

## Introduction

*This fact sheet is intended to provide a guide for **health professionals** not usually involved in undertaking these procedures but who may be asked questions from women about amniocentesis and chorionic villus sampling (CVS). Detailed information about cytogenetic and genetic counselling is available in the Antenatal Screening Wales (ASW) Education Resource Pack (Information Papers).*

*To avoid delay or inappropriate antenatal testing, it may be beneficial to refer families requesting a test because of their genetic family history to the all Wales genetics service for advice.*

*Women should be provided with a verbal discussion and a suitable patient information leaflet.*

## Fact Sheet

### When is the procedure performed?

#### Amniocentesis

Amniocentesis is usually performed after 15 weeks of pregnancy and most commonly between 15 and 20 weeks of pregnancy.

#### CVS

CVS can be performed after 10 weeks of pregnancy, but is more commonly performed after 11 weeks. In some circumstances it can be performed after 14 weeks gestation.

### What happens during the procedure?

An ultrasound scan is performed before the amniocentesis and CVS to check the position of the baby and the placenta to assess the best location for:

- taking a sample of amniotic fluid in an amniocentesis
- taking a sample of placental tissue in a CVS.

These procedures are performed under continuous ultrasound guidance.

#### Amniocentesis

A needle is passed through the abdomen and then a syringe is used to withdraw a small amount (15 to 20mls) of amniotic fluid from around the baby. The amniotic fluid in the uterus will be replenished quickly.

Some doctors numb the skin first with a local anaesthetic but the research evidence suggests that local anaesthetic does not reduce pain scores.<sup>1</sup> Therefore, the use of local anaesthesia is not common practice.

#### CVS

There are two ways of performing a CVS (the choice of route will depend on the position of the placenta and experience of the operator):

1. Transabdominal route (used most commonly)  
A local anaesthetic injection is used to numb the abdomen. A needle is pushed through the abdomen and then a syringe is used to withdraw a small amount of placental tissue.
2. Transcervical route (rarely performed)  
A speculum is inserted into the vagina and fine forceps or a cannula is inserted through the cervix to the placenta. A small amount of placental tissue is removed, using either forceps or a suction catheter.

After both procedures the woman may be asked to stay in the clinic for about half an hour to rest. Occasionally the procedure cannot be performed on the day of

the appointment due to the position of the baby in an amniocentesis or the position of the placenta or insufficient placental tissue in a CVS. The woman will be offered another appointment.

### **Discomfort during the procedure**

Some women may feel mild cramp like pain, or pressure during the procedure or they may feel no discomfort at all. The amount of discomfort or pain varies among women and from one pregnancy to the next.

Some women may experience mild cramps (like period pains) after the procedure which should subside after 48 hours.

### **Length of the procedure**

#### **Amniocentesis**

The whole procedure takes about 10 minutes, but this depends on the position of the baby, the amount of amniotic fluid and the location of the placenta.

#### **CVS**

The whole procedure takes about 20 minutes. The sample will be examined to see if there is enough placental tissue to do the test. If not, another sample will be taken. Sometimes CVS may not be performed on the day because it may not be possible to safely access the placenta. This may be due to placental position. Allowing extra days for the placenta to grow, may then make it possible at a later date.

### **Does the procedure harm the baby?**

#### **Amniocentesis**

Direct injury to the baby from amniocentesis is very rare with continuous ultrasound guidance. The doctor will avoid placing the needle near the baby however, it is difficult to stop the baby moving towards the needle. The aspiration needle is fine and pliable.

#### **CVS**

Performing CVS after 10 completed weeks of pregnancy has decreased the risk of harm to the baby.

### **Bringing a partner or friend**

A partner, friend, or a family member can be with a woman during the procedure. It is not advisable to bring children, as child care facilities are not provided.

### **Eating and drinking**

A woman will be able to eat and drink as normal before and after the procedure.

