Chorionic villus sampling (CVS) & amniocentesis

Information for health professionals
Chorionic villus sampling (CVS) and amniocentesis information for health professionals

This information sheet is intended to provide a guide for health professionals who are not usually involved in chorionic villus sampling (CVS) and amniocentesis procedures but who may be asked questions by women about these procedures. CVS is sometimes also called chorionic villus biopsy or placental biopsy (CVB or PB).

Summary

Chorionic villus sampling (CVS) and amniocentesis are invasive diagnostic procedures carried out during pregnancy. They are usually offered to detect chromosomal disorders such as Down's syndrome (Trisomy 21), but are sometimes used for single-gene conditions such as sickle cell disease, thalassaemia major or other rare conditions.

It is important to remember that women can choose whether or not to undergo these procedures. As health professionals, you must ensure that they understand the risks that these procedures carry to the fetus.

It is good clinical practice to obtain formal written consent for amniocentesis or CVS before the procedure. Written or verbal information should include the reason for offering the invasive procedure, an explanation of the procedure and the cytogenetic results that will become available (Royal College of Obstetricians and Gynaecologists 2010).

It is important to know the woman's HIV, rhesus and hepatitis B status before undertaking either of these invasive procedures.

Both CVS and amniocentesis should be performed under continuous ultrasound guidance. Although the procedure itself usually takes about 10 minutes to perform, the appointment may need to be longer to allow for discussion and because the woman may need to rest afterwards.
<table>
<thead>
<tr>
<th>What does it involve?</th>
<th>CVS</th>
<th>Amniocentesis</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>A needle is passed through the abdomen and into the placenta under continuous ultrasound guidance. Small fragments of placental tissue are then aspirated. CVS is usually performed transabdominally (TA). On rare occasions, however, the procedure cannot be done using the TA route, either because the placenta is posterior or the uterus is retroverted. Alternative options are to offer transcervical (TC) CVS or wait until an amniocentesis can be done.</td>
<td>A needle is passed through the abdomen and into the amniotic sac under continuous ultrasound guidance. A small sample of amniotic fluid (15–20mL) is then removed. Amniocentesis is performed transabdominally (TA).</td>
</tr>
</tbody>
</table>

| When is the safest time to carry out the procedure? | CVS can be performed between 10 and 15 weeks of pregnancy. It is most commonly carried out between 11 and 14 weeks. It can be performed later in pregnancy. | Amniocentesis is usually performed after 15 weeks of pregnancy. It is most commonly performed between 15 and 18 weeks of pregnancy. It can be performed later in pregnancy. |

| What is the risk of miscarriage? | The risk of causing miscarriage by CVS is about 1% but the total miscarriage rate following CVS is between 1 and 2%. | The risk of causing miscarriage by amniocentesis is about 1%. |

A ‘normal’ chromosome result means that the fetus appears to have normal chromosomes but does not rule out all abnormalities. The physical development of the fetus is not shown by this test. The 18th to 20th weeks Fetal Anomaly ultrasound scan is performed to look at the physical development and to detect structural anomalies in the fetus. A scan cannot, however, detect all structural abnormalities.
1. Introduction

This information sheet is intended to provide a guide for health professionals not usually involved in chorionic villus sampling (CVS) and amniocentesis procedures but who may be asked questions by women about these procedures. CVS is sometimes also called chorionic villus biopsy or placental biopsy (CVB or PB).

Parents requesting such a procedure because of their genetic family history should be referred for prenatal diagnosis (PND) counselling. This counselling is generally carried out by an obstetrician/fetal medicine specialist, screening midwife or specialist midwife (fetal medicine) or a clinical geneticist.

2. Why is CVS or amniocentesis performed?

CVS and amniocentesis are invasive diagnostic procedures carried out during pregnancy. They are usually offered to detect chromosomal disorders such as Down’s syndrome (Trisomy 21). When used for rarer indications, such as single-gene conditions (e.g. sickle cell disease, and thalassaemia major), clinical management policies should be followed, which will usually include establishing both the parents’ genetic status before the procedure.

