West Hertfordshire Teaching Hospitals NHS Trust

A guide to...

Amniocentesis

Patient information

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Amniocentesis is a procedure that is carried out during pregnancy. The most common reason for a pregnant woman to be offered amniocentesis is to see if her developing baby has a chromosome disorder such as Down's syndrome.

Amniocentesis is usually offered after 15 completed weeks of pregnancy (usually between 15-16 weeks). This is because at this point there is enough fluid around the baby to make it possible to extract a little without great risk to the pregnancy. Nevertheless, there is a small risk of complications with amniocentesis, including miscarriage.

A note about DNA, genes and chromosomes

In most cells of your body you have 46 chromosomes arranged in 23 pairs. Twenty-two of the pairs are matching pairs. The twenty-third pair are the sex chromosomes, which match exactly in women (who have two X chromosomes) but not in men (who have one X and one Y). One chromosome from each pair comes from your mother and one from your father. Chromosomes are made of DNA, which stands for 'deoxyribonucleic acid'. This is your genetic material. It is found in the nucleus of every cell in your body.

Each of your 46 chromosomes carries hundreds of genes. A gene is the basic unit of your genetic material. It is made up of a piece (a sequence) of DNA and sits at a particular place on a chromosome. So, a gene is a small section of a chromosome. A gene is effectively a coded set of instructions to the cells. Each gene has a particular function in your body. For example, a gene may be involved in dictating your eye colour or determining your height. Each gene has a matching 'paired' gene on the paired chromosome. So, as for chromosomes, one gene from each pair is inherited from your mother, the other from your father. Humans have between 20,000 and 25,000 genes.

What is amniocentesis?

Amniocentesis is a procedure that is carried out during pregnancy. It involves taking a sample of the fluid inside your womb (uterus) that surrounds your developing baby. This fluid is called amniotic fluid and it acts as a cushion or protection for your baby. However, the fluid contains some of the baby's cells, which are shed from the baby's skin. These cells can be examined to look at the baby's chromosomes.

The sample of amniotic fluid is taken using a fine needle. This involves putting the needle into your womb (but not into the baby). An ultrasound scan is being done the whole time so that the operator can see exactly where the needle is. Tests are done on the fluid in the laboratory to look at the chromosomes or the genes. The purpose is usually to try to detect these chromosomal or genetic conditions early in the pregnancy. This allows you the chance to consider whether you wish to continue the pregnancy and to be prepared.

Amniocentesis is a diagnostic test. In most cases it tells you for certain whether or not your baby has a certain condition. Compare this with a screening test in pregnancy (for example, blood tests and/or ultrasound screening tests for Down's syndrome). Screening tests give you a risk estimate (that is, they tell you whether it is likely or unlikely) that the baby has a certain condition. They do not give you a definite 'yes' or 'no' answer. If you have a screening test that shows a high risk of a certain condition, you will usually be offered a diagnostic test.

An alternative diagnostic test to amniocentesis is chorionic villus sampling (CVS). CVS involves taking a very small sample of tissue from the afterbirth (placenta) that is attached to your womb. The cells of the afterbirth contain the same genetic material as the cells of your developing baby.

CVS is usually carried out earlier than amniocentesis - between the beginning of the eleventh and the end of the thirteenth week of pregnancy. This is an advantage over amniocentesis as it means that decisions about what you would like to happen with the pregnancy can be made sooner. However, there is a slightly higher risk of complications such as miscarriage with CVS. See separate leaflet called Chorionic Villus Sampling for more details.

Why pregnant women are offered amniocentesis

Amniocentesis is not carried out as a routine test in pregnancy. It is offered if you are thought to have an increased chance of having a baby with a certain condition. Amniocentesis is therefore often offered to pregnant women whose screening tests show a higher-than-average likelihood of a genetic condition. It is also offered to pregnant women who are already known to have an increased likelihood of a genetic condition.

You do not have to have an amniocentesis if you are offered one. It is a choice that you can make. You should discuss the test fully with your doctor, including the potential risks or complications, before you decide whether or not to go ahead.

