Amniocentesis and Chorionic Villus Sampling – UHDB Full Clinical Guideline - Service provided at RDH site only

Reference No.: UHDB/AN/01:21/A4

Contents

1. Introduction

Amniocentesis and Chorionic Villus Sampling (CVS) are invasive prenatal diagnostic tests which are offered to pregnant women who have an increased chance of a chromosomal or genetic disorder.

Amniocentesis is performed from 15+0 weeks of pregnancy onwards, by inserting a needle to obtain a sample of amniotic fluid.

Chorionic Villus Sampling (CVS) is usually performed from (11+0) to (13+6) weeks of pregnancy. Trans-abdominal CVS is performed by inserting a needle into the placenta to obtain a sample of placental villi.

Both procedures have a risk of causing a miscarriage, and a small risk of maternal infection.

2. Purpose and Outcomes

To outline safe procedures for Amniocentesis and CVS, based on approved national guidance for invasive prenatal procedures (RCOG,2021, NHSFASP 2008), and so minimise the risk of miscarriage and maternal infection.

It is recognised that "For the woman and her family, good care in these circumstances encompasses more than the simple performance of a technique".

Clear guidance is included to ensure that all health care professionals are aware of the appropriate care, advice and information to be given to these women, at a time when they are likely to be particularly anxious.

3. Abbreviations

ARC	Antenatal Results and Choices
AVSD	Atrioventricular Septal Defect
CF	Cystic Fibrosis
CMV	Cytomegalovirus
CPM	Confined Placental Mosaicism
CRIS	Computerised Radiography Information System
CVS	Chorionic Villus Sampling
DNA	Deoxyribonucleic Acid
DOH	Department of Health
EDTA	Ethylenediaminetetraacetic Acid
FISH	Fluorescence In-Situ Hybridisation
FMMC	Fetal Maternal Medicine Centre
HIV	Human Immunodeficiency Virus
iCM	ISOFT Clinical Manager
IU	International Units
MHHR	Maternity Hand-Held Records
mls	Millilitres
NHSFASP	National Health Service Fetal Anomaly Screening Programme
NICE	National Institute for Health and Clinical Excellence
NSC	UK National Screening Committee
NT	Nuchal Translucency
XO	Indicates single X chromosome as in Turners syndrome
PAU	Pregnancy Assessment unit
PCR	Polymerase Chain Reaction
PV	Per Vagina
QF-PCR	Quantitative Fluorescence – Polymerase Chain Reaction
RCOG	Royal College of Obstetricians and Gynaecologists
UPD	Uniparental Disomy

4. Key Responsibilities and Duties

Amniocentesis and CVS are performed by a Fetal Medicine Consultant.

Consent for the procedure is obtained by a Fetal Medicine Consultant. A Fetal Medicine Midwife assists during the procedure, communicates the results to the woman and is responsible for maintaining a database of the clinical details of all women who undergo Amniocentesis and CVS.

5. Indications for offering Amniocentesis or CVS

Amniocentesis and Chorionic Villus Sampling (CVS) are offered to pregnant women who have an increased chance of a fetal chromosomal or genetic disorder. Indications to offer a diagnostic test include:-

- Increased risk of abnormality identified through antenatal screening for Down's, Patau and Edward syndrome (combined screening test or 2nd Trimester Serum Screening), using cut-off levels recommended by NHSFASP – currently chance of 1:150 or higher..
- Previous pregnancy affected with a chromosomal or genetic condition.
- Parents known carriers of a genetic condition.
- Family history of a genetic condition.
- Ultrasound scan shows certain fetal abnormalities which are associated with a chromosomal or genetic condition.
- Positive NIPT or NIPD results

6. Information to be given prior to the test

It is the woman's choice whether, or not, to have an invasive test. For the woman and her partner to make an informed choice, the risks and benefits of Amniocentesis and CVS should be discussed, including the following information:-

- Reason for offering the test
- An explanation of the procedure
- Risks of the test (see below)
- What is tested for
- What is not tested for(structural anomalies and single gene disorders)
- Type and significance of results
- Expected timing of results and how these will be communicated
- Accuracy and limitations of laboratory testing

This discussion is backed up by giving written information (please see local booklet "Amniocentesis and Chorionic Villus Sampling-Information for you").

