NHS
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St Richards, Worthing & Southlands Hospitals

Screening for Down's, Edward's and Patau's Syndrome Guideline

Summary statement: How does the document support patient care?	The purpose of this guideline is to provide evidence based guidance for the process of screening for Down's, Edward's and Patau's syndrome
Staff/stakeholders involved in development:	Antenatal Screening Co-ordinator, Joint Obstetric Guidelines Group, Obstetric Consultants and Senior Midwifery Staff
Division:	Women and Children's
Department:	Maternity
Responsible Person:	Chief of Service
Author:	Antenatal Screening Coordinator
For use by:	Midwives and obstetricians
Purpose:	To provide evidence-based guidance on screening for Down's, Edward's and Patau's syndrome
This document supports:	Screening tests for you and your baby (STFYAYB) GOV.UK August 2022
Key related documents:	**UH Sussex (SRH &WH) Maternity Guidelines:** Antenatal care and patient information, Referral when a fetal abnormality is detected, Maternal antenatal screening tests, Small for gestational age and fetal growth restriction, 12 Week Scan: Combined test and declined combined test ultrasound protocol
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If you require this document in another format such as Braille, large print, audio or another language please contact the Trusts Communications Team	
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Version	Date	Author	Status	Comment
1.0				
2.0	February 2011	Antenatal Screening Coordinator	Archived	New Trust Maternity guideline
3.0	August 2012	Antenatal Screening Coordinator	Archived	Minor update to guideline
3.1	April 2014	Antenatal Screening Coordinator	Archived	3 year update
4.0	May 2015	Antenatal Screening Coordinator	Archived	Additions of Edward's and Patau's syndrome Additions of fail safes for Down's syndrome screening
	August 2017	Antenatal Screening Coordinator	Archived	3 year update
5.1	February 2019	Antenatal Screening Coordinator	Archived	Addition of non- invasive prenatal testing Addition of Monitoring Standards
5.2	October 2019	Antenatal Screening Coordinator	Archived	Amendment of time frame for combined test. Process for unexpected Down's, Edward's or Patau's at birth.
6.0	October 2021	Antenatal Screening Coordinator	Archived	Update to tests offered following national PHE update June 2021. Updates include on-line national info and Trust on-line scan requests. Amendments to audit section.
6.1	September 2022	K. Lundie, Antenatal & Newborn Screening coordinator	Archived	Update of failsafe procedures, sending of results and increased NT pathway. Low PAPP A levels to come in line with other written information. Update to education and training requirements. Removal of torch screen for increased NT. No longer required as part of the fetal medicine investigations
7.0	October 2023	K. Lundie, Antenatal & Newborn Screening coordinator	Live	3 year review

Contents

The interpretation and application of clinical guidelines will remain the responsibility of the individual clinician.
If in doubt contact a senior colleague or expert.

Screening for Down's, Edward's and Patau's guideline

1.0 Aim

- To provide adequate high quality information on the screening process to support each woman and person to make an informed decision on whether to accept or decline the offer of screening.
- To ensure that pre-test information, timeliness of results and offer of pre-natal diagnostic testing are all given in a timescale meeting National Screening Committee (NSC) recommendations.
- Provision of adequate information and support to enable the woman and person and their partner to make a decision on the outcome of the pregnancy.
- For women and people 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.
- To provide optimal care and support for women/people with a confirmed diagnosis as a result of screening who opt for TOP.

2.0 Scope

This guideline applies to all staff involved in consenting pregnant women and people for screening for Down's (T21), Edward's (T18) and Patau's (T13) syndrome and subsequent care.

3.0 Responsibilities

Midwives & obstetricians are expected:

- To access, read, understand and follow this guidance.
- To use their professional judgement in the application of this guideline.

Management are expected:

- To ensure the guideline is reviewed as required in line with Trust and National Recommendations.
- To ensure the guideline is accessible to all relevant staff.

