Amniocentesis and Chorionic Villus Sampling (CVS)
**Why we offer these tests**

The usual reason for having one of these tests is to find out if there is any problem with the chromosomes in the baby’s cells. Sometimes the test is done for known genetic conditions.

The normal number of chromosomes in a human cell is 46. At conception the egg and sperm, which each have 23 chromosomes, fuse to form one cell with the full 46. This then develops into the baby and the placenta.

Babies who have abnormal chromosomes may have too few, too many, or a rearrangement. One of the commonest problems is an extra copy of chromosome 21, which results in the baby having Down’s syndrome.

Chromosomes are made up of many genes. Some genes can be identified in particular genetic conditions.

The tests therefore can be used to detect specific gene problems where a prenatal test is available. For example parents who both carry the gene for cystic fibrosis may decide to a CVS to identify if their baby is affected.

CVS is sometimes a better test than amniocentesis to obtain information on certain genetic conditions.

In special circumstances other tests may be carried out on the cells or the amniotic fluid for example the baby’s blood group, some infections and some inherited (genetic) diseases.
Amniocentesis

What is amniocentesis?
Amniocentesis means obtaining a sample of the amniotic fluid, which surrounds the developing baby in the womb. The fluid is then sent to the laboratory for testing.

When and how is it done?
The safest time to do an amniocentesis is from 15 weeks onwards. It involves inserting a fine needle through the mother’s abdomen into the womb and taking a small amount of the amniotic fluid from around the baby. Ultrasound scanning is used to ensure the needle enters the fluid safely, away from the placenta and the baby.

Local anaesthetic is not necessary, indeed the stinging caused by the local anaesthetic can be worse than the test itself!

Rarely, in about 8 in every 1000 women having an amniocentesis, insufficient fluid is obtained on the first pass of the needle and it may need to be reinserted.

What are the risks associated with the test?
The passage of a fine needle into the womb is associated with a small risk of miscarriage, even if the procedure is straightforward. This risk is about 1 in 100 extra to the normal risk of miscarriage in pregnancy. The risk is greatest within the first 24 - 48 hours, although on rare occasions miscarriage can occur up to 6 weeks later.

There is a potential risk of causing rhesus antibody problems if you have a rhesus negative blood group. To prevent this, a blood test is taken following the amniocentesis and a standard dose of Anti-D is given.
Very rarely infection can set in inside the womb, which may not only lead to miscarriage but to serious infection in the mother.

If you are concerned about any of these risks, or have any further queries, please speak to your consultant or specialist midwife.

**What about the results?**
The results usually come in 2 parts. The initial result, available in around 48 hours, tests for the 3 commonest extra chromosomes (Down’s Syndrome; Edwards Syndrome; Patau Syndrome) and the sex chromosomes. The cells are then grown to allow full analysis of the chromosomes in about 2 weeks. Occasionally the initial result is not obtainable eg. if there is blood staining of the fluid.

After the test the specialist midwife will discuss with you how you would like to receive your results.

**Are the results reliable?**
The chromosome analysis from this test is approximately 99% accurate. Rarely the analysis shows up the mother’s chromosomes rather than the baby’s. Also rarely a result will not be obtained because the cells fail to grow.

Tests done for other specific reasons have different degrees of accuracy. These can be discussed with you.

**What happens after the amniocentesis?**
The test is performed as an outpatient and as soon as it is finished, or following your Anti-D injection if you have a rhesus negative blood group, you may go home.

It is advisable to rest for the next 24 - 48 hours. It is not usually necessary to go to bed but you should avoid any heavy lifting or strenuous exercise. Some women get a tightening feeling in the womb afterwards, or may feel a little sore. This is not unusual.
Are there any alternatives to amniocentesis?
An alternative to amniocentesis is CVS. This is where placental tissue is tested rather than the amniotic fluid. This can only be done after 11 weeks of pregnancy. The miscarriage rate may be slightly greater from this procedure than from amniocentesis.

Chorionic Villus Sampling (CVS)

What is Chorionic Villus Sampling (CVS)?
This is where a sample of trophoblast (the tissue of the developing placenta) is taken from the womb. The placental tissue is usually identical to the baby’s tissue. A trans-abdominal CVS may be performed from 11 weeks onwards.

CVS is offered to women with a known Down’s risk if the results are available by 13 weeks. It is also offered to women who have a high chance of having a baby with a genetic disorder or sometimes when a structural difference is identified by chance on the dating scan.

When and how is CVS performed?
CVS involves taking a tiny piece of the developing placenta from where it is attached to the womb.

