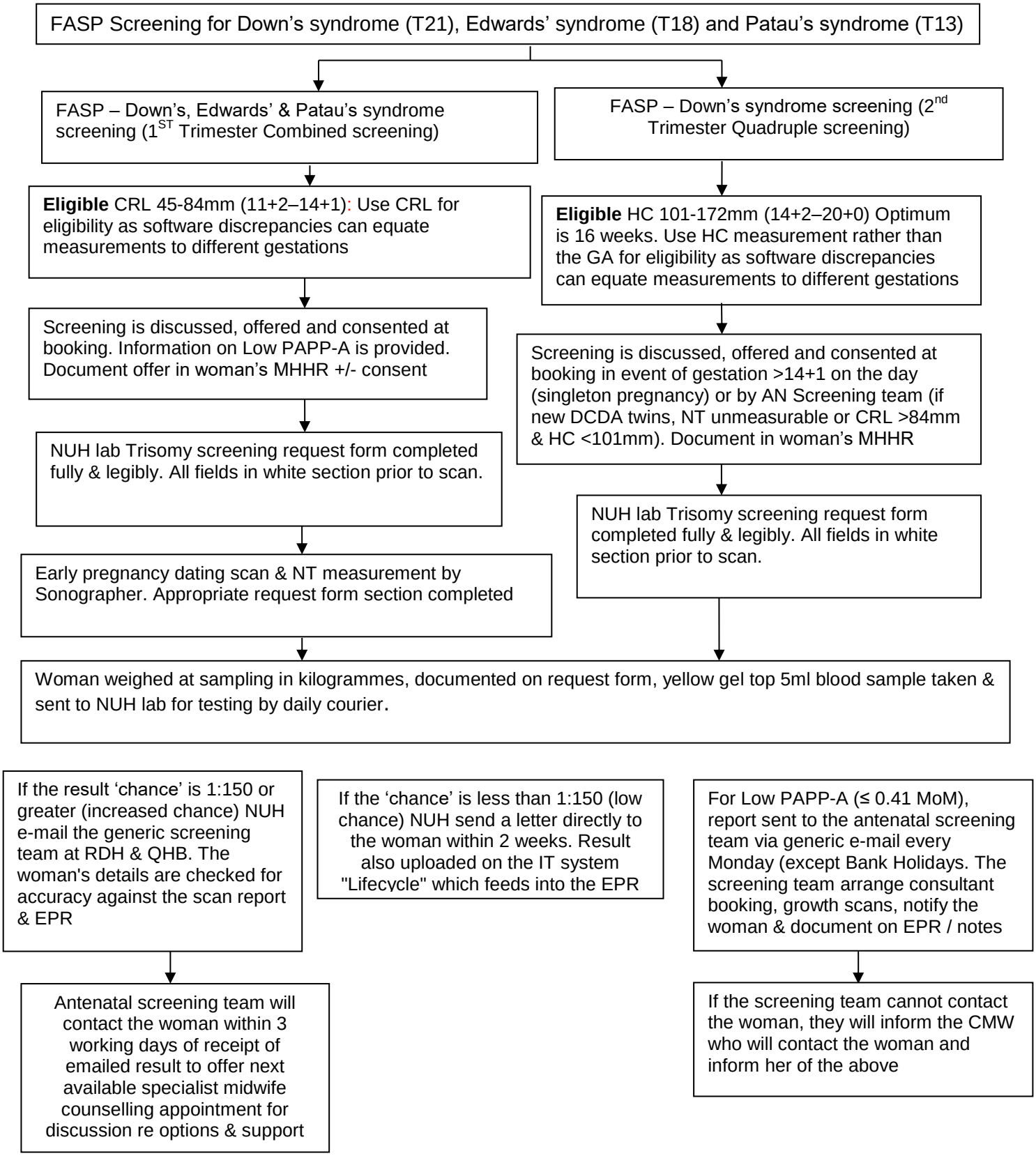
General Pathway for Down's Syndrome, Edwards' Syndrome and Patau's Syndrome Screening in Pregnancy - Full Clinical Guideline

Reference No.: Maternity/07:2023/D4



1. Introduction

Antenatal screening aims to enable parents to make informed choices concerning their pregnancy outcome, through the timely offer of screening. National standards have been set by the UK National Screening Committee (UK NSC) and NHS England (NHSE) in an attempt to ensure a more coordinated approach to screening. Policy and standards are subject to review and development in the light of new evidence.

The NHS Screening programme: Fetal Anomaly Screening Programme (FASP) Standards (FASP, 2021) aim 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.

The following document relates to the Down's syndrome (T21), Edwards' syndrome (T18) and Patau's syndrome (T13) screening programme currently offered by University Hospitals of Derby and Burton- (UHDB) in line with the FASP. The service is bench marked to the NHS Fetal Anomaly Screening Programme - Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome (NHSE, 2021) or any subsequent updated versions.

2. Purpose and Outcome

- Outline the aetiology and incidence of Down's syndrome, Edwards' syndrome and Patau's syndrome including some characteristics and physical differences associated with these three distinct conditions
- Define the current minimum standards for antenatal screening for Down's syndrome, Edwards' syndrome and Patau's syndromes at UHDB
- Provide flowcharts to use in practice to inform practitioners of all the care pathways within the Down's syndrome, Edwards' syndrome and Patau's syndrome screening programmes at UHDB
- Inform practitioners of the Down's syndrome, Edwards' syndrome and Patau's syndrome screening education, training, audit and monitoring mechanisms at UHDB.

3. Abbreviations

AFP	-	Alpha - fetoprotein
AN	-	Antenatal
ANNB	-	Antenatal and Newborn
ANC	-	Antenatal Clinic
AVSD	-	Atrial Ventricular Septal Defect
BSL	-	British Sign Language
CMW	-	Community Midwives
CLC	-	Consultant-led care
CRL	-	Crown Rump Length
DR	-	Detection Rates
EPR	-	Electronic Patient Record
FASP	-	Fetal Anomaly Screening Programme
FM MW	-	Fetal Medicine Midwife
FMMC	-	Fetal and Maternal Medicine Centre
FPR	-	False Positive Rates
HCG	-	Human Chorionic Gonadotrophin
IVF	-	Invitro Fertilisation
MW	-	Midwife
NCARDRS	-	National Congenital Abnormality and Rare Disease Register
NDSCR	-	National Down's syndrome Cytogenetics Register

NHSE	-	NHS England
NIPT	-	Non Invasive Prenatal Testing
NT	-	Nuchal Translucency
PAPP A	-	Pregnancy Associated Plasma Protein A
QA	-	Quality Assurance
QHB	-	Queens Hospital Burton
RDH	-	Royal Derby Hospital
SOFT	-	Support Organisation for Trisomy 18/13
SQAS	-	Screening Quality Assurance Service
UE3	-	Unconjugated E3
UHDB		University Hospitals of Derby and Burton
UK NSC	-	United Kingdom National Screening Committee

4. Aetiology of Down's Syndrome

a) The nuclei of most human cells contain 46 chromosomes. In Down's syndrome, cells contain an extra copy of chromosome number 21 resulting in 47 chromosomes in total. The extra genetic material gained from this gives the characteristics of Down's syndrome also known as 'Trisomy 21' (T21). Most cases (95%) arise when the chromosomes donated by the mother or father have failed to divide correctly.

