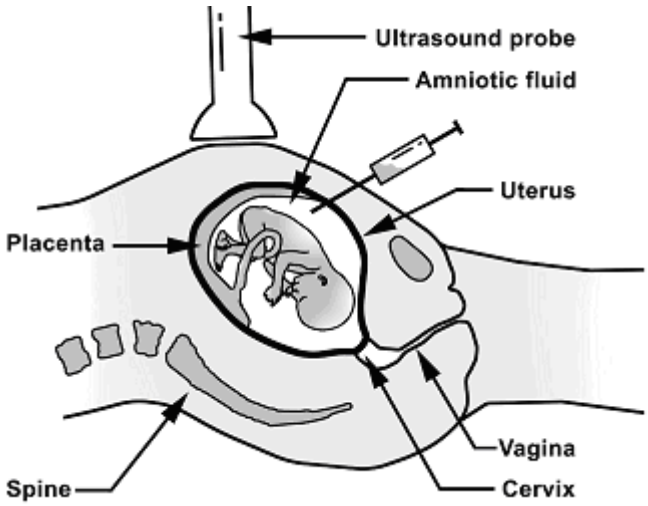
Amniocentesis

What is it?

Amniotic fluid is the fluid around the baby. As the baby grows he or she sheds skin cells into the fluid much as we do when we take a bath. Chromosomes (the structures which hold all the baby's genes) are held in these cells and arranged in pairs. This means we can take some of the fluid and look in the baby's cells to check the baby's chromosomes for abnormalities, such as Down syndrome. This test is called an amniocentesis.

How and when is it performed?

An amniocentesis can be performed after 15 weeks of pregnancy. After an initial ultrasound scan by the doctor your abdomen will be cleaned with antiseptic solution to make sure it is as sterile as possible. The doctor will be scanning you all the time to see where the baby is and where the best pool of fluid is. A fine needle is put through your abdomen into the womb to withdraw some of the amniotic fluid. About 15-20ml of fluid is withdrawn by syringe and the whole procedure takes no more than a couple of minutes. The pregnancy quickly makes up the fluid again. We do not usually give any local anaesthetic for the procedure as this is more uncomfortable than the amniocentesis! Most women describe the procedure as uncomfortable rather than painful and some describe an emptying feeling as the fluid is being withdrawn. Most people say it is no more painful than having your blood taken.



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Is there any special preparation?

There is no special preparation for an amniocentesis. On most occasions you do not need to have a full bladder but sometimes it can help if it is not completely empty. If you are Rhesus negative you will need an injection of anti-D immunoglobulin following the procedure. If your Rhesus status and blood group are not known you may need to have a blood test at the time of the amniocentesis.

Are there any risks?

Unfortunately the test does carry some risks. There is a small risk of miscarriage related to an amniocentesis. This affects about 1 in every 200 women tested. Other risks include failure in being able to perform the procedure, failure of the cells to grow and so not being able to be analysed, and a possibility that it will give a result that is difficult to interpret. However, these risks are small and occur in less than 1 in 100 tests. If there are problems you will be notified as soon as possible and offered a repeat test if that is what you want. There is a very small risk of about 1 in 1000 that amniocentesis will cause a serious infection.

After the test?

Following the test you should rest for the remainder of the day if you wish to, although there is no evidence that rest reduces the risk of miscarriage. It is normal to experience some cramping or spotting of blood from the vagina for a few hours afterwards. If you experience moderate or severe pain, or vaginal bleeding, you can contact the Fetal Medicine Team in work hours on Tel. 01603 286802, or, if you have been referred to us from elsewhere, contact your local hospital's Antenatal Screening Team if you prefer. Out of hours, you can either wait until the next working day but if the bleeding is heavy or the pain severe or you feel feverish, you should contact your GP or the Accident and Emergency Department. These symptoms do not necessarily mean you are going to miscarry but you would need to be checked.

How long will the results take?

Three types of laboratory tests can be used to look at chromosomes – PCR (polymerase chain reaction), micro-array and karyotype

PCR - checks for the most common chromosomal abnormalities. These are Down syndrome (trisomy 21, three number 21 chromosomes), Edwards syndrome (trisomy 18, three number 18 chromosomes) and Patau syndrome (trisomy 13, three number 13 chromosomes). It does not check all the chromosomes, only the most common abnormalities.

The PCR results usually take approximately 4 working days.

Array tests – if required you will be given a separate information sheet with more details about these tests.

Array test results take approximately 3 weeks

Karyotype – a karyotype involves looking at all the chromosomes to see if there is the correct number and if there are any extra or missing pieces. Array tests have now taken over as the test of choice as they give more information than a karyotype. Just occasionally it may be indicated to perform a karyotype but this will be fully discussed with you by the Consultant at the time.

Karyotype results take 2-3 weeks.

Not all gene or chromosome changes have clinical implications for a baby. If a change is found you will be offered further appointments with a specialist to discuss what the result might mean.

Most women undergoing amniocentesis are doing so because their screening test has put them at increased chance of having a baby with Down, Edwards or Patau syndromes. In this situation a PCR result will be offered as this will provide a quick answer for those particular syndromes. Array tests will be performed only when there is an increased risk of other chromosome problems not checked by PCR, either because of family history or an abnormality seen on the baby's scan. Karyotypes will only be offered under individual circumstances.

Are the results accurate?

Amniocentesis will reliably tell you if your baby has Down, Edwards or Patau syndromes or some other major chromosome abnormality, it cannot detect all genetic problems. These other genetic problems are, however, very uncommon.

Occasionally PCR, array or full karyotype do not give a clear result. This is rare but if it occurs, it will be discussed with you and you may be offered another amniocentesis. It is also possible, on a rare occasion, that a woman who has had a previous normal PCR result from an increased chance of Down, Edwards or Patau syndromes, may at a later stage find they are offered another amniocentesis to perform array tests where an abnormality is seen on ultrasound scan. This may show an abnormality not detected by PCR.

How will I get the results?

The doctor will arrange with you how you would like to receive the results. If there is an abnormality you will be contacted and given an appointment, usually the next day, to discuss what the result means, what options are open to you and to answer any questions you will have.

Post Procedure Information:

After amniocentesis, it's normal to have cramps similar to period pain and light vaginal bleeding called spotting for a few hours. You can take paracetamol (but not ibuprofen or aspirin) if you experience any discomfort. You may wish to avoid any strenuous activity for the rest of the day.

Please contact the fetal medicine midwives on (01603) 286802 Monday- Friday 08.30-16.30 or attend your local A&E department out of hour for advice as soon as possible if you develop any of the following symptoms after the procedure:

- Persistent or severe pain.
- A high temperature.
- Chills or shivering.
- Discharge or clear fluid from the vagina.
- Contractions (when your abdomen tightens then relaxes).
- Vaginal bleeding (more than spotting).

Any further questions?

The decision whether to have an amniocentesis or not, may be clear-cut for some but not for others. Please don't feel rushed into making the decision.

You can find further information re amniocentesis from the following:

Antenatal & Newborn Screening/ Fetal Medicine Midwives – 01603 286802

NHS website - https://www.nhs.uk/conditions/amniocentesis/

Antenatal Results and Choices (ARC) - https://www.arc-uk.org/