Women considering amniocentesis or CVS should receive detailed counselling and pregnancy mapping by suitably trained healthcare professionals.

7. Risks of the test

The additional risk of miscarriage is around 0.5% following amniocentesis and CVS (These are national figures). The additional miscarriage risk is for up to 2-3weeks after the test. This percentage increases to 1% for multiple pregnancies.
Late amniocentesis from 22 weeks, it may be appropriate to discuss that this risk may include a small chance of an extremely preterm live baby (rather than miscarriage) with inherent risks of disability, even if the test results are normal.
CVS has a 1% risk of a mosaic result which may lead to the offer of amniocentesis, to establish whether the baby has a mosaic karyotype or that there is Confined Placental Mosaicism (CPM).
There is a small risk (less than 1 in 1000) that amniocentesis or CVS may cause serious maternal infection.
There is a small risk of failure to obtain a result, (rapid or culture or both). This is more common when the procedure is done in the third trimester due to sub optimal quality fetal cells
There is a small risk of obtaining an equivocal result.

The risks associated with third-trimester diagnostic amniocentesis, including the risk of pre-term labour, are likely to be low.

8. Consent

Valid consent must be obtained (RCOG, 2021).
A consent form should be completed and signed by the woman, using DOH consent form 3. A copy of the form is offered to the woman.

9. Review of Blood Results

9.1 Blood Group

The woman's blood group is ascertained to determine whether Anti-D will be necessary.
If no result is available (either in the notes or by phoning Blood Bank) take maternal blood in a pink-top cross-match bottle and send to Blood Bank for grouping.
For rhesus negative women with a gestational age beyond 16 completed weeks, fetal rhesus genotyping results should be obtained to ascertain the need for Anti-D post-procedure. If no result or <16 weeks, a kleihauer should be undertaken 30 minutes after the procedure, and Anti-D administered as issued by blood bank.

9.2 Blood Borne Viruses

The woman's booking blood results are reviewed for the presence of blood borne viruses (HIV and Hepatitis B are currently offered routinely to all pregnant women).
If the woman is HIV positive, or Hepatitis B or C positive, an Amniocentesis or CVS might increase the chance of maternal-fetal transmission. This should be discussed with a Fetal Medicine Consultant.
Where screening results for blood borne viruses are not known, testing should be delayed until HIV status can be determined. The risk of mother to child transmission of HIV for women on highly active antiretroviral therapy is very low. Antiretroviral treatment should be optimised to aim for an undetectable vira lload prior to amniocentesis or CVS.

The risk of mother to child transmission of Hepatitis B is low with viral load less than 6.99 log10copies/ml but increases with higher viral loads.

There is no evidence of risk of mother to child transmission of Hepatitis C based on limited data available.

10. Amniocentesis Procedure

Aseptic technique is followed, including sterile gloves and a procedure pack. (Please see Appendix C - Infection Control Measures). Amniocentesis is performed by fetal medicine consultant,

- under simultaneous direct ultrasound control
- with continuous echogenic needle tip visualisation,
- a 22 G needle is inserted into the amniotic sac to obtain a sample of amniotic fluid,
- the amniotic fluid sample (ideally15 mls) is put into a specimen container.

A transplacental approach should only be used if a clear pool of amniotic fluid can only be reached by passage through the placenta.
If a second attempt is needed a new needle should be used and the number of needle insertions should be documented.
If the sample is blood stained this is documented and the woman advised that the rapid (QF-PCR) result is more likely to fail.
Maternal blood is taken (4 mls in EDTA bottle (purple top)) and sent to the lab with the amniotic fluid. This blood is used during the QF-PCR analysis to exclude maternal contamination.

Fetal viability should be confirmed and documented before and after the procedure.