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 chance will be ~1% 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 (this occurs in 4% of cases). The remaining 1% occurs when there is mosaicism, where normal and Trisomy 21 cells are found within the individual.

b) Incidence

Down's syndrome occurs in approximately 1:1000 births. This figure is similar in all populations and is an overall population chance; it affects both boys and girls equally.

All women have a chance of having a baby with Down's syndrome and this chance increases with age. The older a mother, the more chance she has of having a baby with the condition.

c) Effects of Down's Syndrome

Some physical differences

- Learning disability – wide spectrum. Approximately 20% of children with Down's syndrome have mild learning difficulties, 70% have severe learning difficulties and 10% have profound learning difficulties
- Developmental delay
- 40-50% of children suffer with a congenital heart conditions, more commonly complete AVSD. Cardiac surgery can be less tolerated in children with Down's syndrome
- Over 50% have significant hearing impairment, sensorineural and/or conductive loss
- Gastrointestinal tract abnormalities
- Hypotonia and poor feeding.

These characteristics and physical problems are not exhaustive and if families require further information, please refer them to the Down's syndrome Association (www.dsa-uk.com) and/or a senior paediatrician.

5. Aetiology of Edwards' Syndrome

a) Edwards' syndrome (Trisomy 18) arises when human cells contain an extra copy of chromosome number 18. Edwards' syndrome occurs by the same mechanisms as Down's syndrome (see section 4a).

b) Incidence

About 1 in every 1,500 pregnancies is diagnosed with Edwards' syndrome (SOFT, 2017). In the absence of any prenatal detection programme Edwards' syndrome occurs in approximately 1:7900 births. The incidence increases with increasing maternal age (NHS, 2022).

c) Effects of Edwards' Syndrome

For live-born infants, Edwards' syndrome is usually of far greater clinical severity than Down's syndrome, having a limited lifespan, and often with multiple congenital conditions. About 50% of infants die within the first two weeks after birth, often from central apnoea or congenital conditions. Around 8% of infants with Edwards' syndrome will survive beyond one year, but with severe learning disabilities. Growth restriction (both prenatal and postnatal) is usual. The median life expectancy of these Infants is 14 days (NHS, 2022).

6. Aetiology of Patau's Syndrome

a) Patau's syndrome (Trisomy 13) arises when human cells contain an extra copy of chromosome number 13 (Trisomy 13). Patau's syndrome occurs by the same mechanisms as Down's syndrome (see section 4a).

b) Incidence

About 1 in every 5,000 pregnancies is diagnosed with Patau's syndrome (SOFT, 2017). In the absence of any prenatal detection programme the occurrence in live births is about 1 in 9,500. The incidence rises with increasing maternal age (NHS, 2022).

c) Effects of Patau's Syndrome

For live-born infants, Patau's syndrome is usually of far greater clinical severity than Down's syndrome, having a limited lifespan, and often with multiple congenital conditions. More than 50% of infants die within one month of birth. Around 8%-10% of infants with Patau's syndrome survive beyond one year, but typically with severe learning disabilities. Long-term survival is most likely due to mosaicism (where the extra chromosome is not present in all cells), or due to only part of chromosome 13 being involved (partial trisomy 13) (NHS, 2022).

If families require further information regarding Patau's syndromes, please refer them to the Support Organization for Trisomy 13/18 (www.soft.org.uk) and/or a senior paediatrician.

7. Low PAPP-A

As part of the 1st trimester Combined screening test for Down's syndrome, Edwards' syndrome and Patau's syndrome, blood samples are analysed for the biochemical marker Pregnancy Associated

Plasma Protein-A (PAPP-A). A low level (≤ 0.41 MoM) of the first trimester marker PAPP–A should be considered a risk factor a small for gestational age (SGA) neonate as per the SGA risk-assessment tool.

Low PAPP–A results are sent to the ANNB screening team generic email by NUH Trisomy screening lab on working Mondays and the following actions are completed:

- Contact the woman to explain the results and care plan
- Refer to the information in the MHHR and sign-post to digital information leaflet
- Send by email / post the UHDB Low PAPP-A patient information leaflet. See appendix A
- Where possible arrange a CLC booking to follow the FASP 20-week anomaly scan appointment
- If not possible for a 20-week CLC appointment, arrange a 30-week growth scan appointment with Consultant Obstetrician appointment to follow
- If already on the Small for Gestational Age (SGA) pathway, serial scans will already be arranged, the screening team document on EPR
- Update the local FASP / Low PAPP-A spreadsheet, which kept on the UHDB shared drive
- Change the woman's pregnancy status on the EPR to Consultant-led care
- If the screening team are unable to contact the woman, they ask the CMW do so and to:
  - Inform the woman, explain the results and care plan
  - Give / sign-post to the Low PAPP-A leaflet (see appendix A) at the 15-16 week follow up appointment
  - Recommends transfer to consultant led care (CLC) and gives the growth scan / CLC appointment as per list of actions above.

8. Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome offered at UHDB

a) The 1st Trimester Combined Screening Test

This is offered between 11+2 and 14+1 weeks gestation. Eligibility for first trimester screening is determined at the dating/Nuchal Translucency (NT) scan and can be performed where the CRL measures between 45mm-84mm and the NT measurement is obtainable. The 1st Trimester Combined Screening test enables women with singleton and twin pregnancies to choose screening for:

- Down's syndrome (T21)
- Edwards' syndrome and Patau's syndrome (T13/18)
- Down's syndrome, Edwards' syndrome and Patau's syndrome (T21/18/13)

The Combined screening method involves an ultrasound scan (US) performed by a DQASS accredited Sonographer to confirm viability, estimated due date (EDD) and to obtain the CRL and NT measurements. This is immediately followed by a blood test to quantify the biochemical markers: Pregnancy Associated Plasma Protein-A (PAPP-A) and free Beta Human Chorionic Gonadotrophin (free βhCG).

The chance of a pregnancy being affected by these conditions (depending upon screening choices) is calculated by specifically designed software (currently at the NUH Trisomy Screening Lab) using maternal age, NT measurement, biochemical markers. There are software adjustments for the effect on the maternal biochemical markers of ethnic origin, smoking status, +/-IVF pregnancy, +/- IDDM, and +/- any previous pregnancy affected by T21, T18, T13 or a neural tube defect (NTD).

If the CRL is >84mm, but the head circumference (HC) is between 101mm-172mm the woman should be offered 2nd trimester Quadruple screening.

The 'chance' cut-off for offering further counselling and offer of Non-Invasive Prenatal Testing (NIPT) or diagnostic testing is set nationally to 1 in 150 for all of the conditions screened for. Please note one 'chance' result is given for Down's syndrome and a separate combined 'chance' result is given for both Edwards' and Patau's syndrome jointly. See care pathway flow chart A and B.

b) The 2nd Trimester Down's syndrome Quadruple Screening

The second trimester Quadruple Screening test enables women with singleton and twin pregnancies (see section 8d) to opt for screening for Down's syndrome between 14+2 and 20+0 weeks gestation (**eligibility is confirmed by obtaining a HC measurement of 101mm-172mm)**. If the HC measures >172mm the woman is not eligible for screening as the pregnancy is too late for second trimester screening (irrespective of gestation). This should be documented in the MHHR and EPR.

