Down's, Patau's and Edwards' Syndrome (Trisomies) Screening Guideline

University Hospitals of Leicester NHS
NHS Trust

Trust ref: C6/2001

Contents

1. Introduction and Who Guideline applies to

This guideline is intended for the use of all Medical, Midwifery, Nursing, Sonographers, General Practitioners and Laboratory staff involved in the care of pregnant women/birthing people in both Primary and Secondary care settings.

It is to inform pregnant women/people about the chance of Down's (trisomy 21) and Patau's (trisomy 13) and Edwards' (trisomy 18) syndrome in their current pregnancy as estimated by the screening tests and to ensure women/people understand the implications and consequences of opting into screening and the further assessment and management they may be offered.

NB: Down's (trisomy 21), Patau's (trisomy 13) and Edwards' (trisomy 18) syndrome will collectively be known as Trisomies 21,13 and 18 throughout this guideline apart from where quad testing is carried out as this is to determine a chance of Downs Syndrome alone.

The way the programme is delivered cannot be defined rigidly because it has to be incorporated into the individual care of each pregnant woman/person. Therefore, everyone who is involved in antenatal care is responsible for delivery of the screening programme.

2. Guideline Standards and Procedures

This guideline is based upon recommendations from the Fetal Anomaly screening programme: programme handbook (updated 2023) NICE Antenatal Care Guidelines (2019)

All eligible pregnant women/people who book for their maternity care with University Hospitals of Leicester (UHL) will be offered screening for Trisomies as recommended by the National Screening Committee. The gestation of the pregnancy at the time of screening will dictate the screening test available to the pregnant woman/person.

There is a designated lead for antenatal screening for the UHL Maternity Service (Midwifery Matron for Antenatal Services and Lead for Safeguarding). There is also an Antenatal and Newborn Screening Co-ordinator whose role it is to ensure appropriate processes are in place to offer pregnant women/people appropriate screening tests in pregnancy as per National Screening Committee Guidance. In addition, the screening team also includes a Deputy Screening Coordinator, administration support and a failsafe officer.

Some centres now also offer non-invasive prenatal testing (NIPT) to pregnant women/people. If pregnant women/people are interested in NIPT then they should be advised to seek private services at one of the centres that can provide this service, including the private service at UHL.

2.1 First Trimester Combined Screening

The first trimester combined screening test uses two substances produced by the pregnancy which are circulated in the woman's/person's blood. These substances, free beta HCG and PAPP A are measured. In addition, the nuchal translucency of the baby is measured by ultrasound. These results are mathematically combined to estimate, more accurately, the chance of Trisomy 21/13/18 for that pregnancy. Pregnant women/people will have the options for screening for these trisomies as follows:

1. Decline screening for all these trisomies
2. Accept screening for Trisomy 21 only
3. Accept screening for Trisomy 13/18 (as a single result for both)
4. Accept screening for Trisomy T13/18 & 21

The first trimester combined screening test calculation requires the woman's/persons age, weight, smoking status, ethnicity, specific IVF information (egg collection and implantation date & donor egg age if applicable), chorionicity in the case of twins and details of relevant medical conditions. This information is mandatory if a result is to be given to the pregnant woman/person. In addition, accurate and detailed information from the dating scan and NT measurements are required.

For further guidance on the scan element of this screening test please refer to Ultrasound UHL Obstetric Guideline.pdf

The result of the screening is given as a probability (chance) figure, for example, 1 in 1,000 would indicate a LOW chance of Trisomy 21/13/18 and 1 in 10 would indicate a HIGH chance. A cut-off of greater than or equal to 1 in 150 is used to indicate that the pregnancy has a LOW or a HIGH chance of being affected by one of the Trisomies.
In a twin pregnancy (monochorionic and dichorionic) there is a risk generated for each fetus for combined screening.

It is important to recognise that combined screening test is a ***screening*** test and will not detect all pregnancies affected by Trisomy 21/13/18.
The detection rate for the Trisomies varies slightly; the combined screening test will detect approximately 85% of babies affected with Trisomy 21. This means that despite a low chance result the baby still may be affected with Trisomy 21. The detection rate for Trisomy 13/18 is slightly lower at 80%.