If amniotic fluid appears cloudy or purulent or there are clinical features of intra-amniotic infection consider microbial analysis and antibiotic treatment

11. Trans-abdominal CVS Procedure

Aseptic technique is followed, including sterile gloves and a procedure pack, CVS is performed under direct ultrasound control with continuous echogenic needle tip visualisation. A sterile needle guide attached to the scan probe may be used.
The needle and syringe are heparinised prior to the procedure (to ensure the sample does not get clotted).The Fetal Medicine Consultant infiltrates the skin with local anaesthetic and inserts a needle (18G-double lumen) into the placenta to obtain a sample of placental villi.
The CVS sample is aspirated into the syringe and then placed in Cytogenetic Transport Medium in a sterile specimen container. (The medium is obtained from Cytogenetics and stored in the fridge).
The size of sample obtained is checked visually, and documented. (For Karyotype an optimal sample size of 10mcg or more of trophoblast is needed, if other analysis such as molecular genetics is also required then more is preferred).
If a second attempt is needed a new needle should be used and the number of needle insertions should be documented.
Fetal viability should be confirmed and documented before and after the procedure.

12. Diagnostic Tests In Multiple Pregnancies

In the case of multiple pregnancies, an Amniocentesis or CVS is performed by a specialist who has the expertise to subsequently perform a selective termination of pregnancy if required.
The uterine contents are “mapped” with care to ensure that separate samples are taken for each fetus and clearly labelled as such.

13. Request Form

The Cytogenetic request form is completed with the clinical details:-

- Date of the test
- Gestation
- Indication for the test
- Sample Type (Amniotic Fluid or CVS)
- Name of the Doctor performing the test
- Type of lab analysis required (e.g. state if require DNA to be stored)
- Whether the mother has had previous Cytogenetic testing
- Destination for an extra copy if needed (e.g. Clinical Genetics)
- State if other than FMMC will be giving results (e.g. Clinical Genetics)

14. ANALYSIS TO BE PERFORMED

QF-PCR

QF-PCR is the rapid test performed for both CVS and amniocentesis, and analyses chromosomes 13, 18, 21 and XY.

Molecular Genetic Analysis

If Molecular Genetic analysis for a specific single gene disorder (e.g. Cystic Fibrosis) is required, the details are written on the request form. Telephone Molecular Genetics (0115 9627743), on the

day of the test, to alert them to expect the sample and ask if maternal and/or paternal blood samples (EDTA bottle) are needed to accompany it.
If the required analysis is performed by a laboratory other than Nottingham (e.g. Oxford for Sickle Cell Disease) then Nottingham Cytogenetics will retain some of the sample to perform karyotype (if requested) and send forward the required material (either part of CVS or amniocentesis sample, or extracted DNA) to the other lab.
For these cases it may be necessary to liaise with Nottingham and the other receiving lab prior to booking the appointment for the test. It may be necessary to arrange the appointment Monday to Wednesday, to enable Cytogenetics to process and transport the sample onwards before a weekend.

Micro-array test analysis
A microchip based testing platform that allows high volume, automated analysis of many pieces of DNA at once. This is performed if fetal abnormalities have been detected on scan and following specific discussions with the patients by the fetal medicine consultant about the test.

Storage of samples
Sometimes a need for certain types of analysis only becomes evident subsequent to the Amniocentesis or CVS sample being sent to the lab. If this is the case, explain and offer the further analysis to the woman and phone Cytogenetics to request it. For example, if an Amniocentesis was performed at 15 weeks for raised Down's Syndrome screening, then on the anomaly scan, performed at 18 to 20+6 weeks gestation, echogenic bowel is seen necessitating the offer of testing for Cystic Fibrosis (CF).

The time scales for further testing are as follows:-

- DNA can be extracted from CVS cultures up to 2 weeks following the reporting date.
- Amniocentesis and CVS samples are fixed and stored for 8 months from the sample receipt (i.e. beyond the woman's EDD) in case further analysis becomes necessary.

E.g. Fluorescence In-Situ Hybridisation (FISH) for 22q deletion (for Di George Syndrome) if cardiac anomaly is later seen on scan.

15. Labelling of Samples

Hospital Identification labels are attached to the request form and the specimen container (which contains the Amniotic Fluid or CVS sample).
The woman is asked to confirm that the specimen container and request form are correctly labelled with her name and date of Birth.