2nd Trimester screening is a blood test which enables the quantification of the biochemical markers Alpha-Feta Protein (AFP), beta hCG, uE3 and Inhibin A. The chance of a pregnancy being affected by Down's syndrome is calculated by specifically designed software at NUH Trisomy screening lab using the above biochemical markers and maternal age. There are software adjustments for the effect on the maternal biochemical markers of ethnic origin, smoking status, +/-IVF pregnancy, +/-IDDM, and +/- any previous pregnancy affected by T21, T18, T13 or a neural tube defect (NTD). The chance cut-off for offering further counselling and diagnostic testing is 1:150.

9. Screening in twin pregnancies

a) 1st Trimester Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome in Twin Pregnancies

Women with a new diagnosis of a twin pregnancy at the dating scan appointment who are eligible and have consented to 1st trimester Combined trisomy screening should be referred to the screening team by the sonographer for a discussion with a midwife with an understanding of screening in twin pregnancies. The accuracy, detection rate and type of result received is different depending on chronicity of the twin pregnancy.

For women screened using the Combined test, where a dichorionic twin pregnancy is identified the chance will be reported for each baby. In a monochorionic twin pregnancy, the chance result is the same for each baby and one 'pregnancy' chance result is reported.

Monochorionic twins

The performance of 1st trimester screening for T21/18/13 in monochorionic twins is comparable to that in singleton pregnancies: a detection rate of 80% for a standardised screen positive rate of 2.5%. The Fetal and Maternal Medicine Centre (FMMC) at RDH counsel the monochorionic twin pregnancies regarding screening and arrange future appointments. At QHB, the ANNB MW counsel monochorionic twin pregnancies then refer/transfer care to FMMC at RDH

Dichorionic Twins

In a dichorionic twin pregnancy, where one is affected and the other unaffected, the performance of the 1st trimester Combined screening test is slightly reduced due to the biochemical blood test markers being less discriminatory. It performs better than 2nd trimester Quadruple screening due to the individual NT measurements. Recent data are not available to state a specific detection rate for

a standardised screen positive rate in this scenario. The Screening teams counsel DCDA twins for screening and ANC midwives arrange consultant appointments.

b) Second Trimester Screening for Down's Syndrome in Twin Pregnancies

The Quadruple test is offered to a woman with a twin pregnancy to screen for T21 only when one or both of the:

- Woman presents later than 14+1 weeks gestation
- NT measurements cannot be obtained after two reasonable attempts
- CRL measurements are greater than 84.0mm on the day of the scan

The Quadruple screening test in twin pregnancies is not as sensitive as the combined test and the decision-making process can be more difficult for a number of reasons. Women considering the Quadruple test should have a discussion with a healthcare professional with a special interest, experience and knowledge of managing multiple pregnancies. This is to help support personal informed choice.

Women with twin pregnancies who are eligible for second trimester screening should be referred to the screening team by their CMW, the sonographer or the ANC midwife for a discussion with a midwife with an understanding of screening in twin pregnancies. The accuracy, detection rate and type of result received is different depending on chronicity of the twin pregnancy.

Monochorionic Twins
The performance of the quadruple test in monochorionic twins is comparable to that in singleton pregnancies.

The chance of a T21 birth from a monochorionic pregnancy is lower than that from a singleton pregnancy due to a higher fetal loss rate amongst affected pregnancies.

Dichorionic Twins
The chance of a T21 birth of at least one baby from a dichorionic twin pregnancy is higher than that from a singleton pregnancy. In dichorionic twins, where one is affected and the other unaffected, the performance is less sensitive due to the biochemical markers being less discriminatory.

Quadruple screening test chance results for dichorionic twins relate to the pregnancy not to individual babies as the individual scan measurements are not used as markers. They are not interpreted in the usual way but used, with a cut-off of 1 in 150 at term, to define a higher chance group. An appropriately trained healthcare professional should interpret and explain these results and pregnancy options due to the complexities involved.

'It should be noted that the 'chance' approach used in calculating a quadruple twin pregnancy 'chance' is referred to as a 'pseudo-chance'. This is the established methodology currently available and simply means that the chance would be accurate in predicting a false-positive rate (which relates only to the marker distributions in unaffected twin pregnancies). The 'chance' is a pregnancy related 'chance' is not fetal specific. The term chance cut-off of 1 in 150 is applied to the 'pseudo-chance'. Because the calculation of chance's in twin pregnancies relies on limited evidence and assumptions the chance estimate should be interpreted by suitably experienced practitioners`.

10. Guidance for Down's, Edwards' and Patau's Syndrome Screening in the Event of a 'Vanished' Twin

NHS FASP are currently reviewing guidance on screening in vanished twins. The definition of a vanished twin is when one fetus in a twin pregnancy is non-viable. It may be partially or completely reabsorbed.

An ultrasound scan within the screening window for the Combined or Quadruple test may show either:

- an empty second pregnancy sac
- a second pregnancy sac containing a non-viable fetus.

Empty second pregnancy sac and the combined test

When there is an empty second pregnancy sac, the Combined test can be used to calculate the chance result.

Second pregnancy sac containing a non-viable fetus and the combined test

When there is a second pregnancy sac containing a non-viable fetus, the Combined test should not be used to calculate the chance result. Local guidance should be in place for the specialist clinical management of these pregnancies. For example, use of NT measurement and maternal age alone. This is not part of the FASP pathway.

The Quadruple test

The quadruple test can be offered to a woman with a vanished twin pregnancy depending on the gestation the twin pregnancy and vanished were identified. Please refer to national FASP guidance and local Trisomy Screening Lab pathways.

OUS will have appropriate policies, procedures and guidelines in place, in line with national and professional guidance to ensure a high quality standard of care for all women. The professional lead for this is the clinical ultrasound manager for Obstetric Ultrasound, with support from the Screening Support Sonographer and deputy.

11. Education and Training

- Education and training is achieved via mandatory professional study day sessions on an annual basis. FASP e-learning is completed as required, which is role-dependent.
- Workshops are also scheduled as appropriate to update staff on developments within the screening programme. The e-learning package 'generic cross-programme Antenatal and Newborn screening module' is to be completed as part of mandatory training by all midwives on a triennial basis. Compliance is monitored annually by the screening team. Registration is via the Learning for Healthcare (e-LfH) website http://portal.e-lfh.org.uk/
- All newly employed healthcare professionals involved in the screening process are offered training and the opportunity to work with the specialist midwives in FMMC and ANNB screening team.
- All education and training provided is evaluated and audited.