### **Miscarriage rate**

#### **Amniocentesis**

The risk of causing miscarriage by amniocentesis, derived from research studies, is about 1% (one in 100 procedures).<sup>2</sup>

#### **CVS**

The risk of miscarriage is about 2% (one in 50 procedures) following CVS.<sup>3</sup> This is higher than the post amniocentesis miscarriage rate. However, the background risk of miscarriage is higher in early pregnancy which may contribute to the higher miscarriage rate for CVS.

Local or individual data is of variable quality and the number of procedures performed may not be large enough to reach statistical significance and should not usually be used, particularly if women are seeking reassurance that the miscarriage rate is lower than quoted above.

## Causes of miscarriage and when it is more likely to occur

### Amniocentesis and CVS

The exact cause of the miscarriage following an amniocentesis or CVS is unknown. Miscarriage following the procedures is thought to be due to infection or bleeding but the mechanisms are not proven.

There is currently no evidence to answer the question frequently asked by women regarding if or when a post amniocentesis miscarriage is likely to occur.

When miscarriages happen clinical experience suggests that miscarriage occurs up to two weeks following the procedure and that the risk diminishes after three weeks. Pregnancy outcomes following amniocentesis and CVS are currently being audited, and more information on this issue may become available.

### Activities to prevent miscarriage occurring

It is not possible for the woman to prevent a miscarriage after amniocentesis or CVS. Although there is no evidence, some doctors advise that women should take things easy for a couple of days, avoid intercourse, any heavy lifting or strenuous exercise. Resting in bed is not necessary.

Women are advised to arrange for someone to drive them home after the procedure.

## Cytogenetic results

### The difference between Polymerase Chain Reaction (PCR) marker analysis and karyotyping

Unlike karyotyping, PCR marker analysis is a test that does not require cell culture. PCR results can therefore be obtained quickly. The PCR test usually only looks for defined chromosome problems in the baby; Down's syndrome (Trisomy 21), Edwards' syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13). Normally there are two copies of each chromosome however, with these syndromes there is an extra copy of the chromosome in each cell. If monosomy X (Turner's syndrome) is suspected an additional PCR test analysis is done in the laboratory to look for the sex chromosomes.

The PCR result is usually available within three working days. Occasionally karyotyping results from CVS may be inconclusive because of an abnormal cell line confined to the placenta. This may then require further testing by amniocentesis later in the pregnancy.

### PCR result

This test is very accurate, but can only give information about the chromosomes being tested. A result from a rapid test will confirm whether the baby either does or does not have Down's syndrome, Patau's syndrome or Edwards' syndrome. However, other changes involving other chromosomes can occur. Between 2 and 5%, (two to five out of a hundred, depending on the laboratory) of amniotic fluid PCR samples fail to yield a reportable result, mainly because of maternal cell contamination of the amniotic fluid. More accurate figures should be available from the local laboratory.

### Karyotype result

Karyotyping involves culturing or growing the cells floating in the amniotic fluid before they can be examined under the microscope. The cells take about 10 days to grow in the laboratory.

It therefore takes longer to get a result. The test looks at changes in the number and appearance of all the chromosomes. The result from the karyotype test usually comes back within two to three weeks.

On occasions even though the PCR does not produce a result which indicates trisomy 21, 18 or 13, there could still be another abnormality in the chromosome make-up

that is picked up on karyotyping (if karyotype is offered locally).

## **Does a normal karyotype mean there is nothing 'wrong' with the baby?**

Some chromosome changes are so small that they are invisible even when viewed under the microscope.

The karyotype test will not detect:

- alterations in single genes, such as cystic fibrosis (each chromosome contains thousands of genes);
- microdeletions (loss of small segments of a chromosome); or
- other small changes in chromosomes.

A 'normal' chromosome result therefore means that the baby appears to have normal chromosomes, but this does not rule out all abnormalities.

A baby's physical development is not shown by this test. The 18 to 20<sup>+6</sup> week fetal anomaly scan is performed to detect structural anomalies in the baby however, a scan does not detect all structural abnormalities.