16. Transportation of Samples

For details please see guideline for Antenatal and Postnatal Cytogenetic and Molecular Genetic Samples.

17. ANTI-D Immunoglobulin

If the woman is Rhesus D negative (unless already sensitised), or the genotyping reports a resus negative fetus, Anti D Immunoglobulin should be given as soon as possible following amniocentesis or CVS, always within 72 hours. A kleihauer should be performed 30 minutes post-procedure.

It is given intra-muscularly, ideally into the deltoid, with verbal consent.
Document the dose given in the Obstetric notes and Maternity Hand-Held Records (MHHR).
Complete the blood bank form and return one copy to them.

Procedure
30-40 minutes following the procedure take maternal blood in pink-top crossmatch bottle, if > 20 weeks gestational age add an EDTA sample, and send to Blood Bank.
Blood Bank will perform a kleihauer test to assess the volume of fetomaternal haemorrhage and so determine the dose of anti-D required to prevent sensitisation.
Blood bank will then issue the appropriate dosage (minimum 500IU) to be given.

18. Documentation

A Viewpoint report is completed by the Fetal Medicine Consultant (this report is also available on the iCM system).
Documentation includes a record of the clinical indication, gestation, analysis requested, needle gauge used, sample size, fetal viability before and after the procedure, number of uterine insertions and number of “bloody taps”.
The Fetal Medicine Midwife enters data on the Invasive Procedure audit and the procedures screen on Lorenzo. It is also documented on the antenatal review page of Lorenzo.

19. Advice Post-Procedure

(Please see local booklet “Amniocentesis and Chorionic Villus Sampling (CVS) Information for Women”).
The method of communicating the results is agreed between the woman and the Fetal Medicine Midwife. The woman may choose from the options of a telephone conversation (being phoned by or phoning in to the Fetal Medicine Midwife) and/or a face to face appointment with the Fetal Medicine Midwife. Results may be given to her partner if the woman agrees to this, and documented. Results may be given by Clinical Genetics for women known to their service, if previously agreed with them.
The Fetal Medicine Midwife checks that the woman’s telephone number is correct.
Following Amniocentesis or CVS the woman is encouraged to stay in FMMC to rest for 10 minutes.
Advise the woman that it is normal to experience abdominal tenderness, mild tightenings or mild period-type ache during, and for a few days after, the procedure. Advise her that it is safe to take Paracetamol for any discomfort.
Advise the woman to rest for a couple of days after the test and to avoid heavy lifting, strenuous exercise and sexual intercourse. There is no evidence to support this preventing miscarriage, but it is common practice to advise these precautions.
The woman is advised to be alert for, report and immediately seek medical advice for, any of the following symptoms: pain, contractions, PV loss, PV bleeding, pyrexia, flu-like symptoms. The woman is given the contact numbers for FMMC (in office hours) and PAU/GAU.

20. Communication of Results

Verbal communication of results between professionals (e.g. receiving results from Cytogenetics) involves the recipient repeating the results and the woman’s name with her date of birth or hospital number.
The Fetal Medicine Midwife will communicate the results to the woman in the manner previously agreed with her, as above and explains the significance and accuracy of the results (as below).
The woman is usually informed of the result within one working day of the result being received by FMMC.

Before communicating the results to the woman, check her name and use a second patient identifier such as phone number to ensure correct identity (e.g. if she phones in to FMMC ask her to confirm her date of birth).

Abnormal results

If the result is abnormal, obtain written confirmation of the result (by email) before communicating the result to the woman in the manner previously agreed with her.
The woman and her partner are then offered an appointment to come to FMMC to discuss the result with a Fetal Medicine Midwife or Fetal Medicine Consultant.
N.B. Abnormal QF-PCR results are confirmed by FISH analysis; therefore two reports will be received by email.
NB If CVS direct result shows Trisomy 18 or 13 Cytogenetics will perform FISH to check for hidden Mosaicism (i.e. direct may show T18, but culture result may be Mosaic T18).

