Amniocentesis

This information sheet may be available in different formats.

There are many reasons for having an amniocentesis performed, however the most common reason is to detect whether a baby has a chromosomal or genetic disorder. The intended benefit of having an amniocentesis is that you will be informed as to whether the baby's chromosomes are normal or not.

Introduction

Every year women have an Amniocentesis performed. Not all women need it, but for the ones that do, it means putting themselves and their pregnancy on hold for what might seem a lifetime.

The purpose of this booklet is to provide you with the facts and information about Amniocentesis so that you and/or your partner will know what to expect from this test. At the back of this booklet there is a small section of medical terms that you may find helpful and there are pages for you to jot down questions which you might like to ask at your visit.

Appointments

When you attend for your appointment please report to the X-Ray/Ultrasound reception desk. This is on the ground floor and can be found by following the 'elephant' sign. If you have any queries about your appointment, please discuss this with the office clerks. We always try to keep a strict appointment system as we are aware the stress you may feel.

However, sometimes a visit can take longer than expected causing some delay. We would like to apologise for any inconvenience this may cause.

Staff in the Fetal Centre

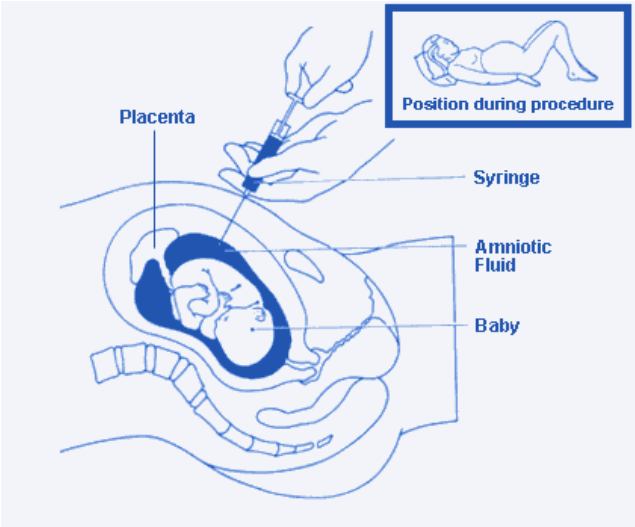
Who will you meet?
Consultants in Feto-Maternal Medicine
Specialist Midwives in Feto-maternal Medicine Tel: 0151 702 4211
Health Care Assistant

Other staff you may meet:
Several other people are involved with your care on the unit, and each one has a very important role.

If you have a query about your appointment please do not hesitate to contact the clerical staff in the ultrasound department. The consultants on the unit work with senior registrars who are training in fetal medicine, and during your visit you will meet them. Sometimes, student midwives and medical students will join them, if you have any concerns about them being present, please let us know.

What is Amniocentesis?

Amniocentesis means the removal of amniotic fluid (liquor) from around the baby. It can be performed as early as 15 weeks gestation.



What are the benefits of having an amniocentesis?

Accurate results regarding chromosomal abnormalities can be obtained early on in the pregnancy.

What alternatives do I have?

A CVS Chorionic Villus Sample (see separate sheet) also provides accurate results, however this test is performed earlier in the pregnancy. You may choose not to have any invasive test and wait for the baby to be born.

How is the test done?

The doctor/midwife will first perform an ultrasound scan to assess the size of your baby by taking measurements of the head, leg and abdomen. An overall estimate is taken of the amniotic fluid around the baby and the placenta is checked for its location in the womb.

Prior to the test, your skin is cleaned with antiseptic wipes and then the procedure begins. Ultrasound is used whilst inserting a needle into the amniotic sac so that the needle can be positioned away from the baby. 20mls of fluid is withdrawn which is sufficient for the laboratory to analyse. The fluid is

immediately put into a sterile plastic pot, which is labelled with your name, date of birth, address and hospital number. The details are then checked with you.

Your sample

The plastic container, which holds your amniotic fluid sample, will be looked at by the Regional Genetics Laboratory.

The cells are then looked at by one member of the prenatal laboratory team and checked by another senior scientist. The result is interpreted and a report is faxed to your consultant.

How long do the results take?

In the majority of cases a rapid preliminary test called QF-PCR is performed on the genetic material (DNA) from the amniotic fluid cells. This detects 80% of all chromosome abnormalities (Down, Edwards and Patau syndromes and sex chromosome abnormalities). The results should be available within 4 working days.

In order to look for any remaining detectable abnormalities, cells from your sample will be grown in culture and slides prepared. The results from this test will be available within 3 weeks from having the amniocentesis. There are some genetic or chromosomal conditions that cannot be detected by these tests.

The DNA from your sample will be stored for 1 year as an internal quality control measure. The slides from the sample will be stored as an internal quality control measure for 5 years.

Otherwise following investigation the cells will be disposed of in accordance with health and safety.

Does having Amniocentesis hurt?

Women frequently ask this question. Everybody's perception of pain differs. It is true to say, however, that most women find test 'uncomfortable'. Some say it is like having a blood test.

