Guideline

Title: Screening for Down's, Edwards' & Patau's Syndromes

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Scope: Midwives, Sonographers, Obstetric doctors, GPs and the Laboratory staff.		
To be read in conjunction with the following documents: • Screening Tests for You and Your Baby (UK National Screening Committee Leaflet) • Screening in Pregnancy Guideline • Down Syndrome - Management of suspected or confirmed Down Syndrome on Postnatal Ward/ Neonatal Unit Guideline		
CQC Fundamental standards:		

Disclaimer –

Since every patient's history is different, and even the most exhaustive sources of information cannot cover every possible eventuality, you should be aware that all information is provided in this document on the basis that the healthcare professionals responsible for patient care will retain full and sole responsibility for decisions relating to patient care; the document is intended to supplement, not substitute for, the expertise and judgment of physicians, pharmacists or other healthcare professionals and should not be taken as an indication of suitability of a particular treatment for a particular individual.

The ultimate responsibility for the use of the guideline, dosage of drugs and correct following of instructions as well as the interpretation of the published material **lies solely with you** as the medical practitioner.

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Guideline Statement

To enable staff to provide a comprehensive prenatal screening service.

Executive Summary

Screening is a process of identifying apparently healthy people who may be at increased risk of a disease or condition. They can then be offered information, further tests and appropriate treatment to reduce their risk and/or any complications arising from the disease or condition. [2]

The UK National Screening Committee (UK NSC) has responsibility for setting screening policy. It recommends that all eligible pregnant women in England are offered screening to assess the risk of the baby being born with Down's (Trisomy 21/T21), Edwards' (Trisomy 18/T18) and Patau's (Trisomy 13/T13) syndromes or a number of fetal anomalies (structural abnormalities of the developing fetus). [1.]

FASP aims to ensure that there is equal access to uniform and quality assured screening across England and that women are provided with high quality information so they can make an informed choice about their screening options and pregnancy choices. Some women may choose not to be screened at all, or accept screening for some conditions and it is important that this choice is respected. The screening policy is to offer screening to assess the risk of the baby being born with Down's, Edwards'/Patau's syndromes. [1, 2]

The test of choice for both singleton and twin pregnancies is first trimester combined screening. Women can choose:

- not to have screening
- to have screening for T21 and T18 / T13
- to have screening for T21 only
- to have screening for T18 / T13 only [1,2]

The first scan usually takes place between 10 to 14 weeks and includes a blood sample taken to test for Down's, Edwards'/Patau's syndromes, with a second scan for fetal anomalies between 18+0 to 20+6 weeks. The timing of the scans allows for further diagnostic tests if required and ensures women have time to consider decisions about continuing their pregnancy. [1, 2]

The National Standards are[1]:

Screening strategy	Thresholds	
	Acceptable	Achievable
T21	Standardised DR 85%	
	Standardised SPR 1.8-2.5%	Standardised SPR 1.9-2.4%
T18/T13	Standardised DR 80%	
	Standardised SPR 0.1-0.2%	Standardised SPR 0.13-0.17%
T21/T18/T13	Standardised SPR 1.8-2.5%	Standardised SPR 1.9-2.4%
Quadruple (T21)	Standardised DR 80%	
	Standardised SPR 2.5-3.5%	Standardised SPR 2.7-3.3%

*The DR and SPR for the quadruple test relate to singleton pregnancies only

The following policy document relates to the screening programme currently offered in Milton Keynes University Hospital NHS Foundation Trust and is benchmarked to the national standards set out in April 2015. It is therefore an evolving document that will need revisiting and revising to encompass any future developments and recommendations from national bodies.

The aims of this screening programme are:

- To offer screening and diagnosis to all 'elligible' women booked before 20 weeks of pregnancy and attending for antenatal care within the area covered by the Milton Keynes Hospital NHS Foundation Trust.
- To provide adequate high quality information on the screening process to support each woman to make an informed decision on whether to accept or decline the offer of screening.
- Provision of adequate information and support to enable the woman and her partner to make a decision on the outcome of the pregnancy.