Normal QF-PCR Result (rapid result from Amniocentesis and CVS)

The QF-PCR counts the number of specific chromosomes (21, 18 and 13, and XY).
Thus it tests for Trisomy 21 (Down's syndrome), Trisomy 18 (Edward's Syndrome), Trisomy 13 (Patau Syndrome), Triploidy (an extra copy of every chromosome) and sex chromosome differences
The QF-PCR result is usually available 2-3 working days following receipt of the sample, and is received in FMMC by emailfrom Cytogenetics or Molecular Genetics.
If the QF-PCR result is normal, the Fetal Medicine Midwife informs the woman that no extra number 21, 18 or 13 Chromosomes have been detected by this rapid analysis. (i.e. Down's Syndrome, Edward's Syndrome and Patau Syndrome have not been detected, and that the sex chromosomes are normal).
N.B. A result from QF-PCR is nearly 100% accurate in confirming whether or not the fetus does or does not have T21, 18 or 13.(Very rare, theoretical chance that a low level mosaic T21 in the baby could be missed on this initial result).

Microarray results a

CVS Mosaic result

CVS has a 1% risk of a mosaic result, which means that a combination of normal cells and cells with abnormal chromosomes was found in the sample from the placenta.
In this instance, the woman and her partner are informed and offered an appointment for discussion with a Fetal Medicine Midwife or Fetal Medicine Consultant. An amniocentesis may be offered to confirm that this is due to Confined Placental Mosaicism, rather than a mosaic karyotype in the baby.

Failure of results

There is a small risk of failure to obtain a result (rapid or culture or both). It may be possible for Cytogenetics to perform FISH analysis to try to obtain a rapid result.
A FISH probe for Trisomy 21 only will be used if the indication for the Amniocentesis or CVS was increased screening risk for Down's syndrome.
rapid result it is usual to await the culture result (which Cytogenetics will phone through to Fetal maternal medicine centre (FMMC).

Equivocal Results

Occasionally results are inconclusive or difficult to interpret.

In some cases, further tests may be offered to clarify interpretation of the result. If required, Cytogenetics will request this, either by phone to the Fetal Medicine Midwife or in the written report. In this situation further counselling is offered with a Fetal Medicine Midwife, Fetal Medicine Consultant or Clinical Geneticist, as appropriate.
For example if a chromosome rearrangement is found, parental Karyotyping (blood in a green-top lithium bottle) may be offered to establish if it is parental or de-novo in origin.

21. Monitoring Compliance and Effectiveness

The RCOG (2010) Green Top Guideline recommends continuous audit of the following standards:-

- Rate of pregnancy loss within 14 days of a procedure
- Local Cytogenetic laboratory culture failure rates for Amniocentesis and CVS
- Proportion of procedures requiring more than one needle insertion - operator specific
- Proportion of procedures with failure to obtain an adequate sample - operator specific
- Maintenance of a register of invasive diagnostic procedures
- Audit should be performed annually and the results made accessible to patients
- Rate of anti-D prophylaxis for women who are Rhesus negative undergoing amniocentesis or CVS
-

Monitoring method	The Fetal Medicine Midwives maintain a Database (located in the Fetal Medicine Shared Drive) of all Amniocentesis and CVS procedures performed. This database is used to enable audit of the above standards.
Report prepared by	Fetal Medicine Consultants and Fetal Medicine Midwives
Monitoring report sent to:	Maternity Development and Governance Committees
Frequency of report	As per agreed Audit forward programme

22. References

BCSH (2006) Guidelines for the use of prophylactic anti-D immunoglobulin. British Committee for Standards in Haematology (2006)

BCSH (2009) Guidance for the Estimation of Fetomaternal Haemorrhage. British Committee for Standards in Haematology (Sept 2009)

DOH (2009) Reference Guide to Consent for Examination or Treatment. DOH July 2009

NHSFASP (2009) Chorionic Villus Sampling (CVS) & Amniocentesis. Information for Health Professionals. NHSFASP July 2009

NSC (2007) Antenatal Screening –Working Standards for Down's Syndrome Screening. UK NSC April 2007

NHSFASP (2011) Consent Standards and Guidance. NHSFASP July 2011

NHSFASP (2008) Amniocentesis and Chorionic Villus Sampling. Policy, Standards and Protocols. NHSFASP April 2008