12. Audit and Monitoring of the Down's, Edwards' & Patau's Syndrome Screening Programme

- The UHDB internal Antenatal and Newborn Screening Programme Board and FMMC Team are responsible for establishing links and enabling quality assurance at a local level in line with national standards. Any issues are escalated via Trust Governance, Divisional Management Team and the Maternity Risk Groups as appropriate.
- Where appropriate (e.g. a suspected or detected abnormality) information is also sent for inclusion in the NCARDRS and the National Down's Syndrome Cytogenetics Register (NDSCR).

- NCARDRS notification forms should be completed contemporaneously, National Disease Registration Service (NDRS) - NHS Digital a copy filed in maternal obstetric notes and sent by secure email with a copy of the scan report to nhsdigital.ncardrsemsy@nhs.net
- The Trust ANNB screening annual audit and report (pertaining to the previous fiscal year is produced by the ANNB Screening Lead Midwives in conjunction with the members of the Antenatal and Newborn Screening Programme Board (approved and signed off by the Director/Head of Midwifery). It is sent to Regional SQAS team at PHE.MidsAndEastQA@nhs.net and shared with NHS England North Midlands Screening & Immunisation Team.

13. Local & Regional Contacts

Royal Derby Hospital Site only:

Antenatal & Newborn Screening Lead Midwife	01332 789924
Antenatal & Newborn Screening Midwives	01332 785435
Antenatal & Newborn Screening Failsafe Officer	01332 785142
Bereavement Specialist Midwife Office	01332 789791
Chair of Antenatal & Newborn Screening Board	Secretary 01332 785204
Clinical Lead Paediatrician: Office	01332 785719
Clinical Specialist Midwives (FMMC)	01332 785409
Duty Consultant Biochemist RDH	01332 789383
Obstetric Ultrasound Office	01332 785326

Queens Hospital Burton only:

Antenatal & Newborn Screening lead Midwife	01283 511511 extension 4297/3100
Antenatal & Newborn Screening Deputy midwife	01283 511511 extension 3100
ANC & Scan reception	01283 511511 extension 4343/4342
Bereavement Specialist Midwife Office	01283 511511 extension 4383

Regional contacts

NHS England North Midlands Screening & Immunisation Team	07783811900
Regional Genetics Centre - City Hospital Nottingham,	01159 627728
Regional Cytogenetics Department	Internal #630 ext 56617
City Hospital Nottingham Cytogenetics	0115 9627617
Sheffield Diagnostic Genetic Services	01142 717009
NUH trisomy screening service	0115 969 1169 ext 76422

Voluntary Sector or Charitable Representatives

- Antenatal Results & Choices (ARC) 020 771 37486 (10 – 5.30pm Mon-Fri)
  Evening service: 8 – 10pm Tues & Thurs (email to arrange a call)
- Down's syndrome Association Helpline 0333 121 2300 (10:00 - 16:00) info@downs-syndrome.org.uk
- Support Organization for T13/T18 (SOFT) 0300 102 7638. email: enquiries@soft.org.uk

13. Resources & Useful Websites

- NHS Screening
  www.screening.nhs.uk
- NHS Choices
  www.nhs.uk/screening
- Fetal Anomaly Screening Programme

https://www.gov.uk/guidance/fetal-anomaly-screening-programme-overview

- Continuous Professional Development
  http://portal.e-lfh.org.uk/
- Antenatal Results & Choices
  www.arc-uk.org
- Down's Syndrome Association
  www.downs-syndrome.org.uk
- Database of Individual Personal Experiences
  www.healthtalkonline.org
- Support Organization for T13/T18 (SOFT)
  http://www.soft.org.uk/

Screening tests for you and your baby digital information (NHSE 2022) has been translated into 12 different languages and there is an easy to read format. There is also an introduction and summary animation with different languages subtitles and BSL. Please see the below links:

- Screening tests for you and your baby (STFYAYB) - GOV.UK (www.gov.uk)
- www.screening.nhs.uk (professionals)
- www.nhs.uk/conditions/pregnancy-and-baby/pages/screening-tests-abnormality-pregnant.aspx (for pregnant women) for online screening information and support).
- Information is also available in an 'easy to read' formats at
  Screening tests for you and your baby: easy guides - GOV.UK (www.gov.uk)

14. References

UHDB Standard Operating Procedure for: *The management of 1st and 2nd Trimester screening blood samples taken in Obstetric Ultrasound / Antenatal Clinic at Royal Derby Hospital for Down's, Edwards' & Patau's Syndromes.* Available on Net-i

Fetal Anomaly Screening Programme standards (FASP, 2021):
https://www.gov.uk/government/publications/fetal-anomaly-screening-programme-standards

Fetal Anomaly Screening Programme handbook (FASP, 2022): Fetal anomaly screening programme handbook:
https://www.gov.uk/government/publications/fetal-anomaly-screening-programme-handbook

NHS England (2022). NHS Public health functions agreement 2022-3. Service specification no. 16. NHS Fetal Anomaly Screening Programme - Screening for Down's, Edwards' ad Patau's Syndromes (Trisomy 21, 18 & 13). Or later versions as appropriate.

NHS National Genetics and Genomics Centre, (2013a). Edwards' Syndrome. [Online] Available from:
National Genetics and Genomics Centre, (2013b), Patau's Syndrome. [Online]. Available from:
Welcome to Genomics Education Programme - Genomics Education Programme (hee.nhs.uk)

Service Specification for Down's, Edwards' and Patau's Syndrome Screening

Service Standards for Fetal Anomaly Screening (Down's, Edwards' and Patau's syndrome) at UHDB are offered in accordance with the FASP Service Specification 2022-2023 or any subsequent version:
NHS Fetal Anomaly Screening Programme (FASP): programme overview - GOV.UK (www.gov.uk)

Pathway A

Key of Responsibilities
- CMW
- Sonographer
- Radiography Assistant
- Fetal Medicine MW/ANSC

Care Pathway for Down's, Edwards' and Patau's Syndrome Screening by the 1st Trimester Combined Screening Test (Singleton pregnancy)

Woman attends midwife for booking 8-10 weeks screening options discussed and signposted to digital information & MHHR QR codes

Box 1

Woman accepts test for
a) Down's
b) Edwards' and Patau's
c) Down's, Edwards' and Patau's

Action by Midwife: Accurately complete NUH screening request form & book dating scan apt with 1st trimester screening (and quad test) document woman's decision in MHHR. Gives request form to woman & inform to bring MHHR with form to the scan apt.

Woman undecided.

Refer "Screening tests for you and your baby".

Still unsure: visit NHS choices website 'antenatal screening' or arrange a discussion with the ANNB MW

Decision must be made before scan appointment. Sonographer cannot discuss options further.

Accepts any of the screening options highlighted in Box 1

Declines Combined screening.

Action by Sonographer: early pregnancy / dating scan only. Document patient's choice to decline 1st trimester Combined screening

Dating scan & if CRL 45-84mm perform NT measurement.

Action by Sonographer: Check woman's consent & correct screening choice on form. Complete US fields on form (i.e. CRL, NT etc). Send woman to phlebotomy room for blood tests (to measure PAPP-A & Free Beta hCG). Copies of US report in HHR & obstetric notes.