Before the procedure your skin is cleaned and some local anaesthetic is used to numb your abdomen where the needle will be inserted.

Using an ultrasound probe to guide the direction, a needle is inserted through the abdomen and the wall of the womb into the placenta.
What are the risks associated with the test?
Every pregnancy carries a risk of miscarriage. CVS may sometimes cause a miscarriage due to injury or infection in the womb.

The additional overall risk of miscarriage from CVS is approximately 1 - 2%. In other words about 1 - 2 in every 100 women who have CVS under ultrasound guidance between 11 - 13 weeks will miscarry.
What about the results?
A quick result is usually available in 24 - 48 hours for some common chromosomal problems such as Down’s syndrome.

The complete results take about 2 weeks to check all 23 pairs of chromosomes thoroughly.

Results for single gene tests take a variable amount of time.

You may choose how you would like to receive the results of your test, either by telephone, or at an antenatal clinic appointment. This will be discussed with you when you attend for the test.

Are the results reliable?
The chromosome analysis from this test is approximately 98% accurate. Rarely a result will not be obtained because the sample is inadequate or the cells fail to grow. In 1% of cases the result may be confusing because sometimes more than one line of cells will grow in the placenta. When this happens it is usually recommended to have an amniocentesis after 15 weeks to check that this does not represent the baby's cells.

Tests done for other specific reasons have different degrees of accuracy. These can be discussed with you.

What happens after the CVS?
After the procedure it is advisable to rest for the remainder of the day. It is not usually necessary to go to bed but you should avoid any heavy lifting or strenuous exercise.

You may notice some ‘spotting’ of blood and cramping for a few hours afterwards. This is normal.

If you experience any unusual symptoms immediately after the test, such as feeling shivery (as if you have flu), fluid loss, bleeding or contractions then you should seek advice immediately.
Who do I contact if I have any problems after these tests?
If you have any abdominal (tummy) discomfort which lasts longer than 24 hours, or any pain, or if you have any watery discharge or bleeding, contact the:

Fetal and Maternal Medicine Centre
Telephone: 01332 785409
Monday to Friday, 8.30am - 5.00pm

Pregnancy Assessment Unit
Telephone: 01332 785796

Alternatively, if you prefer, you can contact your community midwife or GP.

Sources of information/references

Royal College of Obstetricians & Gynaecologists
Amniocentesis and CVS: What you need to know. July 2010

Royal College of Obstetricians & Gynaecologists
Website: www.rcog.org.uk

These organisations offer support:

ARC (Antenatal Results and Choices)
73 Charlotte Street
London W1T 4PN
Helpline: 0207 631 0285 (Monday to Friday, 10am - 5.30pm)
Email: info@arc-uk.org
Website: www.arc-uk.org

Contact a Family
209 - 211 City Road
London EC1V 1JN
Helpline: 0808 808 3556 (Monday to Friday, 10am - 4pm)
Email: info@cafamily.org.uk
Website: www.cafamily.org.uk
**Down's Syndrome Association**  
Langdon Down Centre  
2a Langdon Park  
Teddington TW11 9PS  
Helpline: 0845 230 0372 (Monday to Friday, 10am - 4pm)  
Email: info@downs-syndrome.org.uk  
Website: www.downs-syndrome.org.uk

**Genetic Interest Group**  
Unit 4D, Leroy House,  
436, Essex Road  
London N1 3QP  
Telephone: 0207 704 3141  
Email: mail@gig.org.uk  
Website: www.gig.org.uk/index.html

**Sickle Cell Society**  
54 Station Road  
London NW10 4UA  
Telephone: 0208 961 7795  
Email: info@sicklecellsociety.org  
Website: http://www.sicklecellsociety.org

**SOFT UK (Patau syndrome, Edward syndrome)**  
48 Froggatts Ride, Walmley  
Sutton Coldfield B76 2TQ  
Telephone: 0121 351 3122  
Email: enquiries@soft.org.uk  
Website: http://www.soft.org.uk

**The Miscarriage Association**  
c/o Clayton Hospital  
Northgate, Wakefield  
West Yorkshire WF1 3JS  
Helpline: 01924 200799 (Monday to Friday, 9am - 4pm)  
Email: info@miscarriageassociation.org.uk  
Website: wwwmiscarriageassociation.org.uk/main4.htm
If you have any queries, or require further information please contact your GP or Midwife for advice. Alternatively, telephone the Royal Derby Hospital on 01332 340131 and ask for your ward/clinic.
Smoking is not permitted anywhere in the buildings and grounds of Derby’s Hospitals. For advice and support about giving up smoking please call Free Phone 0800 707 6870.