NICE (2008) Routine Antenatal Anti-D Prophylaxis for Women who are Rhesus D Negative. Technological Appraisal Guidance 156. NICE August 2008

RCOG (2021) Amniocentesis and Chorionic Villus Sampling. Green-top Guideline No.8 RCOG October 2021

RCOG (2011) The Use of Anti-D Immunoglobulin for Rhesus D Prophylaxis. Green-top Guideline No.22 RCOG March 2011

23. Sources Of Information And Support For Women

Leaflets

NHSFASP (2009)	Amniocentesis Test-Information for parents.
NHSFASP (2009)	Chorionic Villus sampling (CVS)-Information for parents.
RCOG (2011)	Information for you. Chorionic Villus sampling and Amniocentesis.

Websites

Antenatal Results and Choices	www.arc-uk.org
NHS choices	www.nhs.uk
NHSFASP	www.fetalanomaly.screening.nhs.uk

See: Local booklet “Amniocentesis and Chorionic Villus Sampling (CVS) Information for Women”

Appendix A

INFECTION CONTROL MEASURES

Nationally recommended infection control practices for amniocentesis and CVS (RCOG 2010 and NHSFASP 2008) are followed to reduce the risk of infection as below :-

General

Prior to the procedure all surfaces are cleaned with detergent wipes.
The couch to be covered with disposable paper roll which is disposed of after each patient and the surfaces wiped as above.
The area is kept clutter free with no storage of inappropriate equipment, to allow for easy cleaning
The procedure trolley is cleaned daily and just prior to the procedure the top/working surface is wiped with an alcohol spray.
The ultrasound probe is decontaminated prior to use and after use on each patient.

Operators

The operator and assistant will:-

- Put on a protective apron.
- Wash their hands ensuring all surfaces of the hands are washed thoroughly using the correct technique.
- Then apply alcohol rub to their hands and rub into all skin surfaces before applying sterile gloves.
- Check that all disposable items are in sealed packs – undamaged and in date.
- Open the packs taking care not to touch the contents.
- Open any additional items onto the sterile field.

The Procedure

Amniocentesis and CVS are performed using an aseptic technique.
Immediately before the procedure, the area of skin where the needle is to be inserted is cleaned with antiseptic. The technique should thoroughly clean the skin site and the area be allowed to dry before proceeding.
A sterile drape, supplied in the pack, is applied to the abdomen and a separate sterile gel sachet for each woman. Used needles are disposed in the sharps bin, which is on the bottom of procedure trolley. Used cotton wool should be placed in the waste bag on the trolley. The site is observed for oozing, and an absorbent dressing is applied if this occurs.
A semi-permeable film dressing should be applied to the puncture site and should be left in place for 48 hours. Consider Spray and plaster if not allergic.

Appendix B

Patient Information

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, a deletion or duplication 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.

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. Micro array testing can also be offered and your consultant will discuss this with you if appropriate.

Non-invasive prenatal diagnosis is available if an increased chance screening result is received. It is also available in the private sector.

If your blood group is Rh D negative you may require an Anti-D injection following the procedure. Unless we know that your baby is predicted to be Rh D negative from the blood test that you will be offered at 16 weeks gestation.

If you are HIV positive or Hepatitis B or C positive, an amniocentesis or CVS may increase the chance of maternal-fetal transmission - this will be discussed between you and the fetal medicine consultant.

Amniocentesis

What is amniocentesis?