If NT unobtainable after 2 attempts offer 2nd trimester screening

If uncertain of woman's consent or choices refer to ANNB MW

Action by Sonographer: If NT measurement is >= 3.5mm, complete blood test & refer woman to FMMC MW at RDH site. QHB site refer to FMMC at RDH (see pathway D)

Blood collection. (Yellow top bottle) for collection of markers PAPP-A & Free Beta hCG

Action by Radiography Assistant: Weigh the woman at sampling in Kg & enter on request form with date & time of sample & signature. Check **all fields** complete & correct.

If CRL <45mm (too early for screening), woman is referred by sonographer to rebook apt scan within the screening window

Sample/Request dispatched to lab.

Action by Radiography Assistant: E-mail the screening team the daily worklist of women screened, to NUH lab.

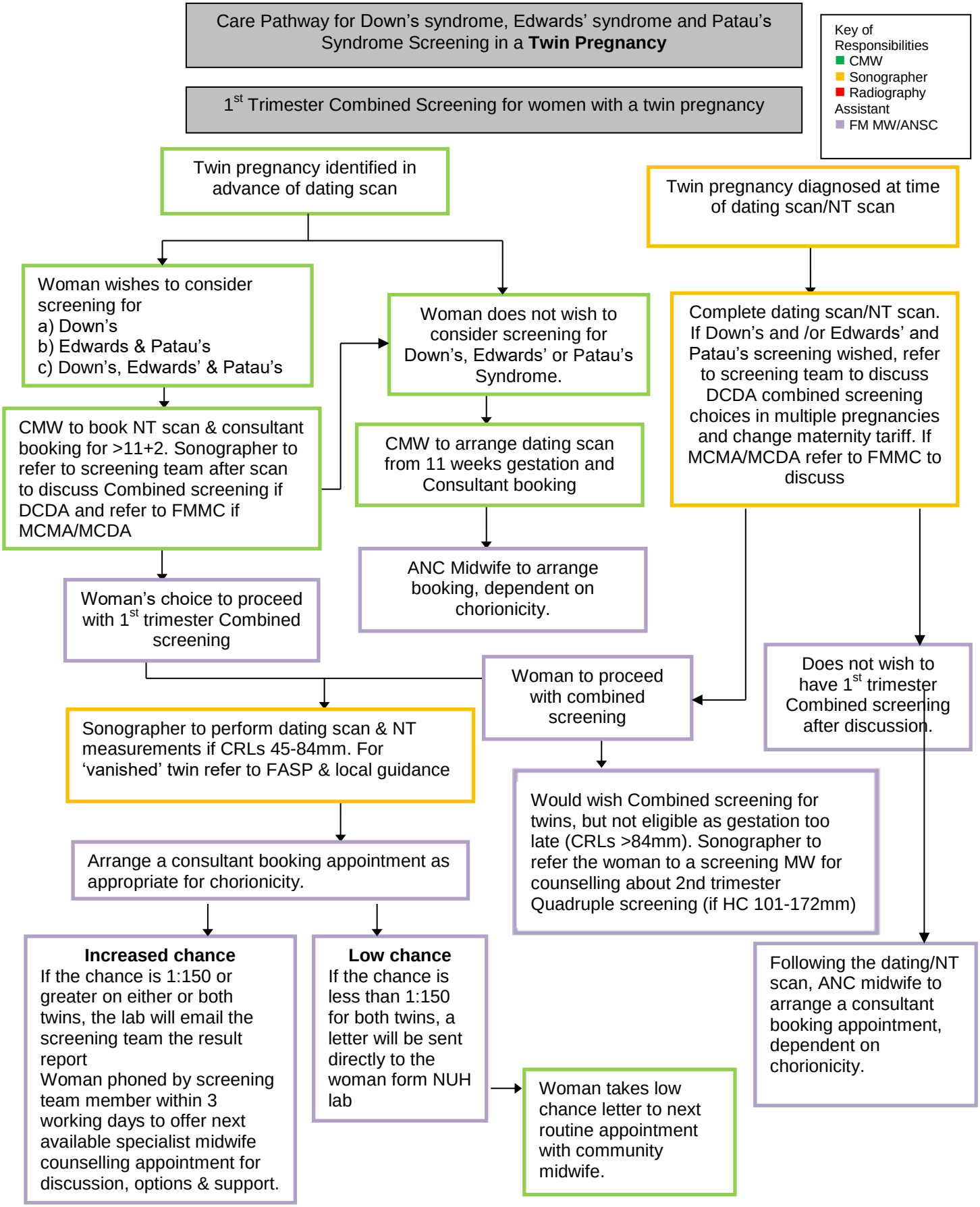
If the chance is 1:150 or greater (increased chance) then the lab will email report to the screening team

If the chance is less than 1:150 (low chance) a letter will be sent directly to the woman from NUH lab

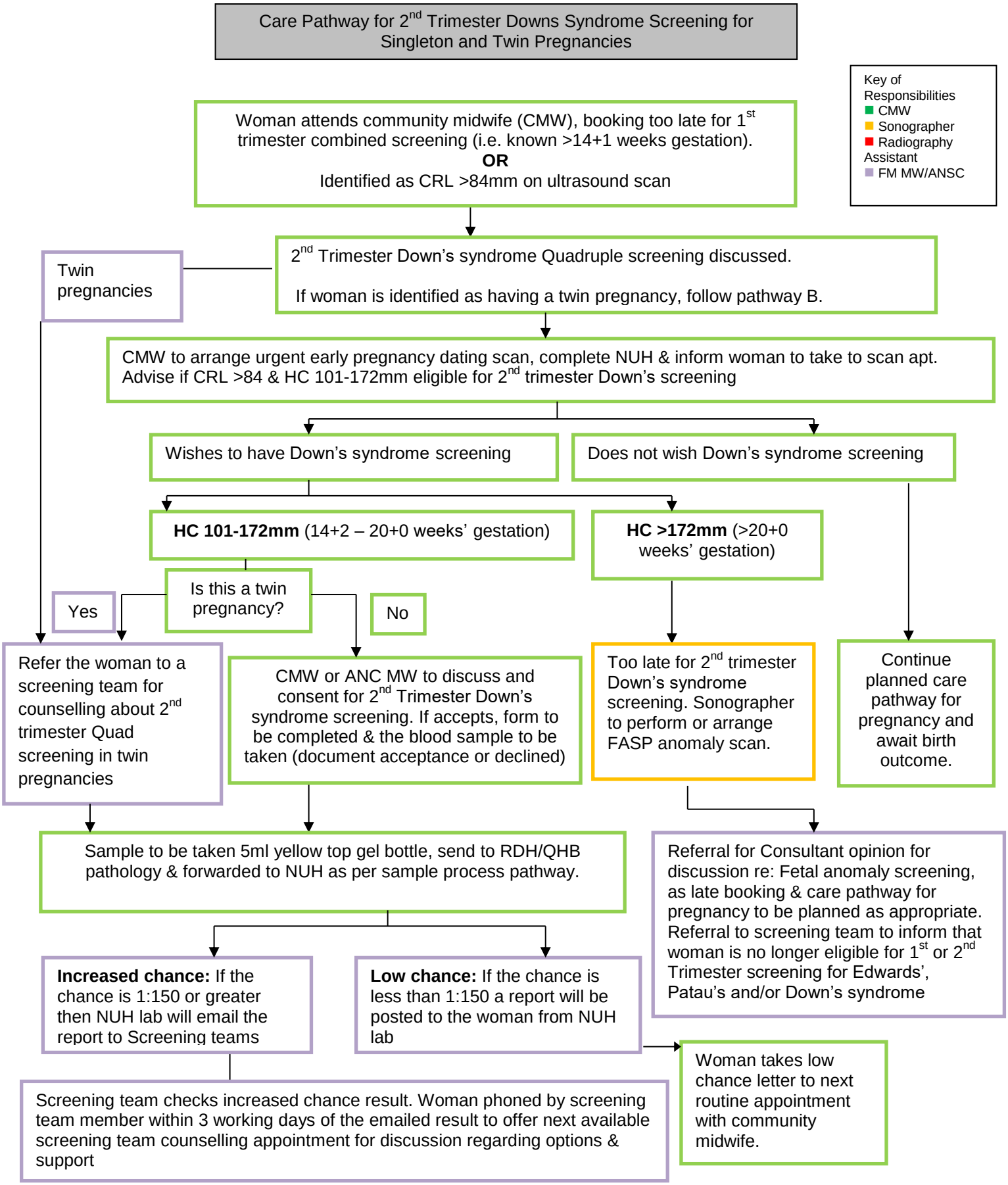
Screening team checks high-chance result report. Woman phoned by screening team within 3 working days of receipt of email high chance result, to offer next available specialist midwife counselling appointment for discussion regarding options & support.

