Reference Number: UHBOBS196 **Version Number: 2** **Published 7/2/24**	**Date of Next Review: 7/2/2027** **Previous Trust/LHB Reference Number: N/A**

Title: Antenatal Invasive Diagnostic Testing – including Amniocentesis and Chorionic Villus Sampling (CVS) Guideline

Introduction and Aim

It is estimated that around 5% of the pregnant population (approximately 30 000 patients per annum) in the UK are offered a choice of invasive antenatal diagnostic testing (most commonly amniocentesis or chorionic villus sampling / CVS). The aim of this guideline is to set a series of evidence-based standards to ensure a high level and consistency of practice in the provision and performance of invasive antenatal diagnostic testing (RCOG 2010).

The words “woman” and “women” have been used throughout this document as this is the way that the majority of those who are pregnant and having a baby will identify. For the purpose of this document, this term also includes girls. It also includes people whose gender identity does not correspond with their birth sex or who may have a non-binary identity

Objectives

- To provide guidance and a clinical pathway for clinicians completing an antenatal invasive diagnostic procedure within the tertiary fetal medicine unit (FMU), UHW.
- To provide a framework for quality and auditable standards.

Scope

- This guideline is written for Fetal Medicine Specialists conducting invasive diagnostic procedures and Fetal Medicine Midwives counselling, assisting with procedures and results reporting. This policy applies to all UHB healthcare professionals in all locations including those with honorary contracts.

Equality Health Impact Assessment	*An Equality Health Impact Assessment (EHIA) has not been completed.*
Documents to read alongside this Procedure	• Antenatal Diagnostic Testing - Amniocentesis - Standard Operating Procedure (SOP) • Antenatal Diagnostic Testing - Chorionic Villus Sampling (CVS) - Standard Operating Procedure (SOP). • Antenatal Screening Wales Policy, Standards & Protocols 2023
Approved by	*Maternity Professional Forum / Perinatal Guidelines Forum*

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Disclaimer

If the review date of this document has passed please ensure that the version you are using is the most up to date either by contacting the document author or the Governance Directorate.

Summary of reviews/amendments			
Version Number	**Date of Review Approved**	**Date Published**	**Summary of Amendments**
1	17.05.2019	20.05.2019	New Guideline
2	7/2/24	7/2/24	Updated by the FMU Team in line with ASW

Indications for offering patient's an invasive procedure (including Amniocentesis and (CVS) chorionic Villus Sampling):

1. High risk screening result.
2. Fetal abnormality (including raised nuchal thickness).
3. A pregnancy at risk of a known inherited condition.
4. A pregnancy at risk of specific viral infections.
5. Previous likely de novo genetic / chromosomal abnormalities after counselling and exploring other options. Patients are given information about Non Invasive Prenatal testing (NIPT) where appropriate.
6. Patient request after extensive counselling.

Please note that other invasive procedures may be carried out within fetal medicine for example amnio drainage, collection of plural fluid or fetal urine etc.

Informed Choice:

Patient's are counselled, given time to decide, issued written information and are granted opportunities to ask questions before conducting the test. Invasive procedures should only be performed with the patient's explicit and informed documented consent (Cardiff and Vale NHS Trust, ASW 2019 & 2022). The risks and benefits should be discussed, including the following information:

- Reason for offering the test (benefits)

- An explanation of the procedure including what is tested
- What is not tested (e.g. structural anomalies, single gene disorders etc.)
- Risks of the procedure as indicated within the consent form's (Cardiff and Vale NHS Trust, ASW 2022)
- Type and significance of results
- Expected timing of results and how these will be communicated
- Accuracy and limitations of laboratory testing

This discussion is supported by giving the following written information, ASW 'Antenatal Screening Wales' Information for women offered further tests for suspected chromosomal condition (ASW 2022).

Procedure:

- Invasive procedures are carried out by Fetal Medicine Consultants within the Fetal Medicine Unit (FMU).
- Amniocentesis may be offered after 15 weeks of gestation by fetal measurements.
- CVS may be offered between 11 and 14 weeks 6 days of gestation by fetal measurements.
- The rationale, benefits, limitations and implications of an invasive procedure must be explained and discussed with the patient by an appropriately trained health professional. Therefore, a patient should be counselled in detail about the procedure, its benefits and risks by the Fetal Medicine Consultant undertaking the procedure and the Fetal Medicine Midwife. Patients are informed that the national risk of

miscarriage; following amniocentesis & CVS is approx. 0.5 % (ASW 2019 & 2022).