2.2 Second Trimester Downs Syndrome Serum Screening

The second trimester serum screening test (Quadruple test) is used to calculate the chance of having baby affected with Down's syndrome (Trisomy 21) only. It measures the levels of Alpha Fetoprotein (AFP), Human Chorionic Gonadotrophin (HCG) unconjugated Oestriol and Inhibin A in the mother's blood. The Quadruple Test calculation also requires the pregnant woman's/persons age, weight, smoking status, ethnicity, specific IVF information (egg harvest collection and implantation date, and donor egg age if applicable), chorionicity in the case of twins, relevant medical conditions and accurate information from the dating scan in this pregnancy. This information is mandatory if a result is to be given to the pregnant woman/person.

The result of the serum screening is given as a probability (chance) figure, for example, 1 in 1,000 would indicate a LOW chance of Trisomy 21 and 1 in 10 would indicate a HIGH chance. A cut-off of greater than or equal to 1 in 150 is used to indicate that the pregnancy has a LOW or a HIGH chance of the fetus being affected by Trisomy 21.
In a twin pregnancy (both monochorionic and dichorionic) the result is generated for the `pregnancy` not the individual fetus by the Quadruple test.

It is important to recognise that the Quadruple test is a ***screening*** test and will not detect all pregnancies affected by Trisomy 21, indeed this test has a detection rate of approximately 80%.
Therefore, as for the first trimester screening test, a low chance result does not mean that the baby is not affected with Trisomy 21.

2.3 Screening

- All eligible pregnant women/people booking with University Hospitals of Leicester NHS Trust should be offered screening for Trisomy 21/13/18 (Down's, Patau's and Edwards' syndrome)

- At the first contact with the Community Midwife (booking appointment) the pregnant woman/person should be given information about Trisomy screening so that they understand the tests that are available to them. There is also information from the NSC "Screening Test for you and Your Baby" available that women/people should be directed to via the leaflet or electronically about Trisomy Screening. This is to ensure they understand what the screening is and the implications of this screening test to enable them to make an informed choice.

- The offer, discussion and the pregnant woman's/persons decision should be documented in the Maternity health records.

- The pregnant woman's/persons choice about Trisomy screening must be documented on E3 at booking to allow the clinic co-ordinators to arrange the NT scan for the woman/person if they want screening. There will be a print off of the relevant information at the booking hospital that will be given to the woman/person when they attend for their NT/dating scan.

- The community midwife should complete all of the white section of the Trisomy screening request form (see Appendix 7) accurately as missing or incorrect information could lead to false results or missed screening.

- **Women or people who have history of having a baby affected with full Trisomy 13/18/21 should be referred to Fetal medicine for a discussion about their options for screening and diagnostic tests including the offer of R445 from 1.4.2024 – see Appendix 12.**

- All electronic handheld maternity records should be reviewed by the Antenatal Services Midwives. If a pregnant woman /person has booked late for screening, an urgent referral should be made by the Antenatal Services Midwives, to the Clinic Coordinators to identify a scan slot that will ensure the woman/person receives their screening test within the required timeframe (see Appendix 3).

- A weekly Trisomy screening failsafe list is automatically generated from the maternity IT system and the pregnant woman's/persons Trisomy screening choices are logged. Women/people who request a screening test will be followed up by the failsafe officer 5 weeks after booking to ensure that screening is completed or declined. See Appendix 8 for the Standard Operating Procedure for the Trisomy screening failsafe process.

- If the dating scan shows the pregnant woman/person to be eligible for nuchal translucency screening, it should be offered & completed at the time of the dating scan (see Appendix 1).
    - Pregnant women/people who are 14+2 to 20+0 week's gestation (HC 101-172mm) at the dating scan should be referred to the Antenatal Services Midwives who should offer Quadruple testing for Trisomy 21 on the same day see (Appendix 1).

- If the nuchal translucency cannot be measured after 2 attempts this should be explained to the woman/person and a plan made for Quadruple testing with the community midwife between 14+2 & 20+0 weeks. The information sheet (see Appendix 6) should be stapled to the front of the handheld notes as a reminder to the woman/person and the community midwife that quadruple testing still needs to be completed.
- The Sonographer should complete the orange Gestational details on the Trisomy screening request form (see Appendix 7).

