesus disease is a condition where proteins in a pregnant woman's blood attack her baby's blood cells.

Rhesus disease is only possible if your blood is rhesus negative and your baby's blood is rhesus positive. If this is the case, amniocentesis could trigger rhesus disease if your blood is exposed to your baby's blood during the procedure.

In this situation, you'll be given an injection of anti-D immunoglobulin after having amniocentesis. Immunoglobulin is a solution of antibodies (proteins produced by the body to fight disease-carrying organisms) that can prevent rhesus disease from developing.

Club foot

Having amniocentesis early (before week 15 of the pregnancy) has been associated with an increased risk of the unborn baby developing club foot.

Club foot, also known as talipes, is a deformity of the ankle and foot that's present at birth (congenital).

Because of the increased risk of a baby developing club foot, amniocentesis isn't recommended before 15 weeks of pregnancy.

Miscarriage

There's a small risk of miscarriage (loss of the pregnancy) occurring in any pregnancy, regardless of whether or not you have amniocentesis.

If you have amniocentesis after 15 weeks of pregnancy, the chance of having a miscarriage is estimated to be about 1 in 100. The risk is higher if the procedure is carried out before 15 weeks.

It's not known for certain why amniocentesis can lead to a miscarriage. However, it may be caused by factors such as infection, bleeding or damage to the amniotic sac that surrounds the baby.

Most miscarriages that happen after amniocentesis occur within the first 72 hours of the procedure. However, in some cases a miscarriage can occur later than this (up to two weeks afterwards).

Inconclusive results

After having amniocentesis, it can be reassuring if the results indicate that your developing baby has normal chromosomes and that there are no signs of any developmental problems.

Chromosomes are the threadlike structures inside cells that carry genetic information in the form of genes.

However, you should be aware that amniocentesis can't test for every condition or disease, and it can't guarantee your baby will be born completely healthy.

Read more about the results of amniocentesis.

Results of amniocentesis

After you've had the amniocentesis procedure, the sample of amniotic fluid that surrounds the unborn baby in the womb will be taken to a laboratory for testing.

There are two different types of tests – a rapid test and a full karotype. These are described below.

Rapid test

A rapid test looks for abnormalities on specific chromosomes (the threadlike structures that carry genetic information in the form of genes).

A rapid test can identify a number of chromosomal conditions that cause physical and mental abnormalities. These are:

- **Down's syndrome** – caused by an extra chromosome 21
- **Edward's syndrome** – caused by an extra chromosome 18
- **Patau's syndrome** – caused by an extra chromosome 13

The results of a rapid test should be ready after three working days. The test is almost 100% accurate, but it only tests for the three syndromes listed above.

Full karyotype

Each cell in the body contains 23 pairs of chromosomes. A full karotype checks all of these.

The cells in the sample of amniotic fluid are grown for up to 10 days in a laboratory. They are then examined under a microscope to check the number of chromosomes and their appearance.

Results from a full karotype will usually be ready in two or three weeks. In about 1 in every 100 tests, the results may not be clear. This could be because the mother's blood has contaminated the sample of amniotic fluid, which may have prevented the cells from growing properly.

Negative test results

In most cases, the results of amniocentesis are negative. This means that the baby doesn't have any of the disorders tested for.

However, it's possible to have a negative result from amniocentesis, but your baby may still be born with the condition tested for, or they may be born with another chromosomal condition. This is because a normal test result doesn't exclude every chromosomal disorder.

Genetic disorders are caused by mutations (changes) in the genes (units of genetic material). Each chromosome contains thousands of genes. This means it's not possible to test for every possible genetic mutation, so a baby may occasionally be born with a condition that wasn't detected.

Positive test results

A positive test result means your baby has the disorder that was tested for. The implications of a positive test result will be fully discussed with you.

You should be aware that there's no cure for the majority of chromosomal conditions, so you need to consider your options carefully. These may include:

- continuing with your pregnancy while getting information and advice about the condition so you're prepared for caring for your baby
- ending your pregnancy

Talk to your GP or midwife if you're considering ending your pregnancy. They'll be able to give you important information and advice.

For example, your options for ending your pregnancy will depend on how many weeks pregnant you are when you make the decision.

If you decide to end your pregnancy, you may wish to talk to a counsellor afterwards. Your GP or midwife will be able to arrange this for you.

Read more about genetic testing and counselling.

Additional information

Useful organisations

The Royal College of Obstetricians and Gynaecologists

27 Sussex Place, Regent's Park, London, NW1 4RG
Tel : 020 7772 6200

http://www.rcog.org.uk/

NCT

Alexandra House, Oldham Terrace, London, W3 6NH
Tel : 0300 330 0772

http://www.nct.org.uk/home