4.0 Abbreviations used in this guideline

T21- Down's syndrome	**T18-** Edward's syndrome
T13- Patau's syndrome	**NIPT-** Non-invasive prenatal testing
IVF- in-vitro fertilisation	**NT-** Nuchal translucency
ARC- Antenatal Result and Choices	**ANC-** Antenatal Clinic
SAFE- St George's Antenatal Fetal Evaluation	**NHSE -** National Health Service England

PND- Prenatal diagnosis	**ASC-** Antenatal screening co-ordinator
CVS- chorionic villus sampling	**MIS -** Maternity Information System
ANST – Antenatal screening team	**NSC -** National Screening Committee
CRL – Crown Rump Length	**FASP –** Fetal Anomaly Screening Programme
MIS – Maternity Information System eg Badgernet	**TOP –** Termination of pregnancy
CMW – Community Midwife	**DNA –** Did not attend
GP – General Practitioner	**SBLv3 –** Saving Babies Lives V3
	MA – Maternity Assistant

5.0 Introduction

This Trust will:

- Offer screening to all eligible women and people in line with ~~national standards~~ NSC standards.
- Provide all women and people with appropriate support and information at all stages of the screening programme. This Information is available in digital format and written if required.
- Process and report on FASP screening tests in a timely manner as detailed in the NHSE Standards.

Screening standards:

- 1st trimester combined test screening for Down's (T21) and Edward's (T18) / Patau's (T13) syndrome is offered between 11+2 and 14+1 weeks, (CRL measurement between 45.0mm and 84mm).
- Second trimester quadruple testing for T21 can be performed from 14+2 to 20 weeks (CRL >84.0mm/HC 101mm and 172mm).
- Combined and quadruple screening can be offered to twin and singleton pregnancies.
- Vanished Twins: If scan shows empty second pregnancy sac the combined test can be offered. If the scan shows a vanished twin (a second sac containing a non-viable fetus) there could be a contribution to the maternal biochemical markers for many weeks. In these cases the NT measurement of the viable fetus will be taken and a chance calculation will be offered from this information alone. It should be noted that this falls outside the scope FASP.
- Non-invasive prenatal testing (NIPT) is offered to women and people with an increased chance combined or quad test result for both singleton and twin pregnancies.
- If women and people access private NIPT prior to the screening window, they will still be offered a scan and combined screening.

6.0 Screening Care Pathway

6.1 Offer of screening

All women and people will be provided information on how to access the digital NHSE screening information 'Screening tests for you and your baby' prior to the booking appointment. This is available in some alternative languages.

The offer of screening and the decision to accept or decline will be recorded in the woman and person's MIS.

The booking midwife should discuss the risks and benefits of screening.

6.2 Consent process

Consent must be obtained for:

- Down's syndrome (T21) only
- Edward's (T18) and Patau's (T13) only
- Down's (T21), Edward's (T18) and Patau's (T13) syndrome.

This should be documented in the woman and person's MIS.

6.3 Screening declined

If disclosed, the reason for decline should be documented in the notes and entered on the Maternity Information System (MIS).

In cases where combined/quad screening is initially consented to and subsequently declined with the sonographer, the woman and person should be referred to the ANST for further counselling and final decision. This final decision should be documented on MIS.

6.4 Screening accepted

- The CT/Dating ultrasound scan is requested by admin team at the time the online referral is received from the women and person via the Trust on-line 'ICE' programme. The CMW will document accepted or declines at time of booking on MIS.
- The appointment will be sent to the woman and person.
- NT scanning is undertaken and sonographer informs woman and person that they will not receive the combined/quad result from scan, and that to complete the test she/they needs to attend ANC for blood test and weight directly following scan.
- Blood is taken with consent, recorded on MIS, and completed test sent to Portsmouth Downs (T21) Syndrome Screening laboratory.

