



Amniocentesis

(English)

This leaflet gives you information about a procedure you have been offered called an amniocentesis.

You may have been offered an amniocentesis because:

- the result from your screening test for Down's syndrome shows you have a higher chance of having a baby with Down's syndrome;
- you have had an ultrasound scan which shows that your baby may have an increased chance of having abnormal chromosomes;
- you already have a child with a chromosome disorder or you had a previous pregnancy (which did not go to full term) in which a chromosome disorder or congenital anomaly (a problem present at birth) was found;
- you carry an inherited condition (for example, sickle cell disorders); or
- you have an infection such as toxoplasmosis which can cause a problem in your baby's development.

If you decide to have an amniocentesis, you can find information about the procedure on page 6 of the leaflet.

The information may help you decide if you want to have this procedure.

Your midwife and obstetrician can give you more information.



www.antenatalscreening.org

April 2009

A5

Introduction

An amniocentesis is a procedure to remove a small amount (about 15 to 20 millilitres) of amniotic fluid from around your baby in your womb. The cells from your baby that are floating in this fluid can be tested in the laboratory to look at the chromosomes. An amniocentesis can be done after 14 weeks of pregnancy (but is usually done after 15 weeks).

If you have an amniocentesis, there is a 1% chance that you could have a miscarriage (that means one in every 100 women could lose their baby). A miscarriage is most likely to happen up to three weeks after the amniocentesis. No one knows why this happens or who it will happen to. It can happen whether or not your baby has a chromosome problem.

Because an amniocentesis is only offered to some women, you may not be able to have it done in the maternity unit you usually go to. Instead, you may be offered an appointment at a different maternity unit where the obstetrician is trained to do the procedure.

If you have an infection such as HIV, hepatitis B or hepatitis C, you may need extra information and advice from a doctor who specialises in infectious diseases before you decide whether to have an amniocentesis.

Your midwife or obstetrician can explain the test to you.

You need to decide if you want to have this procedure.

If you decide to go ahead with an amniocentesis procedure, we may ask you to sign a form agreeing to the procedure (the consent form) before the procedure is carried out.

Chromosome tests

In each cell of our bodies, we usually have 46 chromosomes (23 pairs). These are numbered from 1 to 22, with the 23rd pair being the sex chromosomes: usually XX for a girl, usually XY for a boy. Each chromosome carries thousands of genes, which are the instructions that tell our bodies how to grow, develop and function.

In Wales, two different chromosome tests can be done on the amniotic fluid. These tests are as follows.

PCR (polymerase chain reaction) test

The PCR test usually only looks for the following three chromosome problems in the baby.

- Down's syndrome – causes some degree of learning disability but is not usually life-threatening for the baby. Occasionally the physical problems associated with Down's syndrome can be life-threatening (such as a heart problem).

- Edwards' syndrome – a severe chromosome disorder that can be life-threatening for the baby.
- Patau's syndrome – a rare and severe chromosome disorder that can be life-threatening for the baby.

Sometimes the PCR test can be used to look for other specific conditions. Your obstetrician or genetic counsellor will discuss this with you.

The PCR test looks for the most common chromosome problems. The karyotype test may find other chromosome problems or changes, but these are less common (see the next section). This can happen even if your PCR result is normal.

Karyotype test

This test takes longer than the PCR test but will detect most chromosome problems.

To do this test, the cells from your baby that are floating in the amniotic fluid are grown (cultured) in the laboratory (this takes about 12 days). The laboratory staff will examine the cells under a microscope to look for changes in the number and appearance of your baby's chromosomes.

A karyotype test will not detect **all** chromosome changes. Some changes are so small that they can't

be seen, even under the microscope, and can only be detected using extra specialist tests. These tests are not done on a karyotype.

The karyotype will not detect:

- changes in single genes, such as cystic fibrosis (each chromosome contains thousands of genes);
- microdeletions (loss of small segments of a chromosome); or
- other small changes in chromosomes.

Single-gene disorders

Single-gene disorders are caused by changes in a single gene. If you have a family history of an inherited disorder that is caused by an alteration in a single gene (such as cystic fibrosis), you may be offered a specific test, called a single-gene test, just for this. You may already have spoken to someone at the genetics service, or your midwife or obstetrician can refer you to the genetics service if you tell them you have a family history of an inherited disorder. If a single-gene test is done on an amniotic fluid sample, you will usually be offered a karyotype test as well.

If the result shows the usual number and arrangements of chromosomes, it does not guarantee that there is not a problem with your baby.

A PCR and karyotype test result doesn't show how your baby is growing and developing. Only an ultrasound scan between 18 and 20 weeks, when your baby is nearly fully formed, can show this.

If there is a problem

There will be some pregnancies where a problem is found. If there is a problem, you will be given information and support to help you make a decision about your pregnancy. There are experts who can give you information to help you decide what to do. These include fetal medicine consultants, geneticists and paediatricians.