- For women with a confirmed diagnosis as the result of screening, who choose to continue the pregnancy, provide optimal management of the pregnancy, birth and newborn period.
- For women with a confirmed diagnosis as a result of screening who opt for termination of pregnancy to provide the optimal care and support including bereavement counselling

Objectives

- To process and report on screening tests in a timely manner as detailed in the UK NSC working standards and programme specific standards.
- To offer appropriate tests and methods of screening that meet national standards. To offer appropriate diagnostic tests to all women with screen positive results within programme specific recommended timescales.
- To promote an appropriate level of knowledge for all health professionals involved in the screening programme as recommended by the UKNSC.
- To minimise the adverse effects of screening: anxiety, misunderstanding, inaccurate information, unnecessary investigation and follow-up, and inappropriate disclosure of patient specific information.
- To have in place systems for risk assessment and management of adverse incidents occurring during the screening process.

1.0 Roles and Responsibilities

There are many health care professionals involved in the screening for Down's, Edwards' & Patau's syndromes including Community Midwives, Sonographers, Sonographer Assistants, MK Pathology department, Screening laboratory at Oxford, Prenatal Screening Team and Obstetricians. Please see the main body of the document for the outline of the roles and responsibilities. Other Health care professionals should be aware of this document.

2.0 Implementation and dissemination of document

This guideline will be available on the Trust intranet.

3.0 Processes and procedures

3.1 1st Trimester Screening for Down's, Edwards' & Patau's syndromes

The combined test uses maternal age, the nuchal translucency measurement (NT) and two biochemical tests, free beta hCG and PAPP-A, together with the gestational age calculated from the crown rump length (CRL) measurement, to calculate the risk of the pregnancy being affected by T21or T18/T13. The optimal time to perform the combined test is between 11 weeks 2 days to 14 weeks 1 day of gestation, which corresponds to a CRL of 45.0 mm to 84.0 mm. In cases where screening is accepted but it is not possible to obtain the NT measurement at the first appointment, at least one other attempt should be offered, this may be on the same day or at a later date. If it is not possible to obtain an accurate NT measurement despite 'twice on the couch' then further attempts do not have to be offered and the woman should be referred in to the second trimester screening pathway. If the ultrasound measurement shows that the CRL is less than 45.0 mm, the woman should be recalled for a further scan to measure the NT. If the CRL is greater than 84.0 mm, the second trimester quadruple test should be offered. The first trimester combined test allows earlier decision making for parents.

3.2 2nd Trimester Screening for Down's syndrome and Spina Bifida

The quadruple test uses maternal age and four biochemical markers measured from 14 weeks 2 days until 20 weeks 0 days - AFP, hCG (total, intact or free beta subunit), uE3 and Inhibin-A. Although this combination of markers has a lower detection and standardised screen positive rate than the combined test, it is the nationally recommended screening strategy in the second trimester. The optimum time for testing in the second trimester is around 16 week's gestational age. There will always be a need for a screening test in the second trimester for those women who book too late for first trimester testing or when an NT measurement cannot be obtained (despite twice on the couch) in the first trimester. An ultrasound scan will be required to date the pregnancy and a fetal head circumference is there commended measurement used for women presenting in the second trimester.

3.3 Vanishing twins

When ultrasound shows there is an empty second pregnancy sac, the biochemical markers appear no different to those in a singleton pregnancy and the combined test of NT, PAPP-A and free beta HCG can be used to calculate the risk.

When ultrasound shows that there is a second sac containing a non-viable fetus (sometimes called 'vanished' twin), it is possible there could be a contribution to the maternal biochemical markers for many weeks. It is recommended that in this event services undertake the risk calculation based on the maternal age and nuchal translucency only (ie without biochemistry).

3.4 Screening in twin pregnancies

Women with a twin pregnancy are eligible for combined screening or quadruple screening dependent on gestational age.