- If at the dating scan a twin pregnancy is diagnosed the pregnant woman/person is still eligible for 1st trimester combined screening for Down's syndrome. If greater than 14+1 week's gestation they should be offered quadruple testing after a clear explanation about the implications of the result due to the reduced detection rate for Down's syndrome in twins.

- If there is a twin pregnancy with demise of one of the twins and only a yolk sac present, the midwives should continue with first trimester combined screening test. If there is a measurable fetal pole for the demised twin, Nuchal Translucency only screening can be offered following discussion with the Antenatal Services Midwives (see Appendix 4). However, this is not a nationally recommended test and so the pregnant woman/person should be offered a Quad test.

- If the pregnant woman/person books for their Antenatal Care after 20 weeks gestation, the Community Midwife should discuss the woman's/persons screening test result based on an age related chance of Trisomy 21alone and refer the pregnant woman/person to the Antenatal Services Midwives should they wish to discuss their options in relation to private Non-invasive prenatal testing or prenatal diagnosis. Refer to a Fetal Medicine Consultant if diagnostic testing is required.

- First trimester screening
  - Undertaken between 11+2-14+1 weeks gestation (CRL 45-84mm)
  - A follow up appointment should be made to discuss and document the result and also to initiate second trimester screening should a result not be available.
  - Process for the screening test appointment - see Appendix 1.
  - Where an NT measurement is found to be greater than or equal to 3.5mm at the dating scan, the screening test should be completed by taking the serum element of the trisomy screening test and sending the sample along with the scan details to the Lab. The pregnant woman/person should then be referred to a Fetal Medicine Consultant.
- Second trimester screening (Quadruple)
  - Undertaken between 14+2 & 20+0 weeks of pregnancy (HC 101-172mm)
  - A follow up appointment should be made to discuss and document the result and also to initiate repeat screening should a result not be available.
  - Process for the screening test appointment - see Appendix 2.

- NB: An NHS number and full address including postcode is a mandatory requirement on the blood request form, so that a low chance letter can be generated and sent to the pregnant woman/person.

- The Community Midwife should remain the point of contact should the pregnant woman/person have any further queries or concerns.

- On receipt of the screening sample at the Trisomy screening laboratory the information supplied on the form should be checked according to national guidance. Incomplete forms and requests for further information should be emailed by the Lab to the Antenatal Screening nhs.net email at UHL. The information is sought by the screening team and emailed back to the Lab by nhs.net email.

Interpreting services should be used where appropriate. Screening tests for you and your baby information should be also downloadable in different languages from www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief

If extra scan lists are created "out of hours" to increase scan capacity, please follow SOP in appendix 13.

2.4 Low chance results

- Pregnant women/people who have a low chance result should be sent a letter from the laboratory informing them of this within 2 weeks of the result being available

- Low chance results should be sent from the laboratory to the requestor. This is usually in an electronic format

- The midwife at the next antenatal appointment should ensure they have received this and document the result in the maternity health record. The midwife will also ensure that the pregnant woman/person understands that this is a screening result and therefore cannot completely exclude the possibility of their baby being affected with this condition.

- If the pregnant woman/person requires further or more detailed discussion about their low chance result they should be referred to the Antenatal Services Midwives or the Screening Coordinators

2.5 High chance results

High chance results should be emailed by the Trisomy screening laboratory to the Antenatal Services Midwives generic AAAsharedmailbox@uhl-tr.nhs.uk or PASmailbox@uhl-tr.nhs.uk email at the hospital where the woman/person is booked and a copy of the report is sent to the requestor.

- High chance results will be emailed to the antenatal services midwives generic email address, as one result per email. This email must be checked by the midwives in antenatal services at several points throughout the day in order to prevent delay in informing the woman/person of their results. The email will also go to the screening team to check receipt by the fetal medicine team Email; annbscreening@uhl-tr.nhs.uk

- The midwife managing the result must respond to the email from the lab by return, to acknowledge that this result is being actioned.