3. Diagnostic testing in multiple pregnancies

A high level of expertise in ultrasound scanning is essential for operators undertaking amniocentesis or CVS in multiple pregnancies (Royal College of Obstetricians and Gynaecologists 2010).

4. HIV, rhesus and hepatitis B status

It is important to know the woman’s HIV, rhesus and hepatitis B status before undertaking either amniocentesis or CVS.

The woman’s rhesus status should be stated on the referral form. It is vitally important that women who are rhesus D negative are offered and given, where consent obtained, an appropriate amount of anti-D immunoglobulin according to their gestation to reduce the risk of rhesus iso-immunisation (refer to your local Trust policy). The anti-D immunoglobulin should be given after the procedure in line with national guidance (Royal College of Obstetricians and Gynaecologists 2010).

Knowledge of the woman’s HIV and hepatitis B status is important to minimise the risk of transmission of the virus to the unborn fetus and to ensure that appropriate precautions are taken by hospital and laboratory staff. If women have declined screening for bloodborne viruses, the potential risks of infection to the fetus if positive should be discussed and that discussion documented (Royal College of Obstetricians and Gynaecologists 2010).
5. Consent

Women who are offered CVS or amniocentesis should be given as much information as they require about the purpose, risks, benefits and limitations of the procedures before making a decision about whether or not they want to have the tests. The information should be given to them by a healthcare professional with experience and knowledge of prenatal diagnosis. It is good clinical practice to obtain written consent for amniocentesis and CVS before the procedure (Royal College of Obstetricians and Gynaecologists 2010). For more information on consent, please consult the NHS Fetal Anomaly Screening Programme consent standards available at www.fetalanomaly.screening.nhs.uk.

6. Results

During the appointment, the woman should be informed about the local arrangements for handling results and options for how she will be informed. All staff involved in this process must obtain up-to-date information on their local arrangements.

7. Ultrasound scan

Prior to any invasive procedure, a scan is required to determine the fetal number, fetal viability, amniotic fluid volume, fetal position and placental position and to confirm gestational age.

During the procedure, the ultrasound probe will be used to:

- identify a target site to obtain an optimal pocket of amniotic fluid or placental tissue;
- track the sampling needle to reduce the likelihood of trauma to the fetus, cord and uterus.

8. When are the procedures performed?

**CVS**

CVS can be performed between 10 and 15 weeks of pregnancy. It is most commonly carried out between 11 and 14 weeks. It can be performed later in pregnancy.

It is important to refer individuals with a family history of single-gene disorders early, so that the parents have the option of first trimester CVS. Where the risk of the unborn fetus being affected is 1 in 2 or 1 in 4 and where there is a substantial risk that the parents will face the option of termination, it may be important for them to have the test and result as early as possible.
### Amniocentesis

Amniocentesis is usually performed after 15 weeks of pregnancy. It is most commonly performed between 15 and 18 weeks. It can be performed later in pregnancy. From 15 weeks of pregnancy, fetal cells are greatest in number and the amount of amniotic fluid is around 150–200mL and therefore adequate for obtaining a 15–20mL cytogenetic sample. Prior to 15 weeks, the rate of miscarriage from this procedure is higher and obtaining the correct volume of amniotic fluid tends to be difficult.

### 9. What happens during the procedures?

The procedures involve removing small amounts of either placental tissue (CVS) or amniotic fluid (amniocentesis). They are undertaken by a clinician with specialist training.

An ultrasound scan is performed prior to the CVS or amniocentesis procedure to check for fetal position and viability and to localise the placenta. This will identify the optimum needle position for taking:

- a sample of placental tissue (CVS) or
- a sample of amniotic fluid (amniocentesis).

Both procedures should be performed under continuous ultrasound guidance.
CVS

CVS is usually performed transabdominally (TA) between 11 and 14 weeks. On rare occasions, however, the procedure cannot be done using the TA route, either because the placenta is posterior or the uterus is retroverted. Strategies such as asking the woman to fill and empty her bladder or waiting another week may be helpful, but if this fails, alternative options are to offer transcervical (TC) CVS or wait until an amniocentesis can be done. This should be fully discussed with the woman to ensure all options are provided along with any associated risks.