3.5 Screening in Milton Keynes University Hospital NHS Foundation Trust

In Milton Keynes University Hospital NHS Foundation Trust screening is primarily offered in the first trimester using the Combined Test. For women who miss the first trimester screening window or in whom it has not been possible to obtain an adequate nuchal translucency measurement, second trimester screening by Quadruple Test will be offered.

3.6 Roles and responsibilities

3.6.1 Community midwives

- To discuss screening with women and their families at the booking appointment and ensure clients have a copy of "Screening tests for you and your baby" issued by the National Screening Committee.
- Document in the notes in the appropriate section that screening has been discussed and if a client has either declined or accepted for both T21 and T13/T18.
- Refer **all** women for a dating scan to be performed when the woman is 12/40 gestation from LMP, by sending a request card to the Ultrasound scan department.
- If the woman has booked late and is over 14+1/40 gestation to offer 2nd Trimester screening as stated above. If accepts 2nd Trimester screening:

- Obtain an accurate weight from the client on the day of the scan/blood sample.
- With consent take a 3.5ml sample in a gold top tube; ensure the tube and request form is labelled clearly with name (first name and surname), date of birth, hospital number, date sample taken and sign.

3.6.2 Sonographers

- Ensure the woman has made an informed decision and it is documented by the community midwife that screening has been discussed and accepted.
- If from dating scan, the fetus measures greater than 84mm or have been unable to obtain a NT measurement, refer back to Community midwife when between 14+2/40 and 20+0/40 gestation.

3.6.3 Sonographer Assistant

- Obtain an accurate weight from the client on the day of the scan/blood sample.
- Ensure the rest of the request form is fully completed with a copy of the scan report stapled to the request form.
- With consent take a 3.5ml sample in a gold top tube; ensure the tube is labelled clearly with name (first name and surname), date of birth, hospital number, date sample taken and sign.

The rest of the roles and responsibilities apply to both 1st and 2nd Trimester screening.

3.6.4 Pathology (MK)

- To spin samples prior to transporting to Oxford.
- Safely package samples to prevent damage to tube during transit.
- Send daily list of samples sent to Oxford to PNS.

3.6.5 Screening Laboratory

- To inform PNS of any incorrect or missing details via Lifecycle link.
- To inform Screening Co-ordinator of an abnormal result.
- For low risk results – to send letter to client informing of their result**.**
- Send a "midwife" result letter to PNS for all low risk results.

3.6.6 Prenatal Screening Department

- To obtain a list from MK laboratory of samples sent to the screening laboratory each day.
- To check the lists against Lifecycle to ensure that all samples have been received in the Oxford laboratory and details are correct.
- Check daily for missing information and complete as appropriate with correct information - request from Community Midwives.
- To observe daily for abnormal results (however it is also the laboratory's responsibility to inform the Prenatal Screening Co-ordinator of an abnormal result).
- If abnormal result, arrange appointment within 3 working days for counselling with one of the PNS team. Send letter to community Midwives office to be hand delivered.

See flow chart in Appendix 1 for further information.

4.0 Statement of evidence/references

References:

[1] NHS Screening Programmes (2015) Fetal Anomaly Screening Programme. Standards 2015-2016

[2] NHS Screening Programmes (2015) Fetal Anomaly Screening Programme. Programme Handbook June 2015

External weblinks: Please note that although Milton Keynes University Hospital NHS Foundation Trust may include links to external websites, the Trust is not responsible for the accuracy or content therein.

5.0 Governance

5.1 Record of changes to document

Version number: 3		**Date:** 02/2016		
Section Number	**Amendment**	**Deletion**	**Addition**	**Reason**
All			Guidance on Edwards'/Patau's syndromes added	Update
Executive Summary			National Standards table	Update
Appendix 3			Aetiology of Down's, Edwards' & Patau's	Update
		Characteristics Section		Not required
		Other physical problems		Not required

5.2 Consultation History

Stakeholders Name/Board	Area of Expertise	Date Sent	Date Received	Comments	Endorsed Yes/No
Circulated to all Maternity staff on the 3/2/16					

5.3 Audit and monitoring

This Guideline outlines the process for document development will be monitored on an ongoing basis. The centralisation of the process for development of documents will enable the Trust to audit more effectively. The centralisation in recording documents onto a Quality Management database will ensure the process is robust.