## **Trisomy 21**

- Trisomy 21 or Down's syndrome is the most common chromosome abnormality. Down's syndrome occurs when there is an additional chromosome 21.
- Babies with Down's syndrome can have multiple problems such as heart defects, and severe learning difficulties. Some people with Down's syndrome are able to lead semi-independent lives.
- The birth incidence of this condition in an unselected population is about 1 in 560 live births.

## **Severity of the condition**

Neither the karyotype nor PCR test can tell how severe the condition will be. An ultrasound scan at 18 to 20<sup>+6</sup> weeks may detect physical abnormalities that are associated with Down's syndrome such as heart defects.

## **Trisomy 18**

- Trisomy 18 or Edwards' syndrome is a very rare chromosome abnormality. It is less common than Down's syndrome. Edwards' syndrome occurs when there is an additional chromosome 18.
- Babies with Edwards' syndrome can have multiple problems such as heart defects, kidney problems and severe developmental delay. Life expectancy for the majority of babies with Edwards' syndrome is usually limited to a few weeks and rarely beyond one year of life.
- The incidence of this condition is about 1 in 3000 live births.

## **Trisomy 13**

- Trisomy 13 or Patau's syndrome is a very rare chromosome abnormality. It is less common than Down's syndrome. Patau's syndrome occurs when there is an additional chromosome 13.
- Babies with Patau's syndrome can have multiple, severe problems such as heart defects, brain abnormalities and severe kidney abnormalities.
- Most babies do not live beyond the first weeks of life and few survive beyond one year of life.
- The incidence of this condition is 1 in 6000 live births.

## **Sickle Cell and Thalassaemia**

Sickle cell disorders and thalassaemia major are serious inherited blood conditions. They affect the haemoglobin in the red blood cells. If both parents have the sickle cell or thalassaemia gene, there is a high chance (one in four, or 25%) that their baby will have a sickle cell disorder or thalassaemia major. Amniocentesis or CVS procedures will be offered in the antenatal period (as early as possible) to detect if the baby has a sickle cell or thalassaemia disorder.

## **Results**

The woman should be informed when she is having the procedure about the local arrangements for handling results.

## **Additional Information**

### **Amniocentesis and Chorionic Villus Sampling – Policy, Standards and Protocols**

and

### **Amniocentesis Information Leaflet for Women**

and

### **CVS Information Leaflet for Women**

Available from:

Antenatal Screening Wales

18 Cathedral Road

Cardiff

CF11 9LJ

Tel: 02920 787837

Website: [www.antentalscreening.org](http://www.antentalscreening.org)

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

73, Charlotte Street

W1T 4PN, London

Helpline 0207 631 0285

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

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

### **Down's Syndrome Association National Office**

Down's Syndrome Association

Langdon Down Centre

2a Langdon Park

Teddington

TW11 9PS

Tel: 0845 230 0372

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Email: [info@downs-syndrome.org.uk](mailto:info@downs-syndrome.org.uk)

Website: [www.downs-syndrome.org.uk](http://www.downs-syndrome.org.uk)

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## References

<sup>1</sup> Royal College of Obstetricians and Gynaecologists (2005) Amniocentesis and Chorionic Villus Sampling, Guideline No.8, Available from:  
[http://www.rcog.org.uk/resources/Public/pdf/amniocentesis\\_chorionicjan2005.pdf](http://www.rcog.org.uk/resources/Public/pdf/amniocentesis_chorionicjan2005.pdf)  
[Accessed on 08-04-08]

<sup>2</sup> RCOG (2006) Amniocentesis; what you need to know. Available from:  
[http://www.rcog.org.uk/resources/public/pdf/amniocentesis\\_patient\\_information\\_a.pdf](http://www.rcog.org.uk/resources/public/pdf/amniocentesis_patient_information_a.pdf)  
[Accessed on 08-04-08]

<sup>3</sup> RCOG (2006) Chorionic Villus sampling (CVS): what you need to know. Available from:  
<http://www.rcog.org.uk/index.asp?PageID=1557> [Accessed on 08-04-08]