The most common reason for a pregnant woman to be offered amniocentesis is to see if their developing baby has a chromosomal disorder such as Down's syndrome. Someone with Down's syndrome has an extra copy of chromosome number 21 in the cells of their body. See separate leaflet called Down's Syndrome for more details.

A screening test for Down's syndrome is offered to all pregnant women in the UK. If this screening test shows a higher-risk result, you may be offered amniocentesis.

Other reasons why amniocentesis may be offered during pregnancy include:

If you have had a baby with a chromosomal, genetic condition or other disorder (for example, a neural tube defect) in a previous pregnancy.

If you have, or your partner has, a genetic disorder, or are carriers for a genetic disorder that could be passed on to the baby. Examples include sickle cell anaemia, thalassaemia, cystic fibrosis and Duchenne muscular dystrophy.

- If there is a history of certain genetic conditions in your family.
- If other tests during pregnancy (for example, scans) have raised the possibility that the baby has a chromosomal disorder such as Down's syndrome.
- If the baby has an increased risk of a chromosomal disorder because of your age.

In the later stages of pregnancy, amniocentesis may also be offered for a number of different reasons. These include:

- To look for any signs of infection if your membranes have ruptured early. Here, the amniotic fluid is tested for germs in the fluid itself. It is not the baby's chromosomes that are being tested.
- To assess the maturity of the lungs of the developing baby.
- To look at the baby's chromosomes if a chromosomal problem is suspected later in the pregnancy.
- It is thought that about five in every 100 pregnant women are offered a prenatal diagnostic test such as amniocentesis or CVS. Amniocentesis is the most common prenatal diagnostic test that is offered to pregnant women.

At what stage of pregnancy is amniocentesis offered?

Amniocentesis is usually offered after 15 completed weeks of pregnancy (most commonly between 15-16 weeks). This is because it has been shown to be safest at this stage of pregnancy.

Amniocentesis before 15 weeks is considered as early amniocentesis. Before 15 weeks of pregnancy, the amount of amniotic fluid levels is lower. This makes it more difficult to obtain enough amniotic fluid for testing. Early amniocentesis is not usually recommended because it carries a higher risk of miscarriage. There is also a higher risk of club foot in the developing baby. (Club foot is a deformity of one or both feet where the foot is twisted so that the sole of the foot cannot be placed flat on the ground.)

As mentioned, sometimes amniocentesis is carried out later in your pregnancy so that other tests can be done to look for certain problems in your developing baby.

How is amniocentesis carried out?

During the procedure itself, first you can expect to have an ultrasound scan similar to other scans during pregnancy. For this, gel is applied to your tummy (abdomen) and the ultrasound probe is passed over the skin of your tummy. This checks the position of the baby and also your afterbirth (placenta). Amniocentesis is usually carried out under 'continuous ultrasound guidance'. This means that the doctor performing the amniocentesis uses the ultrasound probe to allow them to see an ultrasound picture continuously throughout the procedure. By doing this, they can keep a close eye on your baby and the position of the needle used to draw off the amniotic fluid.

The doctor will use the scan picture to help them to find a clear 'pocket' or 'pool' of amniotic fluid around your baby. This means that the fluid can be withdrawn without the needle touching the baby or the umbilical cord. They will usually try to avoid inserting the needle through the afterbirth. However, occasionally, this can be the only way to collect the sample of fluid. If the needle does pass through the afterbirth it is unlikely to cause any harm to either you or your baby.

When the best position is found, your skin is cleaned around the area where the needle will be inserted. A fine needle is then pushed through your skin, the muscles of your tummy and then through the muscle wall of your womb (uterus) into the pocket of amniotic fluid. A syringe will be attached to the other end of the needle so that some of the amniotic fluid from around your baby can then be drawn off.

Usually between 10-20 ml of amniotic fluid are taken. The needle is then taken out and you will have another scan to check your baby. Your baby will quickly start to replace the amniotic fluid that is removed during the procedure the next time they pass urine. The amniotic fluid is normally a straw-like colour. However, sometimes it can be stained with blood. This is not dangerous but can sometimes interfere with the test results (as some of your blood cells will also be present) and the test may need to be repeated.