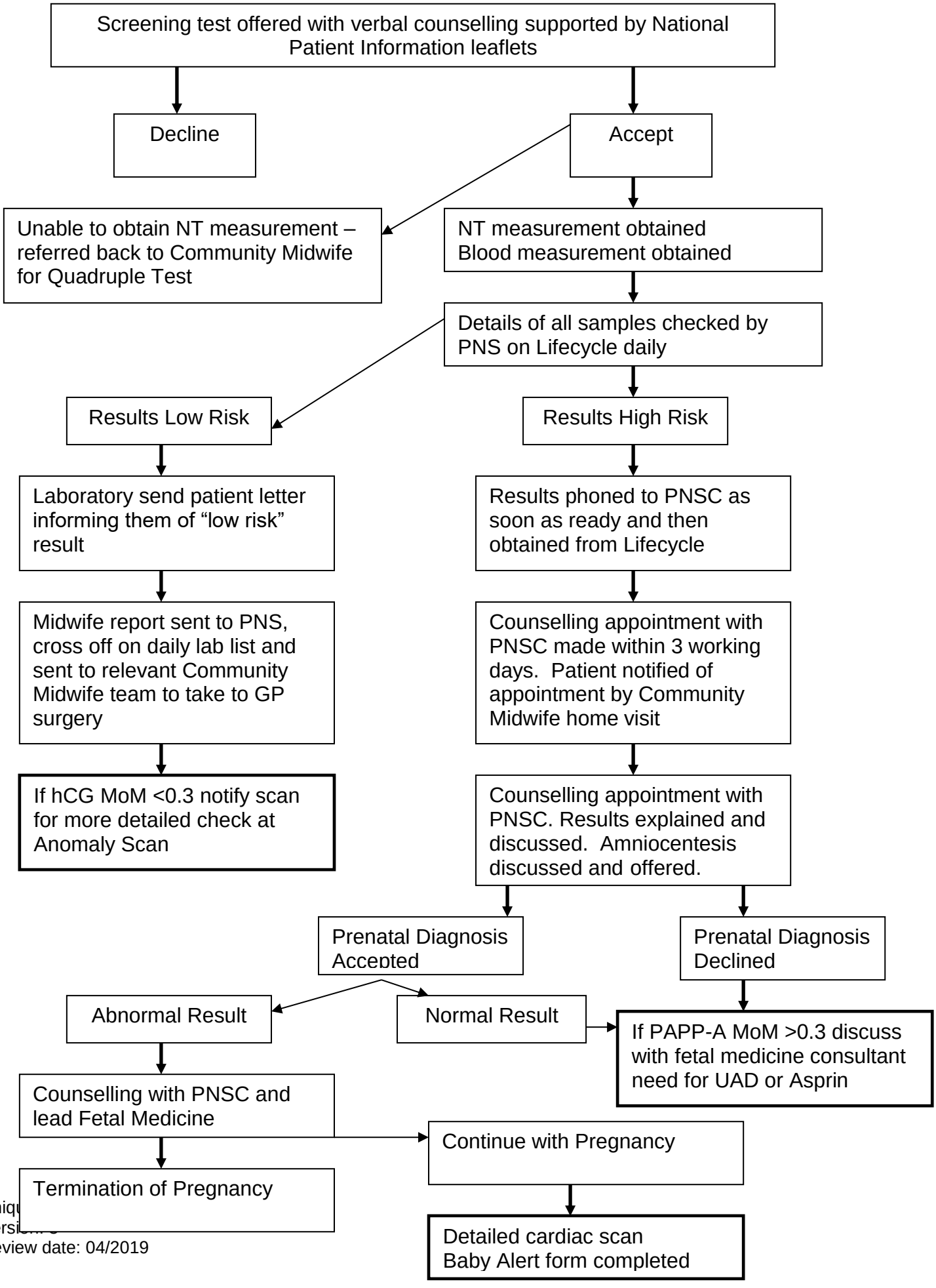
Audit/Monitoring Criteria	Tool	Audit Lead	Frequency of Audit	Responsible Committee/Board
• The uptake of screening • Screen positive rates (SPR) • Detection rate (DR) • False positive rate (FPR) • No of babies born and found to have Down' s syndrome	Oxford Screening Laboratory provides a quarterly report specifically for Milton Keynes, together with an SHA report to compare.	Screening Laboratory Manager	Quarterly	ANSSG committee, PCT, SHA