- The Antenatal Services Midwives should document the result on the Fetal Diagnostic Care Plan

- The result should be confirmed using iLAB or ICE.
- The gestational age at date of sample should be verified as correct
- Hospital notes should be obtained
- An appropriate appointment time for the pregnant woman/person to attend the hospital to discuss their result with the midwives and potential diagnostic test should be identified within 3 working days of the result being received.
- The pregnant woman/person should be informed of their result by telephone, no more than 24 hours prior to the planned appointment time and the appointment offered.
- All discussions with the pregnant woman/person about their Trisomy screening results must be documented on the electronic maternity system.
- If the midwife is unable to contact the pregnant woman/person or the appointment is on a Monday the Community Office should be informed and the Community Midwife should inform the pregnant woman/person of their result and the appointment
- If the Community Midwife is unable to make contact with the pregnant woman/person they should post a note through the door asking that the woman/person contacts the Antenatal Service Midwives as soon as possible. (See Appendix 5)
- The Community Midwife should also let the Antenatal Services Midwives know that this has taken place.
- If the pregnant woman/person fails to contact the Antenatal Services Midwives within a week the above process should be repeated
- A contact number for further enquiries should be given
- The screening team will check that all high chance results have been actioned by the Fetal medicine midwives at least twice per week and record this on the database.

2.6 Equivocal results

- Equivocal results that require further action should be emailed to the Screening Coordinators generic – annbscreening@uhl-tr.nhs.uk
- An individualised management plan should be made by the Antenatal Services Midwives or Screening Coordinator in conjunction with the Consultant

Low Papp A (less than or equal to 0.41MOMs)

- Following a low Papp A result pregnant women/people will need to be informed that this result does not affect the results that they have been given for Trisomy screening.

- Pregnant women/people will need to be informed of the changes required for Fetal surveillance during pregnancy and scan pathway monitoring commenced.

- Arrange to commence Aspirin 150mg as soon as results are available as per Aspirin guideline.

2.7 Results not available follow up

- Results found to be unavailable should be followed up.

- If the health professional identifies that the result is not available at the follow up appointment, they should contact the laboratory to check their records.

- If test has not been performed further testing should be initiated

- A follow up appointment should be made for the pregnant woman/person to receive their result within 2 weeks.

2.8 Antenatal Services Midwives discussion, Implications & further testing

- The Antenatal Services Midwives should offer a full discussion with the pregnant woman/person about the implications of a high chance result and further testing is offered and initiated where accepted

- The Antenatal Services Midwives should discuss the high chance result with the pregnant woman/person and their partner / family, ensuring they have an understanding of the screening test, including its implications and limitations.

- The midwife should inform the pregnant woman/person of their options:

    - Do nothing and accepted their screening result,
    - NHS funded non-invasive prenatal testing (NIPT) Appendix 10&11
    - prenatal diagnostic testing with its associated risk of miscarriage (0.5%).

- NHS funded NIPT samples must be sent to the external laboratory in the dedicated boxes provided and sampling techniques should be followed as per the dedicated laboratory guidance.

- It should be made clear at this point that if there are no abnormalities on scan the diagnostic test result will only be for T13/18/21 and the sex chromosomes (see recommendation 7 for details).

- The pregnant woman/person should be aware that they will be informed of any abnormal chromosome arrangement on diagnostic testing, regardless of consent choices at the time of screening. For example, if the screening result is high chance for Trisomy 13/18 and the baby is affected with Trisomy 21 we will inform them of this result regardless of previous screening choices.

- Sufficient time should be given for decision making.

- If the pregnant woman/person wishes to proceed to a diagnostic test the midwives should ensure an appropriate appointment with a Fetal Medicine Consultant is in place and that the woman/person has a full understanding of the procedure, it's risks and how the results will be communicated

- The midwives should then support the pregnant woman/person through the procedure and plan the process for communicating her results.