Occasionally, it is not possible to get enough amniotic fluid for testing and the needle has to be removed and reinserted. This may be due to the position that your baby is lying in. Hopefully, a second attempt at collecting fluid will be successful. If not, you will usually be asked to return on another day to try again.

You may prefer to have some support during the procedure. If you feel comfortable about this, consider asking your partner, a friend or a family member to accompany you. The whole procedure will probably take about 10 minutes but your appointment will usually last longer to give you time to rest afterwards.

Note: if you have not already been tested for HIV and hepatitis B, you will usually be offered testing before amniocentesis. You may also be offered testing for hepatitis C. This is so that if you do have one of these infections, the risk of you passing it on to the baby during amniocentesis can be kept to a minimum.

What tests are carried out on the amniotic fluid?

There are two main tests that can be done to look at the baby's chromosomes. The first is called a rapid test. It can look for the chromosomal disorders Down's syndrome, Edward's syndrome and Patau's Syndrome. Sometimes sex chromosome disorders such as Turner syndrome can also be detected on a rapid test. This test usually gives results within three days after amniocentesis. The results are usually very accurate in confirming whether or not the developing baby has these chromosomal problems. So sometimes only a rapid test is carried out.

The second test is a chromosomal microarray that looks at all of the baby's chromosomes in detail. It can show up other chromosomal disorders and abnormal genes. This takes longer to get the results - usually, two to three weeks. A chromosomal microarray test may be suggested by your doctor if other genetic conditions are suspected.

Various other tests can also be carried out on the amniotic fluid, depending on your particular situation. These can include tests to measure levels of certain proteins in the amniotic fluid to look for conditions such as phenylketonuria. Your doctor will advise which tests are best for you.

Occasionally, the chromosome test results are uncertain. If this is the case, you may be offered a repeat amniocentesis. However, this is rare and, in most cases, definite results are possible. There is also a very small chance that the test results for the rapid test are normal but that the chromosomal microarray test shows up a problem. Very rarely, a woman's chromosomal microarray result may be reported as normal but she will still have a baby born with a chromosomal disorder or other problem. This is because some changes in chromosomes may be so small that they are very difficult to see. Amniocentesis cannot exclude all possible disorders.

Also, it should be understood that amniocentesis results do not provide information about the physical development of your baby. The foetal anomaly ultrasound scan that is done at around 18-20 weeks of pregnancy can help to look for physical problems. However, it is also not possible for this scan to show up all abnormalities.

You should ask your doctor or midwife to explain how long it will take for the results of your amniocentesis. You should also ask them how you will receive the results. For example, you may be given another appointment or sometimes results are given by telephone.

Are there any complications of amniocentesis?

Most women who have amniocentesis during pregnancy have no complications. However, amniocentesis does carry the risk of some complications. You need to balance the small chance of these complications against having the extra information about the baby and their genetic make-up. Complications can include the following.

Miscarriage

There is a small risk of miscarriage with every pregnancy, whether or not you have amniocentesis or CVS. This is the background risk of having a miscarriage. It used to be thought that for women who have had amniocentesis after 15 weeks of pregnancy, there is about a 1 in 100 additional, or extra, risk that they will have a miscarriage. (This means that, out of 100 women who have amniocentesis, one will have a miscarriage that they would not otherwise have had.) Recent research suggests the risk is actually much lower than this. This might be because the way that the procedure is carried out is safer than it used to be. However, it is generally thought that any extra risk of miscarriage is lower if amniocentesis is carried out by someone who is very experienced at the procedure. The risk of miscarriage is higher if amniocentesis is carried out before 15 weeks.

It is not certain why there is a small chance that amniocentesis can lead to a miscarriage. It may be that it is caused by infection, bleeding, or damage to the amniotic membranes (the sac containing the amniotic fluid that surrounds the baby) caused by the procedure.

If a miscarriage does occur, it is most likely within the first 72 hours after an amniocentesis. However, a miscarriage can occur up to two weeks afterwards. Procedure-related miscarriages are uncommon more than three weeks after amniocentesis.