- Woman and person's contact number checked and recorded.
- A list of samples sent is emailed to laboratory, this list forms a tracking and failsafe system for each test; from sending of sample to receipt of result.
- If woman and person fails to attend for blood test following NT, they are contacted by telephone and letter sent outlining timescale for completing test.
- If the pregnancy gestation is outside of combined screening cut-off (>14+1), she/they will be counselled by the ANST and offered quad testing and appointment made if accepted. If the woman and person leaves the department prior to discussing the quad test, they should be contacted by the ANST.
- Following 2 unsuccessful attempts to measure nuchal translucency, quad testing should be explained as recommended in the FASP. This should be documented in MIS.

6.5 Failsafe

- The eligible population (i.e. Down's (T21) and Edward's (T18) / Patau's (T13) screening requested by woman and person) will be identified using the MIS.
- The failsafe administrator/ASC will undertake a weekly check to identify women and people within this eligible group who appear to have not been tested. These cases will be investigated and women and people who have not had screening and who booked after 13+1 weeks will be offered quad testing. Those who booked < 13+1 weeks will be investigated. If the woman and person is found not to be at fault, a Trust funded NIPT will be offered and a datix raised. The offer of NIPT fall outside the scope of the NSC recommendations.
- The 'CRIS' radiology system can be checked daily to match NT scans against blood test attendees.
- Prior to taking screening bloods the MA will check that the woman and person have been booked on MIS. If unbooked ANST are to be notified who will consent and take all booking bloods as well as CT/Quad screening tests The CMW team leaders will be notified and the ANST will update failsafe spread sheet.
- If a woman and person DNA's, their CT/Quad scan apt the ANST they will check the MIS and miscarriage spread sheet to check to see if had miscarriage or TOP. If there is no record the ANST will contact the CMW. If unable to establish why DNA the ANST will arrange and send a repeat appointment to the woman and person either by email, post, call or text. An email will be sent to SG team if there are any concerns.

7.0 Results

7.1 Low chance result

The woman and person will be sent a copy of ~~her/~~their result by letter within 10 working days of the ANST receiving the result from the laboratories. The date of posting is to be recorded on the result sent to the woman and person.

The result (including the PAPP-A MoM) will be recorded within MIS.

The CMW should check with the woman and person at their next appointment that they have received a result in the post and has been recorded in MIS. If not the ANST should be contacted to investigate.

7.2 High chance result

Since 1st June 2021 NHSE recommends the offer of NIPT screening for T21, T18 and T13, following a higher chance result from the combined or quadruple test in singleton and twin pregnancies. A higher chance result is between 1 in 2 and 1 in 150. This Trust uses the SAFE test laboratory at St George's for NIPT testing.

As part of the NHS FASP care pathway NIPT screens for T21, T18 and T13 and will not screen for other chromosomal conditions or assess the baby's sex.

Following a higher chance result from the combined or quadruple test, the woman and person will be contacted and counselled over the phone and/or offered an appointment within 3 working days to discuss the result. The following choices should be offered:

- No further testing
- NIPT screening
- Prenatal diagnosis (PND), such as chorionic villus sampling (CVS) or amniocentesis. Note: Where a woman and person is having a CVS or amniocentesis and taking low dose heparin (dalteparin), this must be omitted on the day of the procedure; and can be re-commenced the following day.

For women and people choosing to have NIPT, the options are:

- T21, T18 and T13
- T21 only
- T18 and T13 only

NIPT screening will report individual chance results for T21, T18 and T13. These results are reported as either lower chance or higher chance; a numerical value is not reported. This test can be performed up to 21+6 as part of the FASP pathway.

In twin pregnancies, the higher chance result report should state that one or both babies may have the condition screened for.

Women and people should expect their NIPT results around 2 weeks from sample collection.

During this discussion, women and people should be informed that the laboratory may keep NIPT screening samples for quality assurance (for example, validation) and testing development purposes for up to 5 years. The healthcare professional should inform the laboratory

if a woman and person does not want their sample to be kept for these purposes. These discussions must be recorded in the woman and person's MIS.

Women and people with in-vitro fertilisation (IVF) or donor egg pregnancies are eligible for the offer of NIPT. The relevant details must be recorded accurately on the NIPT screening request form.