It is important to note that the risk of miscarriage associated with TC CVS is about 1–2% and, given the choice, some women may opt for an amniocentesis which has a slightly lower risk of miscarriage (1%) (Royal College of Obstetricians and Gynaecologists 2010).

Local anaesthetic may be given prior to the procedure.

Immediately before the procedure, the woman's abdomen is cleaned to ensure that the CVS takes place in the most sterile conditions possible.

During the TA procedure, a needle is passed through the abdomen and into the placenta under continuous ultrasound guidance. Small fragments of placental tissue are then aspirated.

After the procedure, the woman may wish to rest for a while in the clinic.

Amniocentesis

Amniocentesis is always performed transabdominally (TA). Although a local anaesthetic may be given prior to the procedure, research suggests that most women will not notice a beneficial analgesic effect and so the procedure is usually done without the use of anaesthetic.

Immediately before the procedure, the woman's abdomen is cleaned to ensure that the amniocentesis can take place in the most sterile conditions possible.

During the procedure, a needle is passed through the abdomen and into the amniotic sac under continuous ultrasound guidance. The needle stilette is removed once the needle is in the correct position.

A small sample of amniotic fluid (15–20mL) is then removed using a syringe attached to the needle. The amniotic fluid in the uterus replenishes quickly (the next time the fetus passes urine).

After the procedure, the woman may wish to rest for a while in the clinic.
10. Discomfort during the procedure

Discomfort either during or after the procedure varies from woman to woman and in each pregnancy. Some of the sensations women have reported include:

- period type pain during or after the procedure,
- a sharp, stinging sensation when the sample needle is inserted into the abdominal skin,
- pressure-type feeling either in the lower abdomen or in the lower back or vagina as the needle is removed.

11. Length of the procedure

CVS

The whole procedure usually takes about 10 minutes. An appointment for CVS will sometimes take slightly longer than amniocentesis because, although the procedures are similar, more elaborate preparation may be needed for CVS.

Amniocentesis

The whole procedure usually takes about 10 minutes. The time taken can depend on the position and movements of the fetus and whether this hinders access to amniotic fluid pools.

12. Post procedure care

Agreement should be reached between the healthcare professional and the woman about how the test results will be given. The healthcare professional should check that the woman's correct telephone number is recorded on the hospital system and that she has been given the healthcare professional's contact details.

The woman should also be advised who to contact in case of any complications post procedure. If she experiences one or more of the following signs or symptoms, she should contact a hospital or a healthcare professional:

- Feeling generally unwell (shivery, nauseous, abdominal discomfort)
- Pyrexia
- Persistent bleeding from the vagina
- Persistent lower abdominal/back pain
- Clear watery type loss (not urine) from the vagina
- Offensive smelling discharge from the vagina.
13. Barriers to the completion of the procedure

Occasionally the procedure cannot be performed on the day of the appointment for a variety of reasons. In these cases, the woman should be offered another appointment.

Occasionally, in either procedure, an adequate sample may not be obtained.

The cytogenetic laboratory may also fail to obtain a result despite receiving an adequate sample. This is relatively unusual, but if it does happen a further or alternative procedure will be recommended.

As a health professional, you should find out from your local service the failure rate for this procedure.

14. Bringing a partner or friend

The woman may want a partner, a friend or a family member to attend the appointment with her.

15. Eating and drinking

Most hospitals advise that it is safe to eat and drink as normal before and after CVS or amniocentesis providing the pregnancy is under 24 weeks gestation.
16. Do the procedures harm the fetus?

Direct injury to the fetus from either CVS or amniocentesis is very rare because continuous ultrasound guidance is used. It is not possible, however, to prevent the fetus moving towards the needle during an amniocentesis.

Miscarriage rate after CVS or amniocentesis

Because CVS and amniocentesis are invasive procedures, miscarriage is a possible complication. The risk of causing miscarriage by CVS is about 1% but the total miscarriage rate following CVS is between 1 and 2%. The risk of causing miscarriage by amniocentesis is about 1%.