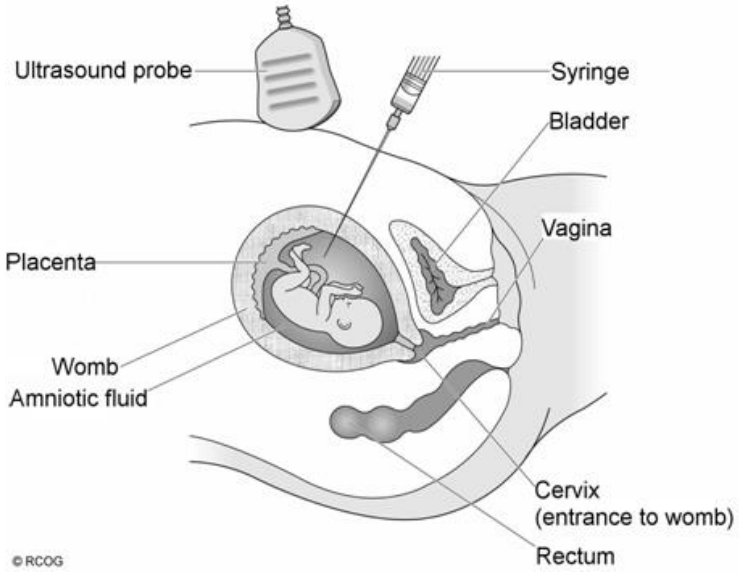
Amniocentesis means obtaining a sample of the amniotic fluid, which surrounds the developing baby in the womb. The fluid is then sent to the laboratory for testing.

When and how is it done?

The safest time to do an amniocentesis is from 15 weeks onwards or once the membranes are fused. 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. The additional overall risk of miscarriage from amniocentesis is approximately 0.5%. 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 if appropriate.

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 QFPCR (rapid test) results are available in around 2 - 4 working days and tests for the 3 commonest extra chromosomes (Downs Syndrome; Edwards Syndrome; Patau Syndrome and sex chromosomes). The cells are then grown to allow full microarray analysis of the chromosomes and takes approximately 10-14 days. Occasionally the initial result is not obtainable e.g. if further testing is required, if there is blood staining of the fluid and in this situation the fetal medicine team will discuss your options with you.

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 greater than 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 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. You should not drive for 24 hours as it is likely your insurance will not cover you for any incident s during that time.

Are there any alternatives to amniocentesis?

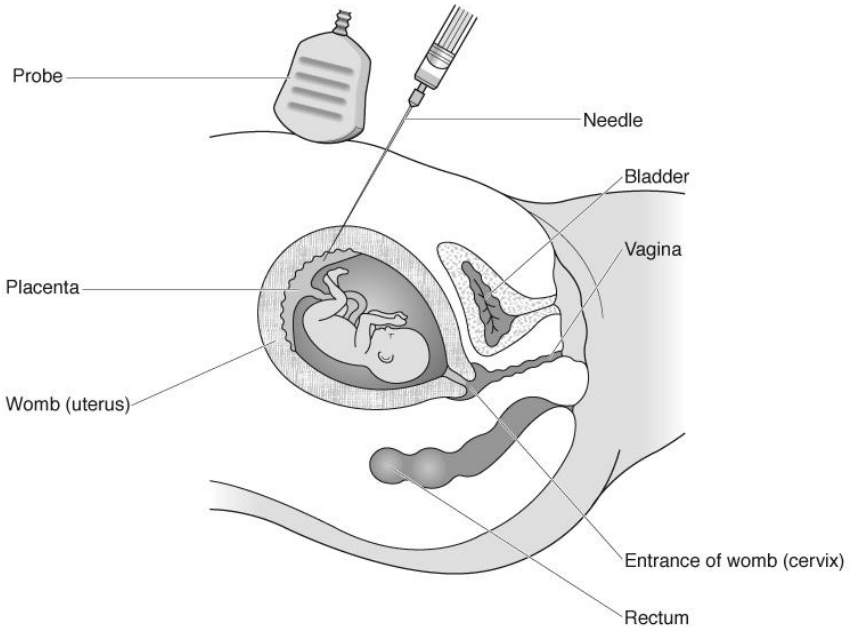
An alternative to amniocentesis is CVS or placental biopsy. This is where placental tissue is tested rather than the amniotic fluid. This can be done from 11+ weeks of pregnancy.

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 increased trisomy risk if the placenta is accessible for testing. 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.

Once the needle is in place, a small amount of placental tissue is obtained by moving the needle. The needle is then taken out and the sample assessed. If not enough tissue is obtained, the procedure may be repeated immediately without an increase in risk.

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 0.5%. In other words about 1 in every 200 women who have CVS under ultrasound guidance between 11 - 13 weeks will miscarry.