- Written information (and adequate time to read the information) should be given to the patient before they are asked to provide consent.
- The patient must be provided with a verbal explanation in a way they are able to understand; detailing the procedure, the risks of the procedure, the genetic information that will be available, the results handling process, pregnancy choices following the results and their right to accept or decline the procedure with informed choice.
- The patient's privacy must be respected at all times, the discussion and procedure should be performed in a room where privacy is assured.
- Invasive procedures can be performed for patients who are hepatitis B or C carriers, but with due regard to the RCOG guidance. The procedure can be performed for patients who are HIV positive but additional care and treatment may be required as outlined in the existing BHIVA Guidelines. 'Danger of Infection' should be identified on the sample and request form.
- A consent form is signed by the patient after counselling prior to the procedure. A copy of the completed consent form should be made available to the patient and the original must be filed in the antenatal maternity record.
- An USS probe decontamination should be completed and operators should adhere to local infection control policies, including enclosing the probe in a sterile bag during the procedure and using an individual sterile gel sachet for each patient.
- The fetus is scanned before the procedure to check viability, confirm fetal size, placenta position and procedure accessibility.

- The patient's abdomen is cleaned with Chlorhexidine 2% and tricept, then sterile drapes are used to protect maternal clothes.
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- 10 ml 1% lignocaine S/C for some invasive procedures may be used to provide local anaesthesia.
- The needle is inserted under direct USS guidance and an appropriate sample taken. Ultrasound scanning during the procedure should be provided by a trained professional to the competencies of the RCOG.
- Fetal viability is checked and shown to the woman prior to and following the procedure.
- The sample tube/universal container should be labelled (use a patient identification label if available) and placed in the leak-proof plastic bag section of the genetics laboratory request form.
- Following the procedure, ALL samples should be checked with the patient to ensure correct labelling.
- The sample together with an EDTA maternal and paternal (if applicable) blood samples are sent to the All Wales Medical Genomics Laboratory in UHB. Genetics laboratory request form must be completed indicating which tests are being requested, providing relevant clinical details and full patient details (use a patient identification label if available). This blood is used during the QF-PCR analysis to identify maternal contamination and the remaining maternal blood and paternal blood is banked for further analysis if required.
- If the amniotic fluid sample is blood stained this needs to be documented, note the QF-PCR result has a higher chance of failing.
- Patients having a procedure for a high-risk screening test result, will only be offered QF-PCR (T21, T18 & T13).
- QF-PCR and SNP array is offered to the patient with high NT (>3.5mm), fetal abnormalities etc.

- QF-PCR includes analysis of the sex (X,Y) chromosomes for CVS and amniocentesis if the indication is cystic hygroma, because of the
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- association with Turners Syndrome. It may also be performed for other reasons following agreement with the genetics department.
- Specific gene testing may be requested via the genetics department.
- CMV PCR DNA Amniotic fluid may be requested if the patient has a known or suspected CMV infection (via Virology not Genetics).
- The patient should be informed that subtle chromosomal changes and single gene defects will not normally be detected by SNP array however, if there is a known genetic condition in the family this should be written on the form even if this is not the reason for the test. The implications of this should be explained, i.e. not all chromosomal conditions will be identified.
- Any additional or specialist genetic testing such as single gene, karyotype or multi gene testing will be discussed and coordinated by the genetics department.
- The patient's blood group is checked and anti-D given (with informed consent) to those who are Rh negative as per the 'Anti D Prophylaxis for Women who are Rhesus D (RhD) Negative' guideline. If Anti D is accepted it should be administered and given as soon as possible after the procedure and certainly within 72 hours (ASW 2010 &RCOG 2002).
- Patient's that are rhesus negative =/> 20 wks gestation require a Kleiuhauer sample taken approx. 20-40 mins following the procedure to assess the volume of fetomaternal haemorrhage and determine if an additional dose of anti-D is required to prevent sensitisation. (BCSH 2009 and 2014).

