STANDARD OPERATING PROCEDURE

Antenatal Screening for Down's Syndrome, Edwards' Syndrome and Patau's Syndrome

Screening is offered so women can find out how likely it is that the baby has Down's Syndrome (T21), Edwards' Syndrome (T18) or Patau's Syndrome (T13)

These conditions occur when there is an extra copy of chromosome 13, 18 or 21- an autosomal trisomy. The baby will have 47 chromosomes in each cell instead of the normal 46. This problem arises at conception and affects how the baby will develop.

Trisomy 21

Incidence - 1:1000 Babies have an extra copy of chromosome 21 in every cell. Down's Syndrome can vary in severity from mild to severe. It is associated with learning difficulties, communication problems and specific physical problems – heart defects, problems with the digestive system, hearing and vision.

Trisomy 18

Incidence - 3:10 000 Babies have an extra copy of chromosome18 in every cell. Most babies will not survive the pregnancy or will pass away shortly after birth. It is possible to survive to adulthood but this is very rare as the condition is associated with multiple severe abnormalities.

Trisomy 13

Incidence – 1: 2:10 000 Babies have an extra copy of chromosome13 in every cell. Most babies will not survive the pregnancy or will pass away shortly after birth. It is possible to survive the first year of life and possibly longer - but again this is very rare as the condition is associated with abnormal development and multiple severe organ defects.

Screening tests are optional and can be accepted or declined.

Screening should be offered to all pregnant women and their decision clearly documented on E3, in the green hand held notes (page 9) and in the hospital notes.

Screening Tests Available

Combined Test

First Trimester Combined Screening – 11+2-14+1/**CRL 45mm – 84mm**. A dating scan, nuchal translucency measurement and blood test at the same visit.

Bloods – Free Beta HCG, Alpha-fetoprotein, Papp-a (Pregnancy-associated Plasma Protein).

Screening Options in the Ist Trimester

1. Down's, Edwards' and Patau's Syndromes
2. Down's Syndrome only
3. Edward's and Patau's Syndrome only
4. None of these conditions

Quadruple Test

Second Trimester Screening at 15+0 to 20+0/**HC 101mm – 172mm**. A blood test carried out based on an accurate gestation following a dating scan.

Bloods – Free Beta HCG, Alpha-fetoprotein, Unconjugated Estriol, Inhibin A

The optimum time to perform the Quadruple test is at 16 weeks.

The Quadruple test will give a result for Down's Syndome in the second trimester. The Anomaly scan is the screening test for Edwards' Syndrome and Patau's Syndrome in the second trimester.

Patient Information

- At the first contact with a Health Professional the leaflet 'Screening tests for you & your baby' should be given to the woman.
- This could be given by the Midwife at home, in Antenatal Clinic, at the GP's surgery or in a community based centre
- Giving written information at the earliest possible opportunity enables the woman to have knowledge of the tests available and begin to consider her options prior to the booking interview.

Booking Visit / Midwife Consultation

- It should be clearly documented in the hand held notes that a discussion regarding screening has taken place.
- The following points will be included in this discussion:
    1. Ensure a basic understanding of the conditions
    2. Screening is optional

3. Screening will give a higher/lower chance result
4. Purpose, risks, benefits and limitations of a screening test
5. What the test involves
6. Detection rates and screen positive rates
7. Receiving results
8. Diagnostic testing

- Document clearly in the hand held notes if screening is declined
- If accepted – clearly document which screening test has been accepted i.e. all three conditions, T21 only, T13 & T18 only. A consent form will be completed in the USS department prior to scan and electronically scanned for a record of consent.
- The biochemistry form will be commenced when the woman attends the Ultrasound department. It will be completed in the clinic and checked that all the required information is filled in and correct. The person responsible for this will sign the form on taking the blood sample.

Screening Declined

- Document in hand held notes, hospital notes and E3
- No further action.

First Trimester Combined Screening Accepted

- Document consent and choice of conditions in hand held notes (p.9). Enter on E3 and document in hospital notes
- A consent form is signed in the Ultrasound Department – clearly identifying which condition(s) the woman requires screening for. It is electronically scanned and the paper copy given to the woman
- Commence the Antenatal Screening Request Form (R21/version 8) in the Ultrasound Department - the receptionist will provide the woman with a form to give to her sonographer.
- The woman should be fully aware of what test she has consented to prior to the scan.
- A dating scan will be performed and a NUCHAL TRANSLUCENCY measurement taken if the **CRL is 45-84mm**.
- The USS details, NT measurement and the operators FMF code are recorded on the form by the sonographer.
- The USS report and the request form are forwarded to ANC where the woman will complete her booking
- The woman will be weighed in ANC, bloods will be taken and the completed request form will be sent to the laboratory with the labelled blood sample.
- Advise how results will be communicated and advise woman to contact Screening Midwife if no lower chance letter is received within 3 weeks
- The person taking the blood is responsible for labelling the sample, signing the form and ensuring that all sections have been completed correctly.