Although the procedure-related risks are similar, the post CVS miscarriage rate is higher than the post amniocentesis miscarriage rate because the background risk of miscarriage (whether the procedure was done or not) is higher in early pregnancy when a CVS is normally carried out.

These miscarriage rates are derived from national clinical audits. Local or individual data are of variable quality and the number of procedures performed may not be large enough to reach statistical significance and should not usually be used. This is particularly relevant if a woman is seeking reassurance that the miscarriage rate is lower than quoted above.

Cause of miscarriage after CVS or amniocentesis

The exact cause of miscarriage following a CVS or amniocentesis is unknown. It is possible that miscarriage following either procedure could be due to ruptured amniotic membranes, infection or bleeding.

Women offered a CVS or amniocentesis will often have risk factors for miscarriage such as maternal age, abnormal blood tests or abnormal scan findings. It is therefore difficult to estimate the background risk of pregnancy loss for the groups choosing to have invasive tests.

Miscarriages that would have occurred even if the procedure had not been undertaken will still occur. Pregnancy outcomes following CVS and amniocentesis should be audited locally and more information on this issue may become available in the future.

Reducing the risk of miscarriage

Clinical experience suggests that miscarriage as a consequence of the procedure can occur up to two weeks following the procedure and that the risk diminishes after three weeks.

Some doctors advise that women should rest for a couple of days post procedure, avoiding intercourse, any heavy lifting or strenuous exercise. There is no evidence to support this. Resting in bed is not necessary. It is considered good practice to advise women to arrange for someone to drive them home after the procedure.
17. Cytogenetic results

What is the difference between QF-PCR and karyotyping?

Quantitative fluorescent polymerase chain reaction (QF-PCR) marker (‘rapid’ test analysis) and karyotyping are two types of cytogenetic tests. QF-PCR marker analysis is a laboratory process that does not require cell culture whereas karyotyping does. PCR results can therefore be obtained more quickly.

PCR testing usually only looks for three specific defined chromosome conditions in the fetus: Trisomy 21 (Down’s syndrome), Trisomy 18 (Edwards’ syndrome) and Trisomy 13 (Patau’s syndrome). Normally there are two copies of each chromosome, but if these syndromes are present there is an extra copy of that chromosome in each cell. If monosomy XO (Turner’s syndrome) is suspected from the scan, a PCR test will be performed to look at the sex chromosomes.

The PCR result is usually available within 3 working days (the national target).

Occasionally, karyotyping results from CVS may be inconclusive because of an abnormal cell line confined to the placenta. This is referred to as ‘confined placental mosaicism’. Further testing by amniocentesis may be needed later in the pregnancy to investigate this result. The chance of a confined placental mosaicism is approximately 1–2% (Lestou and Kalousek 1998).

QF-PCR result

QF-PCR is very accurate, but can only give information about the specific chromosomes being tested. A result from a rapid test is nearly 100% accurate in confirming whether or not the fetus either does or does not have Trisomy 21, Trisomy 13 or Trisomy 18. Other chromosomes may be tested for, but this must be specifically requested and discussed with the relevant cytogenetic laboratory.

If another condition is suspected, a full karyotype can sometimes be offered. As a significant proportion of abnormal scans are associated with karyotypes other than these trisomies, full karyotyping is usually recommended when the indication is an abnormal scan.

A small number of amniotic fluid PCR samples fail to yield a PCR result, mainly because of maternal blood contamination of the amniotic fluid.

Full karyotype result

Karyotyping involves growing the fetal cells floating in the amniotic fluid and making a preparation showing the chromosomes, which are then examined under the microscope. The test looks at changes in the number and appearance of all the chromosomes.

The cells take about 5–10 days to grow in the laboratory. Results from karyotyping therefore take longer to obtain than results from PCR (usually 10–14 days).
Further guidance on cytogenetic testing can be found in the NHS FASP Working Standards document available from www.fetalanomaly.screening.nhs.uk/standardsandpolicies.