5.4 Equality Impact Assessment

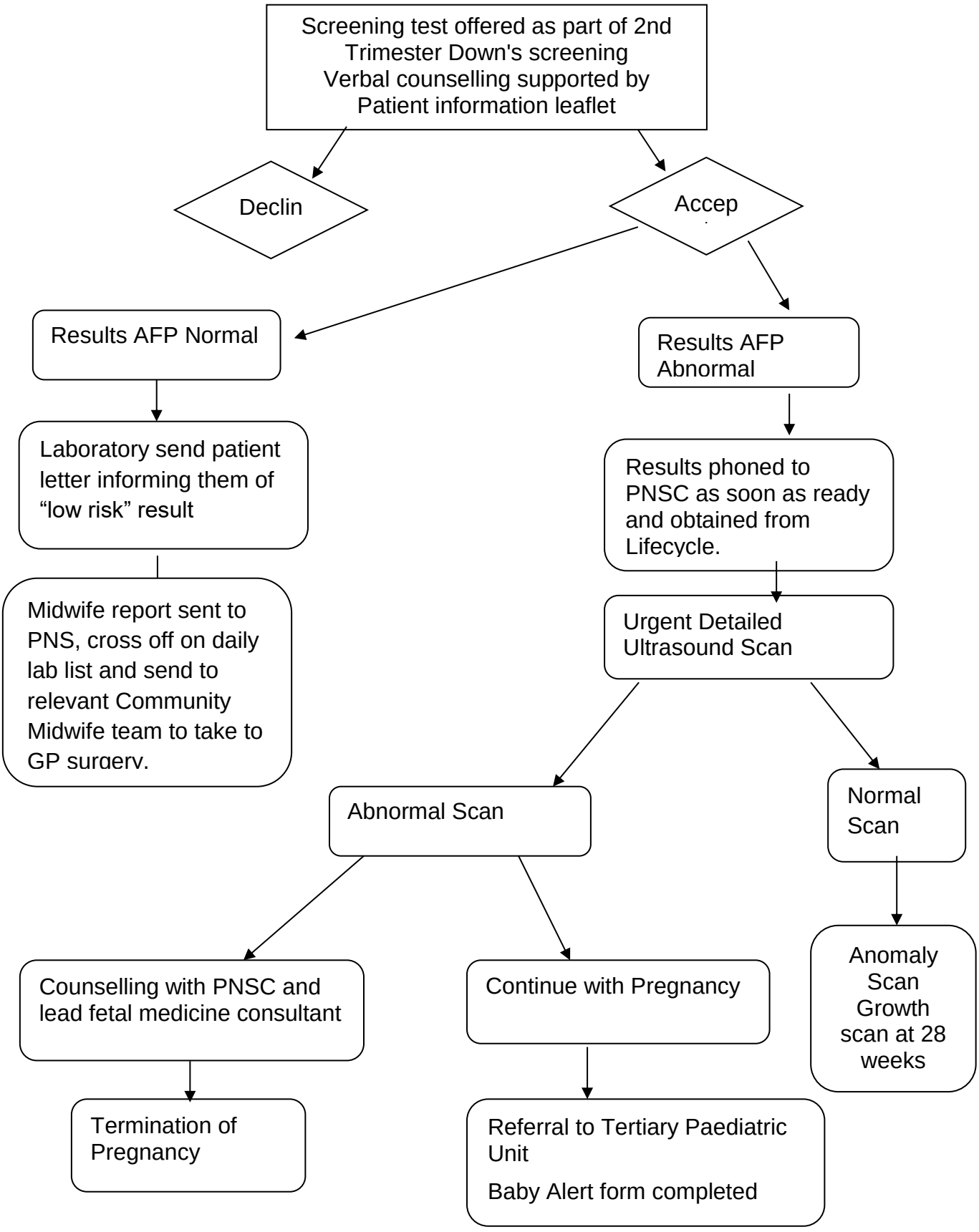
This document has been assessed using the Trust's Equality Impact Assessment Screening Tool. No detailed action plan is required. Any ad-hoc incident which highlights a potential problem will be addressed by the monitoring committee.