- A sticker is placed on the screening sheet/ in the screening file - so the woman's details can be entered on the screening database as part of the failsafe process
- Sheets from Bolton One Leisure Centre, Ingleside and Fairfield Hospital are forwarded to the ANDU clerk for entry on the database. The clerk accesses the file in the Bolton ANC regularly to retrieve this information.

*If a dating scan is performed and the **CRL is <45mm** – and first trimester combined screening is requested - a further appointment for a nuchal translucency scan and blood test will be given by the USS department. This should be as soon as possible within the appropriate timeframe

*If the NT is unobtainable after 2 separate episodes on the bed – the Quadruple test is offered in the second trimester. Appointment made at reception – ANDU/ANC – on LE2.2

*If a woman attends and the **CRL is >84mm** - the Quadruple test will be offered in the second trimester. Appointment made at reception – ANDU/ANC – on LE2.2

All appointments for blood tests should be entered on LE2.2 on the **Screening Bloods** Clinic

Women are attended for their appointment when they check in at ANDU reception

Non-attendance should be followed up as part of the Antenatal Clinic DNA Policy

Second Trimester Screening Accepted

- Document consent in hand held notes, enter on E3 and document in hospital notes.
- Ensure the woman is aware what information this test provides
- A dating scan will be/have been performed. The NT will have been unobtainable or the CRL >84mm
- The USS report and the request form is forwarded to ANC where the woman will complete her booking
- If the **HC >101mm** the Quadruple test can be taken to avoid a return visit
- If the **HC is <101mm** – give appointment to return for test at 15-16 weeks and enter on LE2.2
- The test request form is completed by a HCA on return to the blood clinic
- On returning for the screening test the woman will be weighed in ANDU, bloods will be taken and the completed request form will be sent to the laboratory with the labelled blood sample.
- The person taking the blood is responsible for labelling the sample, signing the form and ensuring that all sections have been completed correctly
- Advise how results will be communicated and advise woman to contact Screening Midwife if no lower chance letter is received within 3 weeks
- A sticker is placed on the screening sheet/ in the screening file - so the woman's details can be entered on the screening database as part of the failsafe process
- Sheets from Bolton One Leisure Centre, Ingleside and Fairfield Hospital are forwarded to the ANDU clerk for entry on the database. The clerk accesses the file in the Bolton ANC regularly to retrieve this information.

*If the woman is unable to attend the appointment the screening test will be rescheduled at her convenience

Women are attended for their appointment when they check in at ANDU reception

Non-attendance should be followed up as part of the Antenatal Clinic DNA Policy

Receipt of Results

After analysis all results will be reported to the referring ante-natal clinic by the biochemistry department.

Lower Chance Result

- A lower chance result is received as a paper report
- Result signed off by Screening Midwife and forwarded to clerk
- A letter will be sent to the woman informing her of her lower chance result
- Letters are specific to which screening option was selected
- Result entered on E3 by clerk on sending letter
- Date letter sent entered on database as part of failsafe process
- Community Midwife to ensure result received at next contact – as part of failsafe
- Database should be checked daily to identify tests taken and no letter sent/result received. These women can be investigated – results confirmed or arrange retesting
- Contact details provided should further information/support be required

Higher Chance Result

- A higher chance result will be phoned to the Specialist Midwife, or a designated colleague.
- The hard copy is downloaded from the Perkins Elmer eReports system by Screening Midwife
- All details are rechecked – name, DOB, USS date, USS details, gestation
- Result documented on E3 and in hospital notes – T21, T13/18 or all three
- Recorded in Screening Midwife's diary – High chance results/diagnostics
- Higher Chance result entered on **Down's Syndrome Screening Programme Audit Collection Programme**
- The woman will be contacted by telephone within 3 working days and invited to attend ANDU to discuss the result and the options available to her, or discussion via telephone if she prefers
- Specialist counselling with Screening Midwife and further investigations/diagnostic testing will be offered.
- Allow time to consider options and discuss with partner/family
- Details provided of FASP websites, FASP literature provided, ARC helpline
- Contact details for Screening Midwife for further discussion, information or support – and to notify of decision

*If the Specialist Midwife is unable to contact the woman by telephone after 3 days – a letter will be sent, as a final attempt/no response to the letter a visit from the Community Midwife will be arranged.