- All of the above should be documented fully in the electronic Maternity Health Records

2.9 NIPT Results communication

NIPT results when the test was offered on the NHS following a high chance screening result:

- NIPT results will be emailed to the Antenatal Services Midwives and communicated appropriately to the pregnant woman/person
- Results will come by email from the Lab, high chance results will come in an individual email – more than once daily checking of fetal medicine shared emails is required.
    - Confirm receipt of results by responding to the email.
    - Clinical queries: Telephone – 02073077409 or email elaine.holgado@tdlpathology.com
- No result will be notified by an individual email and should be acknowledged by return email. The lab then should be informed when the repeat sample has been taken and sent (if repeated).
    - If there is no result the pregnant woman/person can be given the choice to have one more NIPT test, prenatal diagnosis or no further testing.
- The laboratory reception will notify the contact on the request form if the sample cannot be processed e.g. insufficient blood, expired tube etc. No report will be generated if the sample is not tested.

NIPT results when the test was offered on the NHS following a previously affected pregnancy:

- Reported same way as other NIPT samples taken for higher chance combined or quadruple screening test.
- Reported as higher chance, lower chance or no result.
- No numerical values are reported.

2.10 Diagnostic test results

- Diagnostic test results should be obtained by the Antenatal Services Midwives and communicated appropriately to the pregnant woman/person.

- Florescent In-situ hybridisation (FISH) test or QF-PCR will be carried out for pregnant women/people who have an increased chance. This will give a result for Trisomy 21/13/18. The Antenatal Services Midwives should ensure that these results are available and reported to the woman/person within the expected timeframe. This will normally be within 3 working days.

- Where pregnant women/people have an increased chance and an NT measurement of >3.5mm and / or a fetus with suspected structural malformation, they will still have the FISH/QF-PCR test **and** a micro-array. Micro-array results should be made available to the pregnant woman/person as soon as it is received. This will usually take between two to three weeks.

- Pregnant women/people who have had a normal FISH/QF-PCR result early in pregnancy but the fetus is noted to have a structural problem at the anomaly scan, can be offered a repeat invasive test for a full assessment of chromosomes & micro-array.

- The result, following rigorous checking of all demographic details to ensure that the result is correct should be communicated to the correct pregnant woman/person and documented in the electronic Maternity Health Records.

- The midwife should attempt to contact the pregnant woman/person by telephone as soon as the result is available – this process should have been agreed with the woman/person at the time of the procedure.

- The midwife should document that they have given the pregnant woman/person their result or that they have been unsuccessful in making contact with them specifying the date and time. Further attempts should then be made until contact has been achieved.

- In the event of an abnormal result an appropriate appointment should already have been arranged for the pregnant woman/person to discuss their result and their options with the Antenatal Services Midwives and a Fetal Medicine Consultant

3. Education and Training:

- Antenatal and newborn (ANNB) screening training is provided as part of induction training for midwives, obstetricians and midwifery care assistants.

- ANNB screening training is provided on annual mandatory training for midwives, MCA's and children's nurses.

4.Monitoring Compliance

What will be measured to monitor compliance	How will compliance be monitored	Monitoring Lead	Frequency	Reporting arrangements
Key performance indicators for ANNB screening Monitoring of Trisomy screening standards	NHS England and PHE QA teams will monitor through the ANNB screening programme boards NHS England and PHE QA teams will monitor through the ANNB screening programme boards		Quarterly Annually	CMG Q&S board

5. Related guidelines:

Ultrasound UHL Obstetric guideline (B52/2011)

Aspirin in Pregnancy UHL Obstetric guidelines (C36/2011)

Fetal surveillance – Small for Gestational Age Fetus Obstetric guideline (C38/2017)

Booking bloods and urine test UHL Obstetric guideline (C15/2011)

6. Supporting References:

1. Fetal Anomaly screening – programme handbook updated 2023.
2. Fetal anomaly screening programme standards updated 2021
3. NICE Antenatal Care Guidelines (2021). https://www.nice.org.uk/guidance/ng201/antenatal-care

7. Key Words

Trisomy screening, Down Syndrome, Patau Syndrome, Edward's Syndrome.

The Trust recognises the diversity of the local community it serves. Our aim therefore is to provide a safe environment free from discrimination and treat all individuals fairly with dignity and appropriately according to their needs.
As part of its development, this policy and its impact on equality have been reviewed and no detriment was identified.