Some women will want to prepare themselves for the birth knowing that their baby has a problem. Some women may decide to end their pregnancy, and others may decide to continue with the pregnancy and have their baby adopted. These are difficult decisions and you will be given time and information to help you make a decision that is right for you.

Finding out that your unborn child has a problem is distressing and deciding what to do is hard. Most women want and need some support. This might come from your partner, family or friends or from the health professionals who are caring for you. If you want, your partner

or a friend can come to hospital appointments with you.

The karyotype can detect many types of chromosome abnormalities. This can mean that although your baby does not have Down's syndrome, Edwards' syndrome or Patau's syndrome, the test result could show a different problem.

If this happens, you would be able to discuss the result with a doctor and with a midwife who specialises in antenatal screening. You would also be able to discuss the result with someone who specialises in genetics.

To find out whether your baby has a problem, we may need you and the baby's father to provide blood samples.

More information

You can get more information from the following.

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

Helpline: 0207 631 0285
Website: www.arc-uk.org
E-mail: info@arc-uk.org

If you decide to have an amniocentesis

Preparing for an amniocentesis

- You can have breakfast or a light lunch before your appointment.
- Your bladder needs to be full for the scan that is done before the amniocentesis, but you may be asked to empty it for the amniocentesis procedure.
- You may be more comfortable if you wear loose clothing.
- You can bring your partner or a friend with you for support during and after the procedure, but please do not bring children with you.
- If possible, arrange for someone to drive you home.

How the amniocentesis procedure is done

The procedure takes about 10 minutes and you will have it done as an outpatient, usually in the antenatal clinic. You will be awake for the procedure, and lying down.

You will have an ultrasound scan before the amniocentesis. This is to check the position of your baby and look for the best place for taking the sample of fluid from inside your womb. Your abdomen is cleaned with an antiseptic solution to reduce the risk of infection. The doctor inserts a needle through your skin

and the wall of your womb, and then takes a small amount of fluid from around your baby. The doctor will be watching the ultrasound scan to guide the needle and so avoid getting close to the baby. The test can be uncomfortable, but should not hurt.

Occasionally the procedure cannot be done due to the position of the baby. If this happens, the doctor may suggest that the procedure is done on another day.

There is a small chance that the doctor will not be able to get any amniotic fluid from around your baby. This means the test cannot be done. The doctor may suggest that the procedure is done again on another day.

What happens after the procedure

After the procedure, you may be asked to stay in the clinic for up to 30 minutes to rest. Some women have tummy cramps afterwards, rather like period pains.

If your blood group is Rhesus negative, you will be offered an injection of anti-D after the procedure. This is to reduce the chance of antibodies developing in your blood, which could happen if your baby's blood group is Rhesus positive.

Some doctors may advise you to take things easy for a couple of days after the procedure and to avoid having sex, or doing any heavy lifting or strenuous exercise. You won't need complete bed rest.

If you have any pain or discomfort, you can take a normal dose of paracetamol.

Most women are back to normal after two days.

What to look out for

- Severe pain, which you can't control by taking mild painkillers (like paracetamol).
- Any bleeding or unpleasant discharge from your vagina.
- Any fluid leaking from your vagina.
- If you suddenly feel unwell and have a high temperature or flu-like symptoms.

These symptoms do not always mean there is a problem but you may need more care and attention. For advice, please contact:

- the clinic where you had the amniocentesis; or
- your midwife.

Getting the results of the test

The midwife, doctor or geneticist will discuss with you how you want to get your result. You might prefer to get this information in person so that you can speak to someone who

can answer your questions. Results of tests for single-gene disorders take different lengths of time. The genetics team will discuss with you how you will get the results of single-gene tests.

PCR result

Your PCR result is usually available within three days.

About 1 in 25 amniocentesis samples are not suitable for the PCR test. If this happens, you will have to wait for the karyotype result. However, this does not mean there is a problem with the baby. In about 4% or 5% of PCR tests (four or five tests in a hundred) we may need blood samples from you and the baby's father to help us get a clearer result. Again this does not usually mean there is a problem with the baby.

Karyotype result

The karyotype results are usually ready within about two weeks.

Sometimes it takes a bit longer to grow the cells in the laboratory, and you will have to wait a few extra days for the result. If the cells grow more slowly, this is not because there is a problem with your baby.

Occasionally, in about 0.3% of tests (that is, one test in 300), the cells do not grow in the laboratory and you will not get a result from the karyotype. If this happens, you may be offered another amniocentesis.

The test does not give a clear result in 1% of tests (that is, one out of 100). Sometimes, the cells grow in a way which makes it difficult for the laboratory to say if there is a definite problem, and you may be offered more tests.

The karyotype test will show the sex of your baby. If you do not want to know the sex of your baby, please tell your midwife or doctor.

You can get more information about amniocentesis results from your midwife or from the clinic where you had the amniocentesis done.