Women and people receiving a higher chance or 'no result' NIPT result should attend an appointment in ≤ 3 working days of maternity services receiving the result (FASP NIPT-S04).

This appointment should be face to face or virtual, depending on the woman/person's choice, and discuss the option of:

- No further testing.
- Having PND, which should be completed in ≤ 3 working days of the woman and person receiving the NIPT result (FASP NIPT-S05).

The result should be given by the ANST who has knowledge of NIPT and the ~~NHS~~ FASP pathway. This discussion, including decisions, should be documented in the woman and person's MIS. The discussion should include that if accepting PND for an increased chance result with a normal NT (less than 3.5 mm) a PCR result for T21, 13 & 18 may only be issued. (CVS samples are processed by other Trusts so we are bound by their guidelines for offering array CGH or PCR only).

Written information or link for NHSE digital information about all procedures should be given.

Contact number for support group Antenatal Result and Choices (ARC) should be offered.

Where an affected pregnancy is confirmed, the CMW and GP should be informed by the ANST.

7.3 Nuchal measurement equal to or above 3.5 mm

Complete combined test to assess chance for T21, T18 & T13.

ANST will discuss with woman/person subsequent management of raised nuchal translucency regardless of combined test result. This includes:

- Offer of NIPT if CT results come back as an increased chance. Woman and person should be advised will that this is not a diagnostic test and only screens for T21/18/13. No other chromosome abnormalities, cardiac or underlying genetic conditions can be ruled out.
- Referral for specialist fetal cardiac scanning (<4mm fetal echo arranged between 21-23 weeks' gestation; >4mm fetal echo arranged between 16-18 weeks' gestation
- Result is entered on MIS system and maternal and birthing parent alert added.
- Offer referral to fetal medicine consultant.

8.0 Process for unexpected Down's, Edwards's or Patau's syndrome at birth

In cases where a baby is born with T21, T13 and T18 syndrome following low chance combined or quad screening, the following processes should happen:

- Notify Portsmouth screening laboratory.
- Complete DATIX.
- Notify National Congenital Anomaly and Rare Disease Registration Service.
- Review of nuchal translucency and anomaly scans.
- Offer postnatal follow up with fetal medicine consultant.

9.0 Low PAPP-A

An incidental finding of low PAPP-A (≤ 0.41 MoM) will be identified by the ANST. The women and person and GP will be notified to commence aspirin as per SBLV3 recommendations. UA dopplers will be request at time of anomaly scan by the ANST. Subsequent growth scan will be requested in line with the Trust 'Small for gestational age and Fetal growth restriction guideline'

10.0 Education and training

Midwives are required to complete annual training. This is monitored by the practice development team who can be contacted for further information.

11.0 Monitoring and Audit

Suggested audit questions:

- The number of women and people with nuchal translucency ≥3.5 mm where the result was reported by the lab within 3 days of receipt of the sample
- The number of women and people with nuchal translucency ≥3.5 mm with documented evidence that the woman and person was offered further testing within 3 working days of receipt of the result
- The number of women and people with nuchal translucency ≥3.5 mm who were referred for fetal echo by 23 weeks
- This guideline is also monitored through the quarterly and annual NHSE Key Performance Indicators (KPIs).

References

Screening tests for you and your baby (STFYAYB) - GOV.UK (www.gov.uk 2022)

Screening tests in pregnancy - NHS (www.nhs.uk 2021)

NHS Fetal Anomaly Screening Programme (FASP): programme overview - GOV.UK (www.gov.uk 2021)

Fetal anomaly screening programme handbook - GOV.UK (www.gov.uk 2023

Fetal anomaly screening programme: standards - GOV.UK (www.gov.uk 2021)

Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome - NHS (www.nhs.uk 2021)

CVS and amniocentesis diagnostic tests: description in brief - GOV.UK (www.gov.uk 2021)

Appendix 1: T13, 18 and 21 Failsafe