**Does a normal karyotype mean there is nothing ‘wrong’ with the fetus?**

Many serious diseases are not genetic and could never be found with a chromosome test. Not all serious genetic diseases will be detected by a chromosome test alone because some chromosome changes are so small that they may go unnoticed when viewed under the microscope. Some can be found with special tests if it is known what to test for and if specifically requested.

The full 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 fetus appears to have normal chromosomes but does not rule out all abnormalities.

The physical development of the fetus is not shown by this test. The 18<sup>th</sup> to 20<sup>th</sup> weeks Fetal Anomaly ultrasound scan is performed to detect structural anomalies in the fetus. However, a scan cannot detect all structural abnormalities either.

It is a good idea for health professionals assisting in these procedures to gain more information from the relevant genetic departments.
18. Abnormal results

A small number of women will unfortunately receive abnormal test results and those few will need to make choices about the treatment path they want to follow.

If the results show that the fetus has one of the disorders being tested for, the woman and her partner should be given the opportunity to discuss this fully with the specialist midwife, obstetrician/fetal medicine specialist or clinical geneticist as soon as possible.

After an abnormal result, the woman may also be referred to a consultant paediatrician, consultant geneticist or genetic counsellor for further information and counselling. Although some disorders are treatable, others are not.

Whatever the outcome, the woman must feel assured that the health professionals caring for her will support her decision to:

- continue the pregnancy and use the information received to help prepare for the birth and care of her baby;
- continue the pregnancy and consider adoption; or
- terminate the pregnancy.

Some women who make an informed decision to terminate their pregnancy find it helpful to talk with a health professional or a counsellor about their experience afterwards.

It may also be helpful to recommend the support groups listed at the end of this leaflet. These are also available in the parent information leaflets ‘Amniocentesis test – information for parents’ and ‘Chorionic villus sampling (CVS) – information for parents’ available from www.fetalanomaly.screening.nhs.uk/publicationsandleaflets.

Websites maintained by these support groups often have useful information about the various conditions and describe the experiences of women faced with difficult choices following diagnosis of chromosome and other abnormalities.

If a woman decides to have a termination of pregnancy for an abnormal result, she may have the choice of having either a surgical or medically induced termination. The method selected will depend, however, on the nature of the anomaly and how advanced the gestation is.
19. Examples of chromosome abnormalities that can be detected using CVS or amniocentesis

Trisomy 21 (Down’s syndrome)

- Trisomy 21 is the most common chromosomal condition detected.
- Trisomy 21 occurs when there is an additional chromosome 21.
- Babies with Trisomy 21 can have multiple problems such as cardiac defects and severe learning difficulties. However, some adults with Trisomy 21 are able to lead semi-independent lives.
- The birth incidence of this condition in an unscreened population is about 1 in 560 live births, but is strongly linked to maternal age and is therefore different in different geographical areas.
- As women get older, the risk of having a pregnancy affected with Trisomy 21 becomes greater. However, since women under the age of 35 have more children, most Trisomy 21 affected babies are born to younger women.

Neither karyotyping nor the PCR test can predict how severe the condition will be in any given individual. An ultrasound scan at 18 to 20 weeks may detect physical abnormalities that are associated with Trisomy 21, such as cardiac defects, but should not be used as the primary screening test.

Trisomy 18 (Edwards’ syndrome)

- Trisomy 18 is a rare chromosome abnormality. It is less common than Trisomy 21.
- Trisomy 18 occurs when there is an additional chromosome 18.
- The incidence of Trisomy 18 at birth is 1 in 1500.
- Babies with Trisomy 18 can have multiple problems such as cardiac defects, renal abnormalities and severe developmental delay. Many affected fetuses miscarry or die during later pregnancy, but when born alive, life expectancy for the majority of babies with Trisomy 18 is usually limited to a few weeks and rarely beyond one year.