What about the results?

A quick result is usually available in 2 - 3 working days for some common chromosomal problems such as Down's syndrome/ Edwards syndrome/ Pataus syndrome and sex chromosome disorders

Microarray testing takes approximately 10-14 working days.

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 a fetal medicine appointment. This will be discussed with you when you attend for the test.

Are the results reliable?

CVS is estimated to give a definitive result in 99 out of every 100 women having the test. 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 next 24 – 48 hours. It is not usually necessary to go to bed but you should avoid any heavy lifting or strenuous exercise.

You may notice some cramping for a few hours afterwards. This is most likely normal. However, 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 - 6.00pm

Pregnancy Assessment Unit
Telephone: 01332 785796 (24 hours)

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

Sources of information/references

Royal College of Obstetricians & Gynaecologists
Amniocentesis and CVS: Information for you. 2011

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

These organisations offer support:

ARC (Antenatal Results and Choices)
345 City Road
London EC1V 1LR
Helpline: 0845 077 2290 or 020 7713 7486
(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
Telephone: 0207 608 8700
Helpline: 0808 808 3555 (Monday to Friday, 9.30am - 5pm)
Email: helpline@cafamily.org.uk
Website: www.cafamily.org.uk

Down's Syndrome Association
Langdon Down Centre
2a Langdon Park
Teddington TW11 9PS
Helpline: 0333 121 2300 (Monday to Friday, 10am - 4pm)
Email: info@downs-syndrome.org.uk
Website: www.downs-syndrome.org.uk

Genetic Alliance UK
Barclay House,
London WC1N 3BH
Telephone: 0207 831 0883
Email: contactus@geneticalliance.org.uk
Website: http://www.geneticalliance.org.uk

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

SOFT UK (Patau syndrome, Edward syndrome)
48 Froggatts Ride, Walmley
Sutton Coldfield B76 2TQ
Telephone: 0121 351 3122 or 0792 305 6132
Email: enquiries@soft.org.uk
Website: www.soft.org.uk

The Miscarriage Association
17 Wentworth Terrace
Wakefield
West Yorkshire WF1 3QW
Helpline: 01924 200799 (Monday to Friday, 9am - 4pm)
Email: info@miscarriageassociation.org.uk
Website: www.miscarriageassociation.org.uk/

UK Thalassaemia Society
19 The Broadway, Southgate
London N14 6PH
Telephone: 020 8882 0011
Email: office@ukts.org
Website: www.ukts.org

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.

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Documentation Control:

Reference Number: **UHDB/AN/01:21/A4**	**Version: UHDB 1 Status: FINAL** **Service provided at RDH only**			
Version / Amendment	Version	Date	Author	Reason
	1	Sept 2013	Lorna Parsons Fetal Medicine Midwife Miss S Raouf Consultant Obstetrician	New
	2	Nov 2017	Miss S Raouf - Consultant Obstetrician C. Davenport – Specialist Midwife	Review
UHDB	1	Oct 2020	Miss S Raouf - Consultant Obstetrician C. Davenport – Specialist Midwife	Review / merge
	2	Dec 2023	Miss S Raouf - Consultant Obstetrician C. Davenport – Specialist Midwife	Review
Intended Recipients: All staff caring for women undergoing Amniocentesis or CVS				
Training and Dissemination: Cascaded through lead midwives/doctors, Published on Intranet, NHS mail circulation list. Article in BU newsletter.				
To be read in conjunction with: Antenatal Screening for Down's Syndrome (D4), Antenatal and Postnatal Cytogenetic and Molecular Genetic Samples (F5), Referral Pathway for Suspected Fetal Abnormality (F3), Anti-D Administration in Pregnancy (A2)				

Consultation with:	Midwifery, Obstetric Staff
Business Unit sign off:	02/01/24 : Maternity Guidelines Group: Miss A Joshi – Chair 10/01/24: Maternity Development & Governance Committee CD: Mr R Devaraj - Chair
Divisional sign off:	16/01/2024
Implementation date:	23/01/2024
Review Date:	January 2027
Key Contact:	Joanna Harrison-Engwell