

# The Amniocentesis



Information for Patients and Families

## **The Amniocentesis**

The following gives you information about the amniocentesis. It tells you what an amniocentesis is, when and how it is done, what happens after the test, and possible benefits and risks of taking the test. This leaflet is designed to be used alongside the discussions you have with your health care professionals and help you to ask the questions that are important to you.

### **What is the amniocentesis?**

The amniotic sac is the bag of fluid in which the baby floats in the womb. Amniocentesis is a way of taking some of the fluid in that sac for genetic testing during pregnancy. It is most commonly used to check the baby's genes or chromosomes for specific genetic conditions. It may be offered to you for a number of reasons.

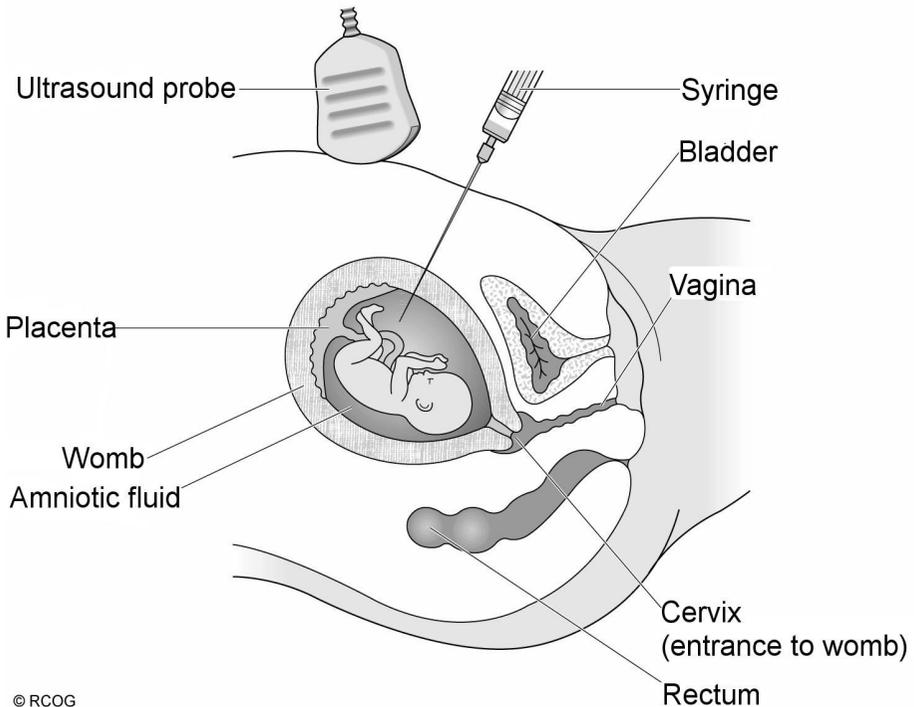
- You are an older mother (35 years or older) and therefore have an increased risk of having a child with a genetic condition such as Down syndrome.
- You or your partner has a genetic condition which may be passed on to the baby.
- There is a genetic condition in you or your partner's family, and there is a risk that the condition may be passed on to the baby.
- You have had a previous child affected by a genetic condition.
- You have had another type of test that is done during pregnancy (such as an ultrasound, nuchal translucency scan or blood test). It has shown that there is an increased risk that your baby has a genetic condition.

### **When is the amniocentesis done?**

Amniocentesis is usually done after 15 weeks of pregnancy.

## How is the amniocentesis done?

Amniocentesis involves taking a small amount of the amniotic fluid that surrounds the baby in the womb. First, an ultrasound scan is done to check the position of the baby and the placenta (or after-birth). The skin over the womb area is then cleaned with antiseptic solution. Next, a fine needle is passed through the skin and the abdomen (the tummy or belly) into the womb, and a syringe is used to remove a sample (about 15mls or 3 teaspoonfuls) of the fluid that surrounds the baby. The fluid surrounding the baby contains some of the baby's skin cells and these can be examined in the laboratory to check the baby's genes and chromosomes. Very occasionally, the health professional performing the test is unable to get enough fluid at the first attempt and may need to re-insert the needle.



## **Is the amniocentesis painful?**

Most women consider amniocentesis to be uncomfortable but not really painful. It is usually over in a few minutes. Some women get a tightening feeling in the womb afterwards, or may feel a little soreness for one day. This is not unusual.

## **What will happen after the amniocentesis?**

The test itself should only take a few minutes. It is a good idea to bring a companion with you for support both during and after the test. You should take things easy for a couple of days after the test. Avoid any heavy lifting or strenuous exercise. If you have abdominal discomfort which lasts longer than 24 hours, or if you have fever, or if you have any unusual vaginal discharge or vaginal bleeding, you should let your doctor know.

## **What are the risks of the amniocentesis?**

Up to 1 woman in 100 (1%) will have a miscarriage as a result of having an amniocentesis. We do not really know why this happens. However 99 out of 100 pregnancies (99%) should continue normally. Other than this, there is no evidence that amniocentesis is harmful to your baby.

## **Is the amniocentesis reliable?**

You should discuss with the doctor the accuracy of the particular genetic test that you are considering, as this will differ depending on the type of change in the genes or chromosomes for which the test is being done.

Occasionally (1 in 100 samples) there are not enough cells obtained from the amniocentesis for the test to be done on that sample. When this happens, it is usually obvious in the

laboratory about 7 to 10 days after the amniocentesis. If this happens, you will be notified and offered another test.