Possible complications

Here at Liverpool Women's Hospital the chance of a miscarriage following the amniocentesis is 0.5% (1-200). It can be difficult to say whether a miscarriage is due to the test or would have happened anyway. Other possible causes are infection, the waters breaking or the womb becoming irritable.

Reliability

Amniocentesis is 99.9% reliable for diagnosing chromosomal abnormalities with only a small chance of a wrong result.

Test failure

The rapid test fails in 2.2% of cases because the sample:

- Is contaminated by blood
- Does not give enough information for the laboratory to get a result

The sample will still go on to culture. Failure of the rapid test DOES NOT mean that the culture will also fail. Cultures fail in 0.5% of cases. This can either be due to:

- Fetal cells not growing
- Fetal cells that start to grow then stop
- A different type of cell growth (mosaicism)

In any of these situations you will be contacted and informed of the problem and several options will be discussed with you. The options are:

- Have the amniocentesis again
- Do nothing at all

Amniocentesis results

The results are sent to the fetal centre by fax.

If the results are **normal** we will contact you by 1st class letter.
If the result is **abnormal** then we will contact you at home by phone as it is necessary to speak to you as soon as possible to discuss the result in depth.

Culture failure

The specialist midwife will notify you by telephone to discuss the matter. You may want to have another test, and so another appointment can be arranged for you.

Some general advice

After the test

Following the test you will be taken to a counselling room to rest before going home. Take things easy for the next couple of days and try and avoid household chores that involve lifting, bending and stretching.

Some ladies experience lower abdominal discomfort within the 24-48 hours following the procedure. This is fine and can be expected because of the procedure. Paracetamol can be taken to relieve this.

If you experience any of the following symptoms you should contact the Gynaecology Emergency Room or delivery suite here at this hospital for further advice:

- Vaginal spotting of blood/bleeding like a period
- Develop a temperature

- Feel unwell
- No fetal movements (if you have previously felt movements)
- Loss of fluid from the vagina

Other tests that can be performed on amniotic fluid

Optical density test (0D450)

Pregnant women can sometimes produce antibodies, which can be harmful to their baby.

This condition, known as haemolytic disease, tends to be more common in women who are Rheus negative. Certain antibodies destroy red blood cells and can result in anaemia. As a result of red blood cell destruction, a substance called bilirubin is released. This can be measured using an optical density test. The bilirubin levels will show how much the baby is affected by the antibodies.

Fetal blood grouping – PCR

Amniotic fluid can also provide information about your baby's blood group. This test is only performed in special circumstances.

Viral and infection screen

If the doctor suspects that a woman or her baby has had a virus, amniotic fluid can be used to trace the virus or infection.

Spina Bifida

Sometimes on ultrasound it can be difficult to see spina bifida, especially if the lesion is at the lower end of the spine. A baby with a damaged spinal chord releases an enzyme called acetylcholinesterase which will be present in the amniotic fluid. This test is not routinely performed as most of the time spina bifida is detected by ultrasound.

Support Groups and useful addresses

The Alder Centre
Alder Hey Children's Hospital
Eaton Road
Liverpool
L12 2AP
Tel Daytime: 0151 228 4811
Tel Evening: 0151 288 9759
(19:00 – 22:00 hrs)
Offers support and understanding to anyone who has experienced the death of a child.

Down's Syndrome Association
155 Mitcham Road
London
SW17 9BR

CLAPA (Cleft Lip and Palate Association)
Dental Department
The Hospital for Sick Children
Great Ormond Street
London
WC1 0EP
Tel: 0171 388 1382

The Miscarriage Association
c/o Clayton Hospital
Northgate
Wakefield
West Yorkshire
WF1 3JS
Tel: 01924 200799

***SANDS** (Stillbirth and Neonatal Death society)*
28 Portland Place
London
W1 4DE
Tel: 0171 436 5881

***CRUSE** (Care for the Bereaved)*
Cruse House
126 Sheen Road
Richmond
Surrey
TW9 1UR
Tel: 0181 940 4818

***ARC** (Antenatal Results and Choices)*
73-75 Charlotte Street
London
W1 1LB
Tel: 0207 631 0285

Medical Glossary

Some common terms used in this booklet.

Fetus – The developing baby in the womb.

Chromosomes – Thread like structures in a cell which carry the blueprint or genes that give people their characteristics.

Gene – A single unit of inherited characteristic, situated in a chromosome.

Placenta – Also known as the 'afterbirth', this is a unique organ in the womb that supplies the baby with nourishment and the removal of waste products.

Amniotic fluid – Also called liquor, this is the fluid that surrounds the baby.

Amniocentesis – Puncture of the amniotic sac and the removal of the fluid from around the developing baby for testing.

Ultrasound scan – A way of creating an image of the fetus by the use of high frequency sound waves which are beyond the range of human hearing.

Soft 'markers' – Unusual features seen on scan.

Chorionic Villus Sampling (CVS) – The removal of small fragments of placental tissue.

Cordocentesis – The removal of small fragments of placental tissue.

Miscarriage – The loss of the baby before 24 weeks gestation.

Genetics – The study of human inheritance.