The additional risk of miscarriage after CVS is thought to be slightly higher than that after an amniocentesis done after 15 weeks of pregnancy. The difference in miscarriage risk may be because CVS is carried out earlier in pregnancy. (There is a greater risk of having a miscarriage earlier in pregnancy regardless of whether you have a diagnostic test or not.) Also, CVS is usually carried out because of suspected problems with the developing baby. Because of these problems, there may be a higher 'background' chance of miscarriage in that pregnancy whether or not CVS is done.

Infection

Infection can, rarely, occur after amniocentesis. Less than 1 in 1,000 women who have an amniocentesis will develop a serious infection in their amniotic fluid. Infection can be caused by a number of things - for example:

- By injury to your bowel with the needle used during the procedure so that germs that are normally contained inside the bowel escape.
- By germs that are present on the skin of your tummy (abdomen), travelling along the track of the needle into your tummy or womb (uterus).
- By germs that are present on the ultrasound probe or in the ultrasound gel, travelling along the track of the needle into your tummy or womb.
- Symptoms of such an infection can include a high temperature (fever), tenderness of your tummy and contractions of your womb. However, infection is very unlikely if correct procedures to reduce infection are followed.

There may also be a risk of injury to your developing baby with the needle used to perform the amniocentesis. However, 'continuous ultrasound guidance' during amniocentesis has reduced the chance of this complication and it is now very rare. Injury to the placenta is also a possibility but this doesn't normally lead to any problems and it usually heals by itself.

Rhesus disease in the developing baby

If your blood group is rhesus negative and the baby's blood group is rhesus positive, there is a risk that you may develop small proteins called antibodies against the baby's blood cells after amniocentesis. This means that there is a risk that the baby can develop rhesus disease. So, if you are rhesus negative, you will be advised to have an injection with anti-D immunoglobulin after amniocentesis so as to help to prevent this.

How can I expect to feel after an amniocentesis?

The procedure itself can be a little painful but most women feel that the discomfort is about the same as that experienced when having a blood test. Using a local anaesthetic before the needle is inserted doesn't seem to improve the pain that is felt.

It is best if you can arrange for someone to drive you home after amniocentesis, if possible. You should also take things easy over the following few days but total bed rest is not necessary. Some mild, period-like cramping tummy (abdominal) pains with some light spotting of blood from your vagina can be normal immediately after amniocentesis. You can take paracetamol to help ease the pain.

However, if you develop any of the following, you need to seek medical advice immediately, as they may be signs of complications:

- Severe tummy pain
- Contractions
- Persistent back pain
- Continuous bleeding from your vagina
- A watery fluid loss from your vagina
- A smelly discharge from your vagina
- A high temperature (fever)
- Flu-like symptoms.

What are my choices if the results are abnormal?

Deciding to have amniocentesis can be a very difficult decision and a very anxious time. However, most women who have amniocentesis will have a normal result. (That is, the baby won't have the genetic problem that the test was looking for.) However, before you go through amniocentesis, it is important to think through carefully what difference an abnormal test result would make to you. How would it be likely to affect your decision about whether or not to continue with the pregnancy?

Once you know the results, and if the results show a problem, you will need to make a decision about what is best for you and for your baby. This decision may be very difficult. You may find it helpful to talk things through with:

- Your GP
- Your midwife.
- A doctor who specialises in pregnancy and childbirth (an obstetrician).
- A doctor who specialises in the medical care of children (a paediatrician).
- A genetic specialist.
- An advisor (a counsellor).
- You may also wish to talk things through with your partner or family.

If you feel this is the right decision for you, there is time to have an abortion after amniocentesis shows an abnormality. The type of abortion will depend on how many weeks pregnant you are when you decide to end the pregnancy. You should discuss this with your doctor or midwife. If the decision is late then an induced labour is needed.

Equally, even if the results of amniocentesis do show a problem, you may choose to continue with the pregnancy. With the knowledge of the results, you can start to prepare for the birth and care of the baby, who is likely to have special needs. Your baby may need special care or surgical care immediately after they are born. Prior knowledge that your baby has a certain condition means that you can plan to give birth in a hospital where all the appropriate facilities are available.

In rare situations, amniocentesis may show that the baby has a condition that is treatable. Occasionally, there may be the possibility that treatment can be given while the baby is still in your womb (uterus).

Acknowledgment

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