Woman takes low chance letter to next routine appointment with community midwife.

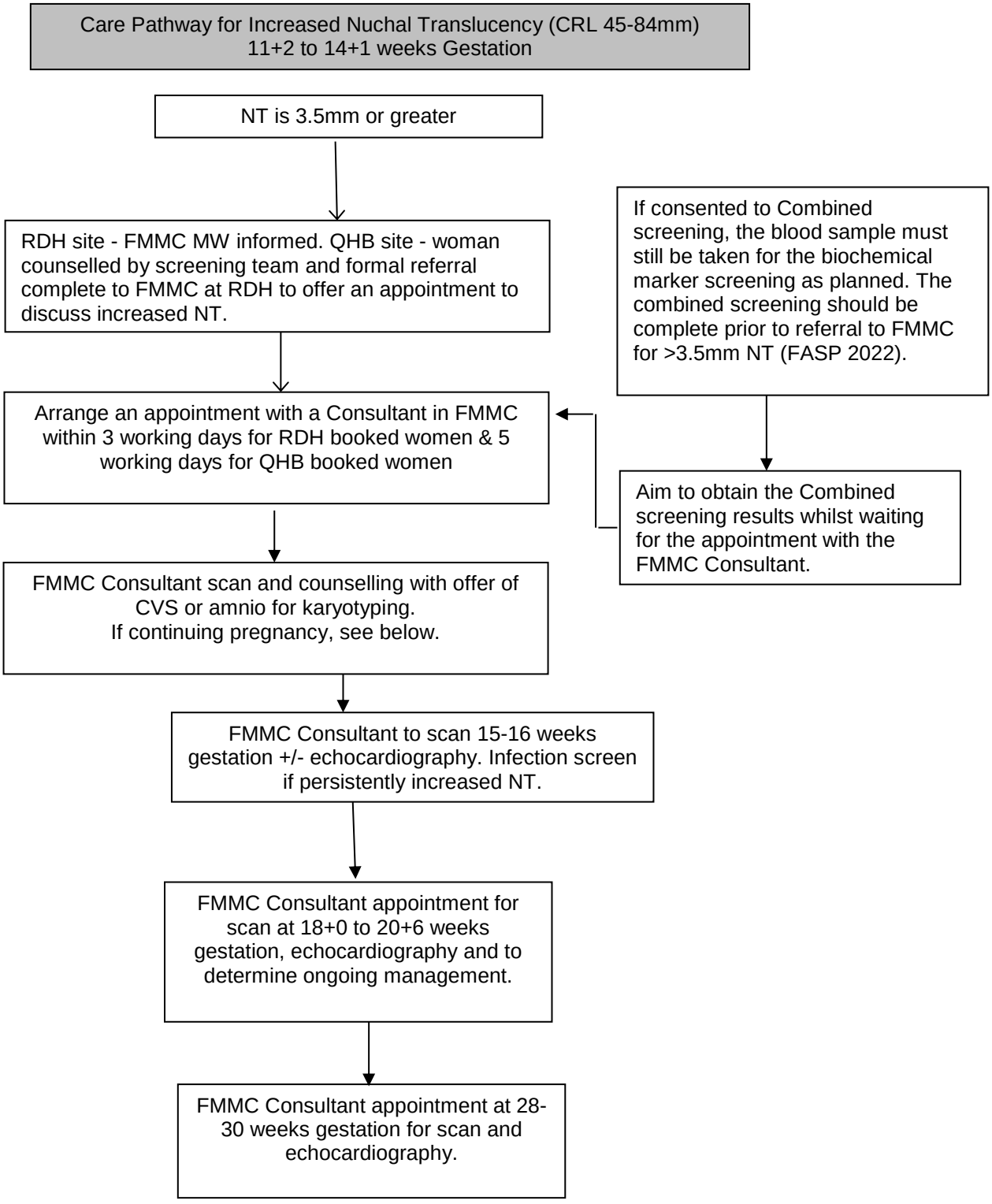
Pathway B


Care Pathway for Down's syndrome, Edwards' syndrome and Patau's Syndrome Screening in a **Twin Pregnancy**
1st Trimester Combined Screening for women with a twin pregnancy
Key of Responsibilities
CMW
Sonographer
Radiography Assistant
FM MW/ANSC
Twin pregnancy identified in advance of dating scan
Twin pregnancy diagnosed at time of dating scan/NT scan
Woman wishes to consider screening for
a) Down's
b) Edwards & Patau's
c) Down's, Edwards' & Patau's
Woman does not wish to consider screening for Down's, Edwards' or Patau's Syndrome.
Complete dating scan/NT scan. If Down's and /or Edwards' and Patau's screening wished, refer to screening team to discuss DCDA combined screening choices in multiple pregnancies and change maternity tariff. If MCMA/MCDA refer to FMMC to discuss
CMW to book NT scan & consultant booking for >11+2. Sonographer to refer to screening team after scan to discuss Combined screening if DCDA and refer to FMMC if MCMA/MCDA
CMW to arrange dating scan from 11 weeks gestation and Consultant booking
ANC Midwife to arrange booking, dependent on chorionicity.
Woman's choice to proceed with 1st trimester Combined screening
Woman to proceed with combined screening
Does not wish to have 1st trimester Combined screening after discussion.
Sonographer to perform dating scan & NT measurements if CRLs 45-84mm. For 'vanished' twin refer to FASP & local guidance
Would wish Combined screening for twins, but not eligible as gestation too late (CRLs >84mm). Sonographer to refer the woman to a screening MW for counselling about 2nd trimester Quadruple screening (if HC 101-172mm)
Arrange a consultant booking appointment as appropriate for chorionicity.
Increased chance
If the chance is 1:150 or greater on either or both twins, the lab will email the screening team the result report
Woman phoned by screening team member within 3 working days to offer next available specialist midwife counselling appointment for discussion, options & support.
Low chance
If the chance is less than 1:150 for both twins, a letter will be sent directly to the woman form NUH lab
Woman takes low chance letter to next routine appointment with community midwife.
Following the dating/NT scan, ANC midwife to arrange a consultant booking appointment, dependent on chorionicity.

