Fetal Medicine – Invasive Testing

This guidance does not override the individual responsibility of health professionals to make appropriate decision according to the circumstances of the individual patient in consultation with the patient and /or carer. Health care professionals must be prepared to justify any deviation from this guidance.

Introduction

Guideline for the counselling and management/procedure of invasive testing in fetal medicine.

This guideline is for use by the following staff groups:

All maternity staff who provide counselling to families around invasive testing and those who carry out invasive testing procedures

Lead Clinician(s)

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Approved by Maternity Governance Meeting on:	21st April 2023
Review Date: This is the most current document and should be used until a revised version is in place	21st April 2026

Key amendments to this guideline

Date	Amendment	Approved by:
April 2023	Full guideline review	MGM

Invasive testing may be offered to women:

- Following a high risk screening result
- Following the diagnosis of a fetal anomaly
- If the family have a known genetic mutation
- Maternal request (i.e baby with a previous chromosomal anomaly)

It is estimated that around 5% of the pregnant population are offered a choice of invasive prenatal diagnostic tests (most commonly amniocentesis or chorionic villus sampling). The type of diagnostic test available and offered is likely to vary depending upon the timing of any initial screening test that is performed.

Amniocentesis is the most common invasive prenatal diagnostic procedure undertaken in the UK. Most amniocenteses are performed to obtain amniotic fluid for PCR +/- microarray to identify chromosomal or genetic mutations from 15 weeks (15+0) onwards.

Chorionic villus sampling (CVS) is usually performed between 11 (11+0) and 13 (13+6) weeks of gestation and involves aspiration or biopsy of placental villi.

The procedure for invasive testing is as follows:

- Ensure the woman's notes and results are available and data has been checked to confirm that they are correct.
- Check RhD status, RhD negative women will require anti-D immunoglobin post procedure as per guideline D2
- Any gestation FFDNA shows negative fetal blood group, women don't require bloods or anti-D.
- <20 weeks if no FFDNA or positive fetal blood group, then a Group and Screen is required prior to amniocentesis and give anti-D.
- >20 weeks and no FFDNA or positive fetal blood group, wait 30 mins after procedure before taking Group and Screen and Kleihauer. The Group and Screen is after the procedure to avoid two separate blood tests for the patient. Anti-d must be given as it's a sensitising event regardless of kleihuer result , minimum 500iu
- Check booking virology results
- Invasive testing and maternal viral infection: Invasive prenatal testing in the first or second trimester can be carried out in women who carry **hepatitis B or C**. The limitations of the available data should be explained and amniocentesis is preferential to CVS.
- If amniocentesis is being contemplated in HIV positive women then:
    - Advice should be sought from the fetal medicine specialists and the HIV Care team about concomitant antiretroviral therapy (if not already receiving treatment).

- Review viral load and treatment regimens prior to invasive prenatal testing in women with HIV and consider delaying the procedure until there is no detectable viral load if the woman is already on treatment.

- It should be explained that it is uncertain whether invasive diagnostic tests are a route for maternal/child transmission.

- Every effort should be made to avoid inserting the needle through the placenta.

- If HIV status is unknown then HIV in-house testing (which takes approximately 90 minutes) should be offered.

- Where women decline screening for blood borne viruses and are being counselled for prenatal
diagnostic procedures, inform and document the potential risk of vertical transmission of infection to the fetus, need for neonatal testing and complete a paediatric alert form.

- Ensure the woman has been fully counselled regarding her options, including non-invasive prenatal testing (NIPT) if appropriate and is aware of the risks and implications of amniocentesis.

- Complete 'Discussion prior to Amniocentesis' sheet (Appendix 1) or 'Discussion prior to CVS
(Appendix 2) and provide patient information from the following websites:
www.nhs.uk/conditions/amniocentesis/
www.nhs.uk/conditions/chorionic-villus-sampling-cvs/

- Women should be informed that third-trimester amniocentesis does not appear to be associated with a significant risk of emergency delivery, however when compared with mid-trimester procedures, complications including multiple attempts and bloodstained fluid are more common in third-trimester procedures.

- Written consent should be obtained prior to invasive procedures.

- The operator should be adequately experienced, operators carrying out unsupervised amniocentesis and CVS should be trained to the competencies expected of subspecialty training in maternal and fetal medicine, the RCOG Fetal Medicine Advanced Training Skills Module (ATSM) or other international equivalent. Competency should be maintained and outcomes audited.

- It is recommended that, in the case of multiple pregnancies, invasive testing is performed by a specialist who has the expertise to subsequently perform a selective termination of pregnancy if required; therefore such cases should be referred to Birmingham Women's Hospital Fetal Medicine Unit (BWH).

- Pre-procedure confirm the gestational age and perform an ultrasound scan to confirm fetal heart beat and placental site.

- Amniocentesis is a sterile procedure performed under continuous ultrasound control using an echogenic tip needle and a sterile probe cover.

- A trans-placental approach may be appropriate if it provides easy access to a pool of amniotic fluid but care should be taken to avoid the cord insertion.

- Post procedure; confirm presence of fetal heartbeat with ultrasound scan.

- Ensure specimen is CLEARLY and CORRECTLY labelled, and cytogenetics form has been completed in full.

- Amniotic fluid specimen is taken directly to the laboratory following a second specimen check. The sample is then transferred to cytogenetics by designated trust courier (if out of hours, then permission for taxi transfer to be sought from head of midwifery).

- The screening coordinator or clinic midwife should call the cytogenetics laboratory to inform them of the pending sample and request a confirmation of receipt.

- Give anti-D immunoglobulin if required.

- Ensure medical notes and patient handheld records are completed.