- An invasive procedure booklet (includes all ASW recommended documentation) is completed after the test. This is kept in the patient's UHW antenatal record (file) with details of the procedure and the results (once available). Note that all 'out of area' patient's will have

- their own UHW FMU antenatal record (file) which is kept within the FMU department at all times.
- UHW patient's will retain their antenatal booking record booklet following the procedure. The remainder of their antenatal record (file) will remain within the FMU at UHW (unless birth is imminent) until all results are received, they are then returned to the patient.
- The patient leaflet 'Information for Patient's following Amniocentesis (Amnio) or Chorionic Villus Sampling (CVS)' should be issued and discussed with the patient, explaining the post-procedure advice and precautions to be taken immediately following the procedure.
- An FMU viewpoint report is completed following the procedure and filed in the maternity record, a copy is uploaded to the Welsh Clinical Portal for the referring unit and GP to access.
- There is a 1% risk of a mosaic result following CVS; this is where a combination of normal cells and cells with abnormal chromosomes are found within the sample from the placenta. This could be due to (CPM) confined placental mosaicism rather than a mosaic karyotype in the baby. An Amniocentesis is offered to determine whether the result is CPM and if it is confirmed then serial ANC growth surveillance is recommended.
- There is a small risk of failure to obtain a result and the patient will be contacted by the Fetal Medicine Specialist team directly and ongoing care arranged.
- The results are e-mailed from UHW Genetics Laboratory to the Fetal Medicine Unit, UHW, Cardiff.

- The patient is usually telephoned by the specialist Fetal Medicine Midwife and given the results (unless results' reporting is pre-arranged via genetics or an alternative arrangement has been made). The results should be provided within 24 working hours (via telephone) of receipt within the FMU department. If the woman doesn't answer the phone and a message asking them to call FMU with a contact number will be left if the answer phone service clearly identifies the patient.
- In case of any post procedure complications, UHW patients should access support from the Gynaecology under 17wks gestation or the Obstetric Assessment Unit after 17 wks gestation. Out of area patients should access support from their local unit.
- An audit of patient evaluation of the service provision may be requested at intervals dependant on staffing levels.

Auditable standards:

1. SNP array/Karyotype offered to all eligible women
2. Written consent is taken in all cases prior to test.
3. Full procedure results received from genetics within 14 days (two weeks).
4. Results given to woman within 24 hours of receipt (or evidence that a reasonable attempt has been made by the FMU department).
5. Rate of pregnancy loss at any gestation after a procedure.
6. Local cytogenetic laboratory culture failure rates for amniocentesis and CVS.
7. Proportion of procedures requiring more than one needle insertion.
8. Proportion of procedures with failure to obtain an adequate sample.
9. Rate of anti–D prophylaxis for women who are RhD-negative undergoing invasive procedures.

References:

Antenatal Screening Wales (2008) Amniocentesis and Chorionic Villus Sampling Policy, Standards and Protocols. Public Health Wales.

Antenatal Screening Wales (2010) Revised Policy, Standards and Protocols. to support the provision of Antenatal Screening in Wales. Public Health Wales

Antenatal Screening Wales (2023) ASW Policy, Standards and Protocols 2023. Public Health Wales.

Antenatal Screening Wales (2022) Information for women offered further tests for suspected chromosomal conditions. Public Health Wales

Antenatal Screening Wales (2022) CVS/Amniocentesis Consent Form. Agreement to (CVS) or Amniocentesis. Public Health Wales.

Cardiff and Vale NHS Trust. Consent Form Ultrasound Guided Invasive Procedures O&G 3a. CF Ultrasound O&G3a/0506

Royal College of Obstetrics and Gyneacologists (2002). Use of Anti D Immunoglobulin for Rhesus Prophylaxis. RCOG Publishing.

Royal College of Obstetrics and Gyneacologists (2010) Amniocentesis and Chorionic Villus Sampling:Green-top Guideline No. 8. RCOG.

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

BCSH (2014) Guideline for the use of anti-D immunoglobulin for the prevention of haemolytic disease of the fetus and newborn. British Committee for Standards in Haematology. 2014.

Public Health England (2021) ‘Screening in pregnancy
Chorionic villus sampling (CVS) and amniocentesis: Information for parents. Public Health England.