Pathway C


Care Pathway for 2nd Trimester Downs Syndrome Screening for Singleton and Twin Pregnancies
Key of Responsibilities
CMW
Sonographer
Radiography Assistant
FM MW/ANSC
Woman attends community midwife (CMW), booking too late for 1st trimester combined screening (i.e. known >14+1 weeks gestation).
OR
Identified as CRL >84mm on ultrasound scan
Twin pregnancies
2nd Trimester Down's syndrome Quadruple screening discussed.
If woman is identified as having a twin pregnancy, follow pathway B.
CMW to arrange urgent early pregnancy dating scan, complete NUH & inform woman to take to scan apt. Advise if CRL >84 & HC 101-172mm eligible for 2nd trimester Down's screening
Wishes to have Down's syndrome screening
Does not wish Down's syndrome screening
HC 101-172mm (14+2 – 20+0 weeks' gestation)
HC >172mm (>20+0 weeks' gestation)
Is this a twin pregnancy?
Yes
No
Refer the woman to a screening team for counselling about 2nd trimester Quad screening in twin pregnancies
CMW or ANC MW to discuss and consent for 2nd Trimester Down's syndrome screening. If accepts, form to be completed & the blood sample to be taken (document acceptance or declined)
Too late for 2nd trimester Down's syndrome screening. Sonographer to perform or arrange FASP anomaly scan.
Continue planned care pathway for pregnancy and await birth outcome.
Sample to be taken 5ml yellow top gel bottle, send to RDH/QHB pathology & forwarded to NUH as per sample process pathway.
Referral for Consultant opinion for discussion re: Fetal anomaly screening, as late booking & care pathway for pregnancy to be planned as appropriate. Referral to screening team to inform that woman is no longer eligible for 1st or 2nd Trimester screening for Edwards', Patau's and/or Down's syndrome
Increased chance: If the chance is 1:150 or greater then NUH lab will email the report to Screening teams
Low chance: If the chance is less than 1:150 a report will be posted to the woman from NUH lab
Woman takes low chance letter to next routine appointment with community midwife.
Screening team checks increased chance result. Woman phoned by screening team member within 3 working days of the emailed result to offer next available screening team counselling appointment for discussion regarding options & support

Pathway D


Care Pathway for Increased Nuchal Translucency (CRL 45-84mm)
11+2 to 14+1 weeks Gestation
NT is 3.5mm or greater
RDH site - FMMC MW informed. QHB site - woman counselled by screening team and formal referral complete to FMMC at RDH to offer an appointment to discuss increased NT.
If consented to Combined screening, the blood sample must still be taken for the biochemical marker screening as planned. The combined screening should be complete prior to referral to FMMC for >3.5mm NT (FASP 2022).
Arrange an appointment with a Consultant in FMMC within 3 working days for RDH booked women & 5 working days for QHB booked women
Aim to obtain the Combined screening results whilst waiting for the appointment with the FMMC Consultant.
FMMC Consultant scan and counselling with offer of CVS or amnio for karyotyping.
If continuing pregnancy, see below.
FMMC Consultant to scan 15-16 weeks gestation +/- echocardiography. Infection screen if persistently increased NT.
FMMC Consultant appointment for scan at 18+0 to 20+6 weeks gestation, echocardiography and to determine ongoing management.
FMMC Consultant appointment at 28-30 weeks gestation for scan and echocardiography.

Pathway E | **Pathway for Trisomy screening blood samples**

Actions by Radiography Assistant / Imaging Department Assistant (IDA) –

In the phlebotomy room (Antenatal Services), Radiography Assistant / IDA to:

1. Review the dating / NT scan information on request form to ensure gestation eligibility for first trimester screening (CRL 45-84mm & NT measured / documented).
2. If too late for 1st trimester Combined screening i.e. CRL >84mm & HC 101-172mm, check woman has consented to 2nd trimester Quadruple screening at booking via documentation in MHHR on the screening page or that she has been consented by the screening team. *Quad T21 only* box to be ticked on request form.
3. Check correct screening test required box ticked on request form.
4. Confirm the correct patient demographic details & check address is correct to receive results letter.
5. Weigh the pregnant woman in Kg & record on the NUH blood request form.
6. Obtain the 5ml screening blood sample in yellow gel top bottle, after gaining verbal consent.
7. Enter date & time of sample on blood form, print name & sign request form.
8. Complete patient information on obtained blood sample bottles.
9. Check there are a minimum of 3 matching identifiers of form & sample (i.e. full name, DOB & NHS no plus hospital number)
10. Check the ultrasound scan information is complete & correct on the request form.
11. Blood samples and screening request forms should be second checked in department prior to being sent to the lab.
12. Email daily scan work list to NUH and screening team.

RDH site:
Blood samples to be stored in ANC refrigerator.

As soon as the morning scan list is completed:

- Samples are sent to RDH Pathology in the POD chute around 13:00
- Samples are spun & sent by the RDH lab via a courier to NUH lab with the previous day's afternoon samples at approximately 14:00.

As soon as the afternoon list is completed:

- Samples are sent via POD chute at end of the day's scan list
- Samples are spun and stored in RDH Pathology fridge overnight
- Samples sent on next day's courier with the morning samples.

Samples taken on Friday afternoon are spun & sent next day by taxi.