Impact	Age	Disability	Sex (gender)	Gender Reassignment	Race	Religion or Belief	Sexual orientation	Marital Status	Pregnancy & Maternity
Do different groups have different needs, experiences, issues and priorities in relation to the proposed policy?	N	N	N	N	N	N	N	N	N
Is there potential for or evidence that the proposed policy will not promote equality of opportunity for all and promote good relations between different groups?	N	N	N	N	N	N	N	N	N
Is there potential for or evidence that the proposed policy will affect different population groups differently (including possibly discriminating against certain groups)?	N	N	N	N	N	N	N	N	N
Is there public concern (including media, academic, voluntary or sector specific interest) in potential discrimination against a particular population group or groups?	N	N	N	N	N	N	N	N	N

Appendix 1: Flow chart for maternal screening for Down's Syndrome



Appendix 2: Flow chart for maternal screening for Neural Tube Defects



Appendix 3: Aetiology of Down's, Edwards' & Patau's

Down's syndrome:

People with Down's syndrome (T21) have extra chromosome 21 in the cells of their body . A baby born with T21 will have a learning disability. They may have communication problems and difficulty managing some everyday tasks. It is impossible to know what level of learning disability a baby with T21 will have. It can vary from mild to severe. Some health problems are more common in people with T21, for example, heart conditions, and problems with the digestive system, hearing and vision. Some problems can be serious but many can be treated. With good healthcare, someone with Down's syndrome is expected to live to around 60 years. People with Down's syndrome have distinctive facial features including almond shaped eyes. Like all children, they also inherit features from their parents. T21 affects 1 in every 1000 births.

- Most cases arise when the chromosomes donated by the mother or father have failed to divide correctly. This type is called Standard or Regular Trisomy 21 and accounts for 95% of people with this condition. Regular Trisomy 21 is not hereditary but it is known from statistical analysis that if a woman has a child with this type of condition then the risk will be higher of it occurring in the next pregnancy.
- Other types of Down's syndrome occur due to translocation of genetic material between chromosome 21 and another chromosome (e.g. Chromosomes 14 and 21 known as Robertsonian translocation). This type occurs in 4% of cases.
- The remaining 1% occurs when there is mosaicism – where normal and Trisomy 21 cells are found within the individual.

Edwards' and Patau's syndromes:

Sadly, most babies with T18 or T13 will die before they are born, be stillborn or die shortly after birth. Some babies may survive to adulthood but this is rare. The correct terminology to use when discussing this with parents is 'life limiting condition.' In Edwards' syndrome (T18) there is an extra copy of chromosome 18 in each cell. All babies born with T18 will have a wide range of problems, which are usually extremely serious - these may include major brain abnormalities. Babies affected by T18 can have heart problems, unusual head and facial features, growth problems and be unable to stand or walk. T18 affects about 3 of every 10,000 births. In Patau's syndrome (T13) there is an extra copy of chromosome 13 in each cell. All babies born with T13 will have a wide range of problems, which are usually extremely serious - these may include major brain abnormalities. Babies affected by T13 can have heart problems, a cleft lip and palate, growth problems, poorly formed eyes and ears, problems with their kidneys and are unable to stand or walk. T13 affects about 2 of every 10,000 births.