Trisomy 13 (Patau’s syndrome)

- Trisomy 13 is a rare chromosome abnormality. It is less common than Trisomy 21.
- The incidence of this condition is 1 in 3000 live births.
- Trisomy 13 occurs when there is an additional chromosome 13.
- Most affected fetuses miscarry; liveborn babies with this condition usually do not live beyond the first weeks of life and few survive beyond one year.
- Babies with Trisomy 13 can have multiple, severe problems such as cardiac defects, brain abnormalities and severe renal abnormalities. Some of these abnormalities can be seen on ultrasound scan from 11 weeks onwards.
Sickle cell and thalassaemia

Sickle cell disease is a variable set of inherited conditions affecting the haemoglobin that can cause chronic anaemia, jaundice, acute pain (crisis), organ damage, infections and strokes in children and adults.

Alpha and beta thalassaemia major are inherited blood conditions that affect the quantity of haemoglobin produced. Alpha thalassaemia major is incompatible with life. Beta thalassaemia major results in severe anaemia. Inheritance of an altered gene from both parents results in a disorder and inheritance of only one altered gene results in a healthy carrier.

All pregnant women receiving abnormal haemoglobinopathy results should receive specialist obstetric and haematological advice.

More information on sickle cell diseases and thalassaemia major is available from the NHS Sickle Cell & Thalassaemia Screening Programme at www.sct.screening.nhs.uk.

Other conditions

There are numerous other abnormal results which will rarely be obtained, usually as a result of sophisticated testing for specific conditions. All abnormal results should be given and explained by specialists, such as those in Fetal Medicine Units or Clinical Genetics Units.
20. Further information, charities and support groups

Antenatal Results and Choices (ARC)
73 Charlotte Street
London W1T 4PN
Helpline: 0207 631 0285
Email: info@arc-uk.org
Website: www.arc-uk.org

Antenatal Results and Choices (ARC) provides information and support to parents before, during and after antenatal screening and diagnostic tests, especially those making difficult decisions about testing, or about continuing or ending a pregnancy after a diagnosis. ARC offers ongoing support whatever decisions are made.

Down’s Syndrome Association (DSA)
Website: www.downs-syndrome.org.uk
Langdon Down Centre
2a Langdon Park
Teddington
TW11 9PS
Phone: 0845 230 0372
Email: info@downs-syndrome.org.uk

The aim of the Down’s Syndrome Association (DSA) is to help people with Down’s syndrome lead full and rewarding lives.

The Miscarriage Association
17 Wentworth Terrace
Wakefield WF1 3QW
Tel: 01924 200799
Email: info@miscarriageassociation.org.uk
Website: www.miscarriageassociation.org.uk

The Miscarriage Association is a registered national charity in England & Wales (no. 1076829) and in Scotland (no. SC039790) and a company limited by guarantee (no. 3779123), working across England, Northern Ireland, Scotland and Wales. It was founded in 1982 by a group of people who had experienced miscarriage and continues to offer support and information to anyone affected by the loss of a baby in pregnancy, to raise awareness and to promote good practice in medical care.
The Health Experience Research Group has created a unique database of personal and patient experiences through in-depth qualitative research into over 60 different illnesses and health conditions. The results of their research are published on two websites (www.healthtalkonline.org and www.youthhealthtalk.org) which are aimed at patients, their carers, family and friends, doctors, nurses and other health professionals. Their target is to complete at least 100 conditions within the next 5–10 years.

They have also recently started two social networking sites for people to add their own experiences of health and illness at www.myhealthtalk.org and www.myyouthhealthtalk.org. The websites are run by the DIPEX Charity.

---

**Sickle Cell Society**

54 Station Road  
London NW10 4UA  
Tel: 0208 961 7795  
Email: info@sicklecellsociety.org  
Website: www.sicklecellsociety.org

The Sickle Cell Society provides information, counselling and care for people with sickle cell disorders and their families.

---

**UK Thalassaemia Society**

Website: www.ukts.org  
19 The Broadway  
Southgate Circus  
London  
N14 6PH  
Phone: 020 8882 0011  
Email: office@kuts.org  

The Society raises the awareness and the health education of the communities of the UK at risk of Thalassaemia.
21. Sources and references


Royal College of Obstetricians and Gynaecologists (2005) Amniocentesis and chorionic villus sampling. London: RCOG.