## **Can all genetic problems be detected with the amniocentesis?**

Test results usually only provide information about the condition that was tested for. Occasionally the test may uncover results relating to other conditions. There is no general test for all genetic conditions.

## **How long will it take to get the results of the amniocentesis?**

The time it takes to receive the results depends on which condition is being tested for. For some conditions it will only take 3 days to get the test results. For others it will take 2-3 weeks. If the result takes longer than this, it does not necessarily mean that something unusual has been found, it may mean that the cells are taking a longer time to grow.



If you are having an amniocentesis for a rare genetic condition, ask the doctor how long it will take to get the results back.

When the test results are ready you may be called back to speak to the doctor, or you may be given the results another way, such as by telephone. You should discuss this with the doctor at the time of the test.

## **What if the result shows the baby has a genetic condition?**

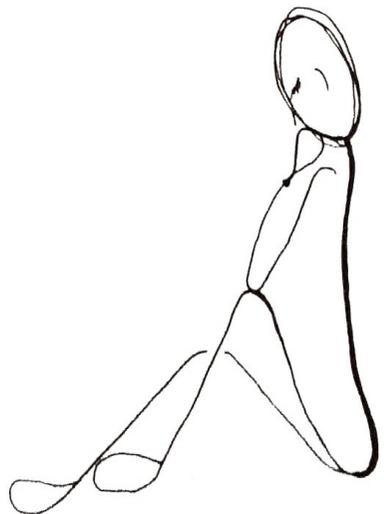
If the result shows the baby has a genetic condition, the doctor will discuss what this means and how this could affect your baby. They will discuss with you if there is a cure or treatment available. They will talk about your options and sometimes the possibility of ending the pregnancy. They will help you consider what is best for you and the baby. Very occasionally the test will uncover an unusual chromosome arrangement where the impact on the baby will be unclear.

## **HIV infection**

If you are HIV positive, there is a small risk that the amniocentesis might cause the HIV virus to be passed on to your baby. It is important therefore that if you are HIV positive you discuss this with your midwife or doctor as measures need to be taken to minimise the risk of transmission to your baby during the amniocentesis.

## **Making a decision about the amniocentesis**

Making a decision about having an amniocentesis test during pregnancy can be difficult. It is important to remember that you do not have to take the amniocentesis test if you do not want to. You should only have an amniocentesis if you and your partner feel that it is important for you to have the information which the test can provide, and do not feel the risks are too great for you to take.



To help you make the decision which is best for you, you should discuss the following information with your doctor:

- Information about the condition being tested for.
- The risk that the baby has the genetic condition you are thinking of testing for.
- Information about the test and what the results will tell you.
- The reliability of the test.
- The risk of having an uncertain result and having to take the test again.
- The risk of miscarriage.
- How long it will take to get the test results.
- How you will get the test results.
- Your options if the baby is found to have a genetic condition.
- How the experience may affect you and your partner emotionally.

These are some of the issues you should think about before making a decision about the amniocentesis test. You can also look at the 'Frequently Asked Questions' leaflet, which lists a number of questions you might want to ask the doctor about genetic testing. It has been developed by people who have been through a similar experience to your own.

Bring any questions or concerns with you to your appointment – write them down. If you need an interpreter, let the department know.

This is only a brief guide to the amniocentesis test. More information can be obtained from your local regional genetics centre ([www.geneticalliance.org.uk/services.htm](http://www.geneticalliance.org.uk/services.htm)) or from these addresses:

### **Antenatal Results and Choices (ARC)**

73 Charlotte St.,  
London,  
W1T 4PN

Tel: 020 76

Providing su  
information f

antenatal tes

[info@arc-uk.org](mailto:info@arc-uk.org)

[www.arc-uk.org](http://www.arc-uk.org)

### **Unique -**

### **The Rare Chromosome Disorder Support Group**

PO Box 2189,  
Caterham,  
Surrey

CR3 5GN

Telephone: 01883 330766

[info@rarechromo.org](mailto:info@rarechromo.org)

[www.rarechromo.org](http://www.rarechromo.org)

Modified from leaflets produced  
by Guy's and St Thomas'

Hospital, London; the Royal

College of Obstetricians and Gynaecologists [www.rcog.org.uk/index.asp?PageID=625](http://www.rcog.org.uk/index.asp?PageID=625) and London IDEAS Genetic Knowledge Park according to their quality

standards.

January 2007

This work was supported by EuroGentest, an EU-FP6 supported NoE contract number 512148

Illustrations by Rebecca J Kent

[www.rebeccajkent.com](http://www.rebeccajkent.com)

[rebecca@rebeccajkent.com](mailto:rebecca@rebeccajkent.com)

### **Genetic Alliance UK**

Unit 4D, Lerov House,  
436 E

OP

7704 3141

ation about

conditions and

et details of support

organisations.

[mail@geneticalliance.org.uk](mailto:mail@geneticalliance.org.uk)

[www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)

### **EuroGentest**

Free-access website providing

information about genetic

testing and links to support

groups across Europe.

[www.eurogentest.org](http://www.eurogentest.org)

### **Orphanet**

Free-access website providing

information on rare diseases and

orphan drugs, and links to

support groups across Europe.

[www.orpha.net](http://www.orpha.net)



**Genetic Alliance UK**  
Supporting. Campaigning. Uniting.