Diagnostic Testing

Following a higher chance result at **first trimester combined screening** the woman will be offered further counselling and diagnostic testing. Her options include:

- Decline further testing/diagnostic testing and await events
- Chorionic Villus Biopsy – from 12 weeks
- Amniocentesis – from 15 weeks
- NIPT – privately at present/awaiting NHS implementation

Following a higher chance result at **second trimester** screening the woman will be offered further counselling and diagnostic testing. Her options include:

- Decline diagnostic testing and await events
- Amniocentesis – from 15 weeks
- NIPT – privately at present/awaiting NHS implementation

Diagnostic Testing Declined

- Document on E3, in hand held notes and in hospital notes
- Complete outcome on **Down's Syndrome Screening Programme Audit Collection Programme**
- Inform USS department prior to anomaly scan
- Email to team advising them of events to date and woman's decision

Diagnostic Testing Accepted

- Document on E3, in hand held notes and in hospital notes
- Complete outcome on **Down's Syndrome Screening Programme Audit Collection Programme**
- Appointment given for CVB or Amniocentesis by Screening Midwife
- Appointment booked on CRIS and LE2.2
- Appointment entered in High Chance Results/Diagnostics Diary – Screening Midwife's office
- Individual counselling as appropriate
- Written information available - FASP literature
- Ensure woman has contact details for Specialist Midwife
- Blood group/IDPS results must be confirmed prior to procedure
- Referral pathways available in Standard Operating Procedures as below

Please refer to:

- **Standard Operating Procedure – Chorionic Villus Biopsy**
- **Standard Operating Procedure – Amniocentesis**

Twin Pregnancies

- Combined Test available for Dichorionic/Monochorionic Twins
- NIPT not recommended on twin pregnancies
- Determine chorionicity of twins
- NT measurements must be obtained on both babies
- Second appointment to be given if NT unobtainable as Quadruple test has reduced detection rate
- Dichorionic – separate chance reported on each baby
- Monochorionic – one chance reported for both babies
- Counsel regarding options - if NT not obtainable Quad Test is available but ensure woman is aware of reduced detection rate/performance
- Discuss diagnostic testing on twin pregnancies and implications of same i.e. options in the event of an affected baby
- Counselling for procedure should take place at a Specialist Unit where test is performed
- Diagnostic testing should be performed by a specialist in a centre where selective feticide could also be offered (RCOG, Guideline No. 8, 2005) – Fetal Medicine Unit/St. Mary's Hospital Manchester

Lower Chance Result

- A lower chance result is received as a paper report
- Result signed off by Screening Midwife and forwarded to clerk
- A letter will be sent to the woman informing her of her lower chance result
- Letter is specific to twin pregnancy
- Result entered on E3 by clerk on sending letter
- Date letter sent entered on database as part of failsafe process
- Community Midwife to ensure result received at next contact as part of failsafe
- Database should be checked daily to identify tests taken and no letter sent/result received. These women can be investigated – results confirmed or arrange retesting
- Contact details provided should further information/support be required

Higher Chance Result

- A higher chance result will be phoned to the Specialist Midwife, or a designated colleague.
- The hard copy will be emailed via nhs.net
- The hard copy is downloaded from the Perkins Elmer eReports system by Screening Midwife

- All details are rechecked – name, DOB, USS date, USS details, gestation
- Result documented on E3 and in hospital notes
- Recorded in Screening Midwife's diary – High chance results/diagnostics
- Higher Chance result entered on **Down's Syndrome Screening Programme Audit Collection Programme**
- The woman will be contacted by telephone within 3 working days and invited to attend ANDU to discuss the result and the options available to her, or discussion via telephone if she prefers.
- Allow time to consider options and discuss with partner/family
- Referral to Fetal Medicine to discuss diagnostic testing if required.
- Details provided of FASP websites, FASP literature provided, ARC helpline
- Contact details for Screening Midwife for further discussion, information or support – and to notify of decision

<u>Please refer to:</u>

- **Standard Operating Procedure – Referral to Fetal Medicine Unit**