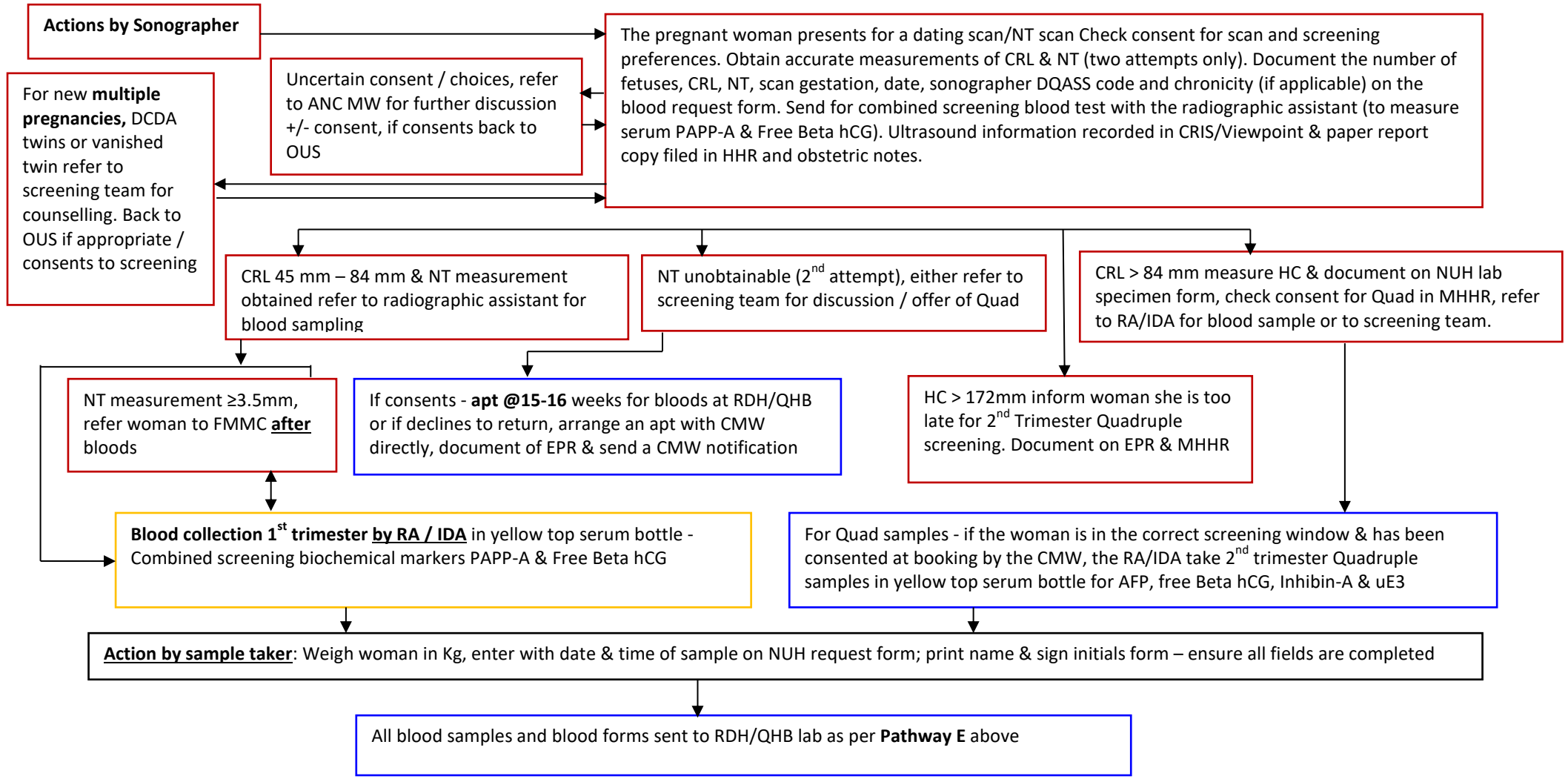
Afternoon samples prior to a Bank Holiday or on Bank Holidays are processed slightly differently depending on the anticipated time-delay and maybe frozen. For further info d/w screening team.

QHB site:
Blood samples are kept in ANC Pathology transport box.
At the end of the day's scan list:

- Samples are taken by the IDAs to QHB Pathology Lab.
- Samples are spun & sent the following morning to NUH lab via a courier
- Samples taken on Friday are spun & refrigerated, stored at QHB Pathology lab & sent to NUH lab on Monday morning via courier
- Samples prior to a Bank Holiday or on Bank Holidays are processed slightly differently depending on the anticipated time-delay. For further info d/w screening team

NUH will contact the Screening teams directly in the case of queries, missing information or samples.

Pathway F – 1st & 2nd Trimester Trisomy screening samples pathway for bloods taken in ANC / Obstetric USS



Appendix A

Patient Information

NHS
University Hospitals of Derby and Burton
NHS Foundation Trust

Results for your NT+ screening test
Low PAPP-A
(Pregnancy Associated Plasma Protein-A)

The result of your combined screening test is now available and you will have had a letter or telephone call with your result.

As part of the first trimester screening test, the blood sample that you had taken was analysed for the pregnancy hormone HCG (Human Chorionic Gonadotrophin) and a pregnancy protein called PAPP-A (Pregnancy Associated Plasma Protein-A). These levels along with the measurement of the fluid at the back of your baby's neck, your age and your weight, have helped to calculate your screening result.

The pregnancy protein that we look for (PAPP-A) is at a slightly lower level than average in your blood.

This is **not** likely to cause you any problems. However a low level of PAPP-A **may** sometimes be linked with a problem with baby's growth rate later in pregnancy.

We recommend that the best course of action is to offer women who have lower levels of PAPP-A, ultrasound growth scans in the third trimester of pregnancy. You will be under the care of a consultant who will arrange these scans which will enable us to assess your baby's growth.

This is not something that you should worry about as it is most likely that when you come for these scans your baby will be growing as it should.

In a small percentage of women a problem with baby's growth rate is found. If this happens, you will be referred to a Fetal Medicine Specialist who will decide how often you need follow up scans.

There is nothing you can do to prevent this or improve your PAPP-A levels.

It is important that you continue to see your community midwife regularly. If she has any concerns about your pregnancy before your appointment, she will refer you to the hospital for an opinion sooner.

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Patient Publications Service

Documentation Control

Reference Number: OBS/07:23/D4	**Version:** **UHDB 1**		**Status: Final**	
Version / Amendment	1	Dec 2010	Tracy Doucas Specialist Midwife Antenatal & Newborn Screening Coordinator Carole Adcock & Sue Rucklidge FM Sp. Midwives	New. To reflect National Screening Committee Standards
	2	Oct 2014	Carole Adcock - FM Specialist Midwife Liz MacGregor – Antenatal Screening Specialist	Review
	3	Aug 2015	Charlotte Daniels Specialist Midwife AN screening Sue Brealey Sonographer	Updated in line with additional screening requirements for T13/T18 & T21 (Public Health England)
	4	March 2017	Cindy Meijer – Risk, Guidelines& Audit Midwife	To add Low PAPP-A pathway
	5	Nov 2017	Tracy Doucas – Antenatal Screening Coordinator	To bring in line with QA post visit recommendations & new PHE national terminology requirements
WC/OG/77	**Burton Trust prior to merged document:**			
	5	Oct 2017	Obstetric Lead for Screening A/N Screening Co-ordinator	Pathway extended to include Patau's and Edwards Syndrome
Version control for UHDB merged document:				
	1	Aug 2022	Jo Wallace – Deputy HOM Rachel McLean - Antenatal and Newborn Screening Lead Midwife Tracy Doucas – Antenatal Screening Coordinator	Review / merge
Intended Recipients: All staff with responsibility for screening women antenatally				
Dissemination: Cascaded electronically through lead sisters/midwives/doctors; Published on Intranet, Article in Business unit newsletter; emailed via NHS.net				
To be read in conjunction with:				
Consultation with:				
Business unit sign off:	02/05/2023: Maternity Guidelines Group: Miss S Rajendran – Chair 19/06/2023: Maternity Governance Group - Mr R Deveraj			
Notification Overview sent to TIER 3 Divisional Quality Governance Operations & Performance: 20/06/2023				
Implementation date:	10/07/2023			
Review Date:	July 2026			
Key Contact:	Joanna Harrison-Engwell			