

Invasive Testing in Fetal Medicine

Key Document code:	WAHT-TP- 094	
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Approved by:	Maternity Governance Meeting	
Date of Approval:	18 th September 2020	
Date of review:	15 th November 2022	
This is the most current document and should be used until a revised version is in place		

Key Amendments

Date	Amendments	Approved by
Sept 2020	New document approved	Maternity Governance

Invasive testing may be offered to women following a high risk screening result, the diagnosis of a fetal anomaly, if the family have a known genetic mutation or 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 immunoglobulin post procedure as per guideline D2
- 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.

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/

Appendix 1

APPENDIX 1 DISCUSSION PRIOR TO AMNIOCENTESIS

<p><i>Please attach patient sticker here or record:</i></p> <p>Name:.....</p> <p>NHS No: <input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/></p> <p>Unit No: <input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/><input type="checkbox"/></p> <p>D.O.B: ___/___/_____ Female</p> <p>Consultant: Ward:</p>	<p>Reason for referral</p> <p>Maternal request <input type="checkbox"/></p> <p>Increased screening results <input type="checkbox"/></p> <p>Previous anomaly <input type="checkbox"/></p> <p>Other reason</p>
<p>Risks</p> <p>Risk Misc. 1% <input type="checkbox"/></p> <p>Failed culture 1 : 200 <input type="checkbox"/></p> <p>Risk infection <1 : 1000 <input type="checkbox"/></p> <p>Amn. Fluid leakage 1% <input type="checkbox"/></p> <p>PCR</p> <p>Not available if bloodstained <input type="checkbox"/></p>	<p>Communication of results <input type="checkbox"/></p> <p>PCR T13 <input type="checkbox"/> T18 <input type="checkbox"/> T 21 <input type="checkbox"/> (No charge)</p> <p>Result 72-96 hours <input type="checkbox"/></p> <p>Full Kayotype 2-3 weeks (Cytogenetics cost £190)</p> <p>Fetal Sex (Cytogenetics cost £70)</p> <p>Method of TOP <input type="checkbox"/></p> <p>Aware of signs of miscarriage <input type="checkbox"/></p>
<p>Amniocentesis Date: ___/___/_____</p> <p>Placental site _____</p> <p>Transplacental tap Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p>Attempts _____</p> <p>Failed Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p>Clear tap / Blood tap / Other</p> <p>Swab count correct Yes <input type="checkbox"/></p> <p>Performed by: _____</p> <p>Signed : _____</p>	<p>HIV Result _____</p> <p>Blood Group _____</p> <p>Gestation in weeks _____</p> <p>Anti D</p> <p>Not required <input type="checkbox"/></p> <p>Given <input type="checkbox"/></p> <p>Batch No: _____</p> <p>Signed: _____</p>
<p>Outcome</p> <p>Declined test Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p>Date of Delivery ___/___/_____</p> <p>Gestation</p> <p>Pregnancy Miscarriage <input type="checkbox"/></p> <p style="padding-left: 20px;">Live Birth <input type="checkbox"/></p> <p style="padding-left: 20px;">Stillbirth <input type="checkbox"/></p> <p>Sex M <input type="checkbox"/> F <input type="checkbox"/> Wt: _____ g</p>	<p>Signature:</p> <p>Midwife _____</p> <p>Patient. _____</p> <p>Community Midwife to visit Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p style="text-align: center;">FOLLOWING DELIVERY PLEASE FILE IN THE WOMAN'S OBSTETRIC RECORD IN HER MEDICAL NOTES</p>

Please note that the key documents are not designed to be printed, but to be used on-line. This is to ensure that the correct and most up-to-date version is being used. If, in exceptional circumstances, you need to print a copy, please note that the information will only be valid for 24 hours and should be read in conjunction with the key document supporting information and/or Key Document intranet page, which will provide approval and review information.

APPENDIX 2 DISCUSSION PRIOR TO CVS

<p><i>Please attach patient sticker here or record:</i></p> <p>Name:.....</p> <p>NHS No: <input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/></p> <p>Unit No: <input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/><input type="text"/></p> <p>D.O.B: ____/____/____ Female</p> <p>Consultant: Ward:</p>	<p>Reason for referral</p> <p>Maternal request <input type="checkbox"/></p> <p>Increased screening results <input type="checkbox"/></p> <p>Previous anomaly <input type="checkbox"/></p> <p>Raised NT <input type="checkbox"/></p> <p>Other known genetic condition <input type="checkbox"/></p>
<p>Risks</p> <p>Risk Misc. 1% <input type="checkbox"/></p> <p>Insufficient sample 1% <input type="checkbox"/></p> <p>Risk infection 1% <input type="checkbox"/></p> <p>Placental mosaicism 1% <input type="checkbox"/></p> <p>PCR</p> <p>Not available if bloodstained <input type="checkbox"/></p>	<p>Communication of results <input type="checkbox"/></p> <p>PCR T13 <input type="checkbox"/> T18 <input type="checkbox"/> T 21 <input type="checkbox"/> (No charge)</p> <p>Microarray <input type="checkbox"/></p> <p>Other specific genetic test</p> <p>Result 72-96 hours <input type="checkbox"/></p> <p>Full Kayotype 2-3 weeks (Cytogenetics cost £190)</p> <p>Fetal Sex (Cytogenetics cost £70)</p> <p>Method of TOP <input type="checkbox"/></p> <p>Aware of signs of miscarriage <input type="checkbox"/></p>
<p>CVS Date: ____/____/____</p> <p>Placental site _____</p> <p>Attempts _____</p> <p>Failed Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p>Swab count Pre – Sign 1.....Sign 2.....</p> <p>Swab count Post – Sign 1.....Sign 2.....</p> <p>Consent form completed <input type="checkbox"/></p>	<p>HIV Result</p> <p>Blood Group</p> <p>Other micro/virology</p> <p>Gestation in weeks</p> <p>Anti D</p> <p>Not required <input type="checkbox"/></p> <p>Given <input type="checkbox"/></p> <p>Batch No: _____</p> <p>Signed: _____</p>
<p>Outcome</p> <p>Declined test Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p>Date of Delivery ____/____/____</p> <p>Gestation</p> <p>Pregnancy Miscarriage <input type="checkbox"/></p> <p>Live Birth <input type="checkbox"/></p> <p>Stillbirth <input type="checkbox"/></p> <p>Sex M <input type="checkbox"/> F <input type="checkbox"/> Wt: _____ g</p>	<p>Signature:</p> <p>Midwife _____</p> <p>Patient. _____</p>
<p>FOLLOWING DELIVERY PLEASE FILE IN THE WOMAN'S OBSTETRIC REORD IN HER MEDICAL NOTES</p>	

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