Leeds Amniocentesis Information Sheet

What is amniocentesis and when can it be done?
Amniocentesis involves putting a fine needle through the tummy wall and into the uterus (womb) in order to obtain a sample of the water (amniotic fluid) from around the baby. As the baby sheds cells into the fluid these can then be collected for analysis. Amniocentesis can be carried out at any stage of pregnancy from 15 weeks onwards.

When will I get my results?
Usually after 48 hours but may take 18 days (see below)

Results after a raised downs test screen.
If this test is done because you are at increased risk for Down's syndrome, for example after a triple test it will be analysed using a test called quantitative PCR. This is a very reliable test that provides a result within 48 hours. However, it only looks for three chromosome anomalies. These are Down syndrome and two rarer but more severe problems, Edwards and Patau syndrome. This test will not tell you the sex of your baby. PCR cannot be performed if the fluid taken from you is discoloured or blood stained. In this situation we would need to culture the fluid (see below).

What is quantitative PCR?
Very simply this is a test performed in the lab which measures the amount of genetic material in the sample. In a baby with a normal set of chromosomes we would expect to see a given amount of genetic material from a specific chromosome. In Down syndrome the baby would have 3 copies of the number 21 chromosome rather than 2. Quantitative PCR would demonstrate this increased amount. The test has been proved to be both very reliable and quick.

Why have I been asked to give a blood test as well?
The lab requires a blood sample from the mother to exclude maternal contamination of the amniotic fluid sample.

Do you know your blood group?
If you are Rhesus negative we need to know as you will need to be given an injection of Anti-D. This is because putting a needle into the pregnancy could cause a small amount of the baby's blood to enter your circulation

Results after an abnormality has been seen on ultrasound
If there is a concern after the scan then in addition to the PCR result we may ask the laboratory to also culture the fluid. This process takes 2-3 weeks. The difference is that now they can look at all of the chromosomes and potentially pick up minor abnormalities which would not be detected on PCR alone. In about 1 in 200 cases the culture will fail and no result will be obtained. If this occurs you may have to have a second test.

Why do we not culture every amniocentesis?
It has been decided that the chance of missing a chromosome abnormality using PCR alone is very small if your baby appears normal on ultrasound. In addition the full culture can very occasionally identify a potential anomaly for which we do not know the significance.

Why does the result take so long?
The results take this long because the cells in the amniotic fluid have to be grown in the laboratory before the chromosomes can be examined down a microscope.

How is the test done?
An ultrasound scan is used to ensure the pregnancy is at the right stage for testing. If the scan showed an unsuspected problem this would be discussed with you. Watching with ultrasound all the time a fine needle is passed through the wall of the uterus into the fluid; about 15ml (3 teaspoons) of fluid is withdrawn. We do not use local anaesthetic for the test as the stinging caused by the local anaesthetic is worse than the test itself.

Consent
Many units will ask you to sign a consent form prior to the procedure. This is to document that you have been informed about the test and have agreed. Others will not ask you to sign as the test is carried out with you wide awake which implies that you are happy for the doctor to proceed.

Are any precautions necessary afterwards?
Your abdomen will feel slightly sore for a few hours afterwards however there are no particular precautions that you need to take. A simple painkiller like paracetamol can be taken if needed. The only thing you will have to show for the test is a small puncture wound, similar to when blood is taken from your arm, from the needle on your abdomen. This requires no special care and does not need to be kept dry so you can bath or shower as normal.

What are the risks?
It is important that you realise that amniocentesis is associated with a small risk to the pregnancy, even if the procedure is straightforward. The main complications are miscarriage or rupture of the membranes of the sac the baby is in. The risk of causing a miscarriage with amniocentesis is between 1 in 100 to 1 in 150. About 1 in every 100 women who have an amniocentesis will leak fluid vaginally in the first 24 hours. This is because the membranes failed to seal when the needle was removed. If this happens we would recommend resting. However, it is important if you have any vaginal bleeding, leakage of fluid, or more pain that you would expect from the procedure you should report this to your General Practitioner or the Hospital

If I am going to miscarry when will it occur?
Most miscarriages will occur within 3 weeks of the test. The reasons for miscarriage are probably either as a result of puncturing the sac or introducing infection. Taking antibiotics has not been shown to reduce this risk.

Can my baby be harmed?
As we watch where we place the needle using ultrasound this should not happen.

Is there anything else I need to know?
If a culture is performed (see above) occasionally normal and abnormal cells are found together in the same sample. This condition (mosaicism) is uncommon, occurring in about 0.6% of amniocenteses. It is usually clear which cells represent the baby's chromosomes but it is sometime necessary to carry out further tests to sort out this problem.

How will I get my result?
The person performing the test will inform you how and when you will get the result. For most people this will be by phone within 48 hours. It is obviously important that you therefore give us a phone number where you are happy to be phoned. It is usual to receive a letter of confirmation of these results but this may be some time after the procedure.

How do you decide whether or not to have a test and which test to have?
It is very important to understand that amniocentesis is capable of diagnosing only a limited number of conditions. For most people this is a straight question of does my/our baby have Down syndrome? As an individual or couple you need to be aware of the limitations of the test and the potential risks and weigh these against the information amniocentesis will provide.
If you have any further queries or questions, please do not hesitate to let us know and we will be happy to discuss them with you.

Audit
It is very important for our records that we audit the outcome of all pregnancies who have any invasive test (amniocentesis, CVS, fetal blood sampling) in pregnancy and as such we would be grateful if you would inform us of the outcome.