- Ensure the woman is aware how her results will be communicated to her. Document in the notes how she wishes to be notified of the results if an abnormality is detected. Ensure that the telephone number in the record for communication of the results is current.

- Advise the woman not to go home unaccompanied.

- Advise her to rest for 48 hours and to report any bleeding, pain or suspected amniotic fluid loss to the EGAU/EPAU if <16/40 and Maternity Triage if >16/40.

Reporting of Results

- All results will be reported directly to the antenatal screening coordinator by email from the cytogenetics laboratory at BWH.

- Normal results will be communicated to the woman by telephone and a copy of the results will be filed in the hospital notes.

- If an abnormality is found the woman will be contacted by telephone and advised that a face-to-face consultation with the screening team has been arranged to discuss these results further and to devise a plan of care.

- Community Midwives will be informed of all results.

Appendix 1

Worcestershire NHS Acute Hospitals NHS Trust

APPENDIX 1 DISCUSSION PRIOR TO AMNIOCENTESIS

Please attach patient sticker here or record:

Name:……..

NHS No: ☐☐☐☐☐☐☐☐☐☐

Unit No: ☐☐☐☐☐☐☐☐☐☐

D.O.B: ____/____/_____ *Female*

Consultant: …… *Ward:* ……..

Reason for referral

Maternal request ☐

Increased screening results ☐

Previous anomaly ☐

Other reason ……

Risks

Risk Misc. 1% ☐

Failed culture 1 : 200 ☐

Risk infection <1 : 1000 ☐

Amn. Fluid leakage 1% ☐

PCR

Not available if bloodstained ☐

Communication of results ☐

PCR T13 ☐ T18 ☐ T 21 ☐ (No charge)

Result 72-96 hours ☐

Full Karyotype 2-3 weeks (Cytogenetics cost £190)

Fetal Sex (Cytogenetics cost £70)

Method of TOP ☐

Aware of signs of miscarriage ☐

Amniocentesis Date:____/____/_____

Placental site _____

Transplacental tap Yes ☐ No ☐

Attempts _____

Failed Yes ☐ No ☐

Clear tap / Blood tap / Other

Swab count correct Yes ☐

Performed by: _____

Signed :_____

HIV Result _____

Blood Group _____

Gestation in weeks _____

Anti D

Not required ☐

Given ☐

Batch No: _____

Signed: _____

Outcome

Declined test Yes ☐ No ☐

Date of Delivery _____/_____/_____

Gestation

Pregnancy Miscarriage ☐
Live Birth ☐
Stillbirth ☐

Sex M ☐ F ☐ Wt:_____ g

Signature:

Midwife _____

Patient. _____

Community Midwife to visit Yes ☐ No ☐

FOLLOWING DELIVERY PLEASE FILE IN THE WOMAN'S OBSTETRIC RECORD IN HER MEDICAL NOTES

APPENDIX 2 DISCUSSION PRIOR TO CVS

Please attach patient sticker here or record:

Name:……..

NHS No: ☐☐☐☐☐☐☐☐☐☐

Unit No: ☐☐☐☐☐☐☐☐☐☐

D.O.B: ____/____/_____ Female

Consultant: …… Ward: ……..

Reason for referral

Maternal request ☐

Increased screening results ☐

Previous anomaly ☐

Raised NT ☐

Other known genetic condition ☐

Risks

Risk Misc. 1% ☐

Insufficient sample 1% ☐

Risk infection 1% ☐

Placental mosaicism 1% ☐

PCR

Not available if bloodstained ☐

Communication of results ☐

PCR T13 ☐ T18 ☐ T 21 ☐ (No charge)

Microarray ☐

Other specific genetic test ……..

Result 72-96 hours ☐

Full Kayotype 2-3 weeks (Cytogenestics cost £190)

Fetal Sex (Cytogenetics cost £70)

Method of TOP ☐

Aware of signs of miscarriage ☐

CVS Date:____/____/_____

Placental site _____

Attempts _____

Failed Yes ☐ No ☐

Swab count Pre – Sign 1……Sign 2……..

Swab count Post – Sign 1……..Sign 2……..

Consent form completed ☐

HIV Result ……

Blood Group ……

Other micro/virology .....

Gestation in weeks ……..

Anti D

Not required ☐

Given ☐

Batch No: _____

Signed: _____

Outcome

Declined test Yes ☐ No ☐

Date of Delivery _____/_____/_____

Gestation

Pregnancy Miscarriage ☐
Live Birth ☐
Stillbirth ☐

Sex M ☐ F ☐ Wt:_____ g

Signature:

Midwife _____

Patient. _____

FOLLOWING DELIVERY PLEASE FILE IN THE WOMAN'S OBSTETRIC REORD IN HER MEDICAL NOTES

Monitoring

Page/ Section of Key Document	Key control:	Checks to be carried out to confirm compliance with the Policy:	How often the check will be carried out:	Responsible for carrying out the check:	Results of check reported to: *(Responsible for also ensuring actions are developed to address any areas of non-compliance)*	Frequency of reporting:
	WHAT?	**HOW?**	**WHEN?**	**WHO?**	**WHERE?**	**WHEN?**
	Data Feeding into national audit	Audit collection Data	Continuously	Antenatal Screening Team	National Audit	Quarterly & Yearly

References

1. Non-invasive Prenatal Testing (Green-Top Guideline No. 74)

2. www.nhs.uk/conditions/amniocentesis/

3. www.nhs.uk/conditions/chorionic-villus-sampling-cvs/

Contribution List

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This key document has been circulated to the following individuals for consultation;

Designation
All Maternity staff – via guideline newsletter

This key document has been circulated to the chair(s) of the following committee's / groups for comments;

Committee
Maternity Governance Meeting