

Amniocentesis

This brochure explains a test called Amniocentesis (amnio) that is available in pregnancy. It is one of a number of tests that can be used to detect birth defects in pregnancy.

What is an amniocentesis?

An amniocentesis is a procedure that allows a small amount of the fluid around the developing baby (amniotic fluid) to be collected. This fluid contains cells from the baby that can be tested for some types of birth defects. The most common type of test done on an amniocentesis sample is a chromosome test. In some cases, tests for infections, spina bifida or other genetic conditions may be done.

Why should I consider an amniocentesis in my pregnancy?

An amniocentesis may be offered when there is an increased risk that the baby may have a chromosome problem or other type of birth defect. Some of the most common reasons include:

- If the woman will be older than 35 at the time of delivery of the baby.
- If an increased risk of a chromosome problem is identified on a screening test, such as maternal serum screening.
- If an ultrasound detects unexpected abnormalities in the baby.
- If the parents have had a previous baby with a chromosome problem, or a parent has a variation of the chromosomes that increases the chance of the baby having a chromosome problem.
- If there is an increased risk that the baby may have a particular genetic disorder, for which a test is available.
- If there is concern that the baby may have been exposed to some types of infections.

It is up to you and your partner whether you choose to have an amniocentesis. You can discuss the test further with your doctor or midwife.

There are many things to consider before choosing whether or not to have a test in pregnancy. The introductory brochure 'Testing for birth defects in pregnancy' will help you to consider these issues and understand some of the terms used. Always discuss any questions or concerns you have with your doctor.



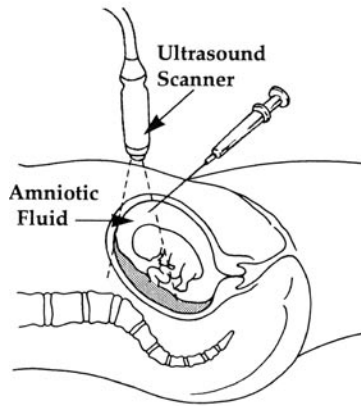
When is an amniocentesis done?

An amniocentesis is usually done around 15-18 weeks of pregnancy. It can be done later if there is a specific reason.

How is an amniocentesis done?

Amniocentesis is an outpatient procedure performed by a specially trained doctor. No special preparation is needed. It takes about 30 minutes. Most of this time is spent having an ultrasound and preparing equipment. The collection of the sample takes about a minute. Your partner or support person is encouraged to attend. It is best that children do not attend. Creche facilities are available free of charge at the hospital, ring 8161 6394 to book.

The procedure



Ultrasound is used to see where the baby, the placenta and a suitable pool of amniotic fluid are located. The woman's abdomen is cleaned with antiseptic. A fine sterile needle is guided through the woman's abdominal wall and the wall of the uterus (womb) into the amniotic fluid. The path of the needle is watched by ultrasound. As the procedure is rapid, a local anaesthetic is usually not necessary.

Most women say that the procedure is no more painful than any type of injection. Some women feel cramping when the needle enters the uterus or pressure as the fluid is being withdrawn. Once the needle is seen to be in the right place, a small amount (a few teaspoonfuls) of amniotic fluid is taken out and the needle is withdrawn. The sample of amniotic fluid is sent to the laboratory for testing. The amount of amniotic fluid in the uterus returns to normal within a few hours. Occasionally the doctor is unable to obtain enough amniotic fluid on the first attempt. If this happens, another sample may need to be taken.

What are the risks of having an amniocentesis for the woman and baby?

Some women experience cramping on the day of the test.

1 or 2 women out of every 100 women who have an amniocentesis experience bruising, some spotting or leakage of amniotic fluid after the procedure. This usually settles within 24 hours but should be reported to your doctor if it occurs.

It is estimated that about 1 woman in every 200 women will miscarry as a result of having an amniocentesis. In other words 0.5% of all the women who have this test will suffer a miscarriage.

What happens after the amniocentesis is done?

Arrangements will be made for you to receive the results. You are advised to rest at home for the remainder of the day. Your blood group needs to be known and, if it is Rh negative, you will usually receive an injection of anti-D at the end of the procedure. Your doctor will explain why this is necessary.

The results of your amniocentesis

The most common test done on an amniocentesis sample is a chromosome test. In order to analyse the chromosomes fully, cells from the baby in the amniotic fluid must first be grown. The final chromosome result is usually available within 2 weeks of the test being done. Under certain circumstances, a more rapid preliminary result known as a FISH result may be available within 24-48 hours.*

If an amniocentesis is done to test for infection or a specific genetic condition your doctor will be able to tell you how long it will take to get the results. In a small number of cases, in order to be able to interpret the results of the chromosome test fully, a chromosome test on a blood sample from the parents or further tests on the baby may also be needed.

In less than 1 in 1000 tests (0.1%), a result cannot be obtained from the sample and a repeat test is required. The amniocentesis will identify the sex of the baby. Parents may choose whether or not they wish to be told.

*A note about FISH (Fluorescent In Situ Hybridisation)

FISH is a rapid method for counting the number of certain chromosomes within cells. It can be used on an amniocentesis sample to get a faster result. It is always followed by the usual chromosome test. A normal FISH test is about 98% accurate in predicting that a baby will have a normal chromosome result. FISH is only offered in special circumstances that you can discuss with your doctor, or by payment of a non medicare-rebatable fee.

Does a normal amniocentesis result mean that my baby will be born healthy?

An amniocentesis is an accurate way of testing for most chromosome problems. However mosaicism and very small chromosome abnormalities cannot be excluded.

Amniocentesis cannot detect all problems with a baby. Having an amniocentesis does not guarantee that your baby will not have a birth defect as most are not caused by abnormalities of the chromosomes.

What if the test shows that my baby has a problem?

If the test shows that your baby has a problem, a medical specialist will talk to you about what this is likely to mean for the baby and what treatments or options are available to you.

Are there other tests that can tell whether my baby has a chromosome problem?

Chorionic villus sampling (CVS) is another test that allows a baby's chromosomes to be tested accurately. This test can be done earlier in the pregnancy. (10-11 weeks). Please read the brochure on CVS for further information about the difference between these tests. It is usual to choose either a CVS or an amniocentesis, not both.

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