Contact & review details

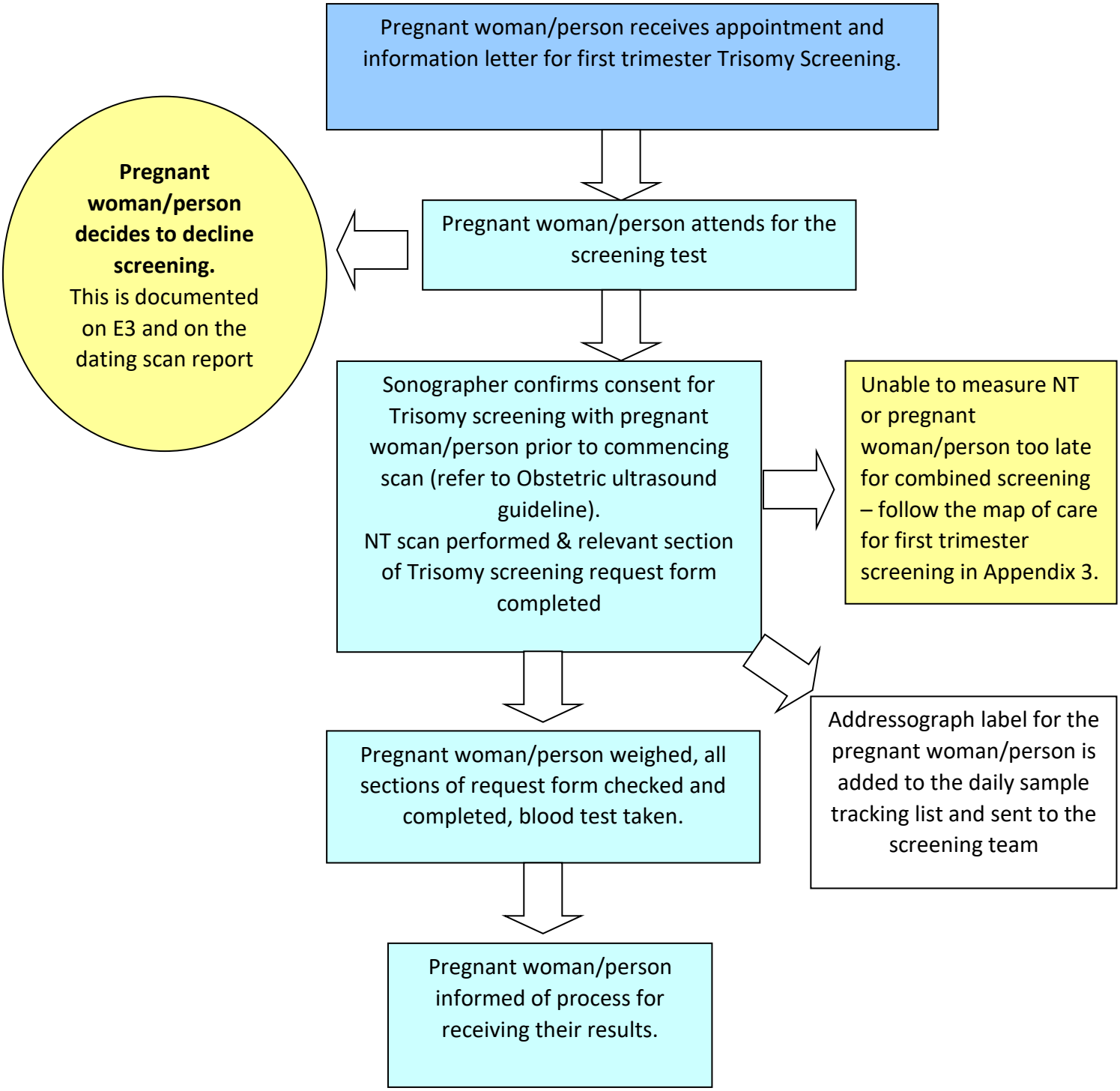
Original Author / Lead Officer:	Louise Payne, Senior Midwife for Community Services Lorraine Matthews, Midwife Quality and Safety Helen Ulyett, Antenatal and Newborn Screening Coordinator	
Reviewed by:	H Ulyett and L.Payne	
Approved by:	**Guidelines Group and Maternity Service Governance Group**	**Date Approved: 16/06/2021**

REVIEW RECORD

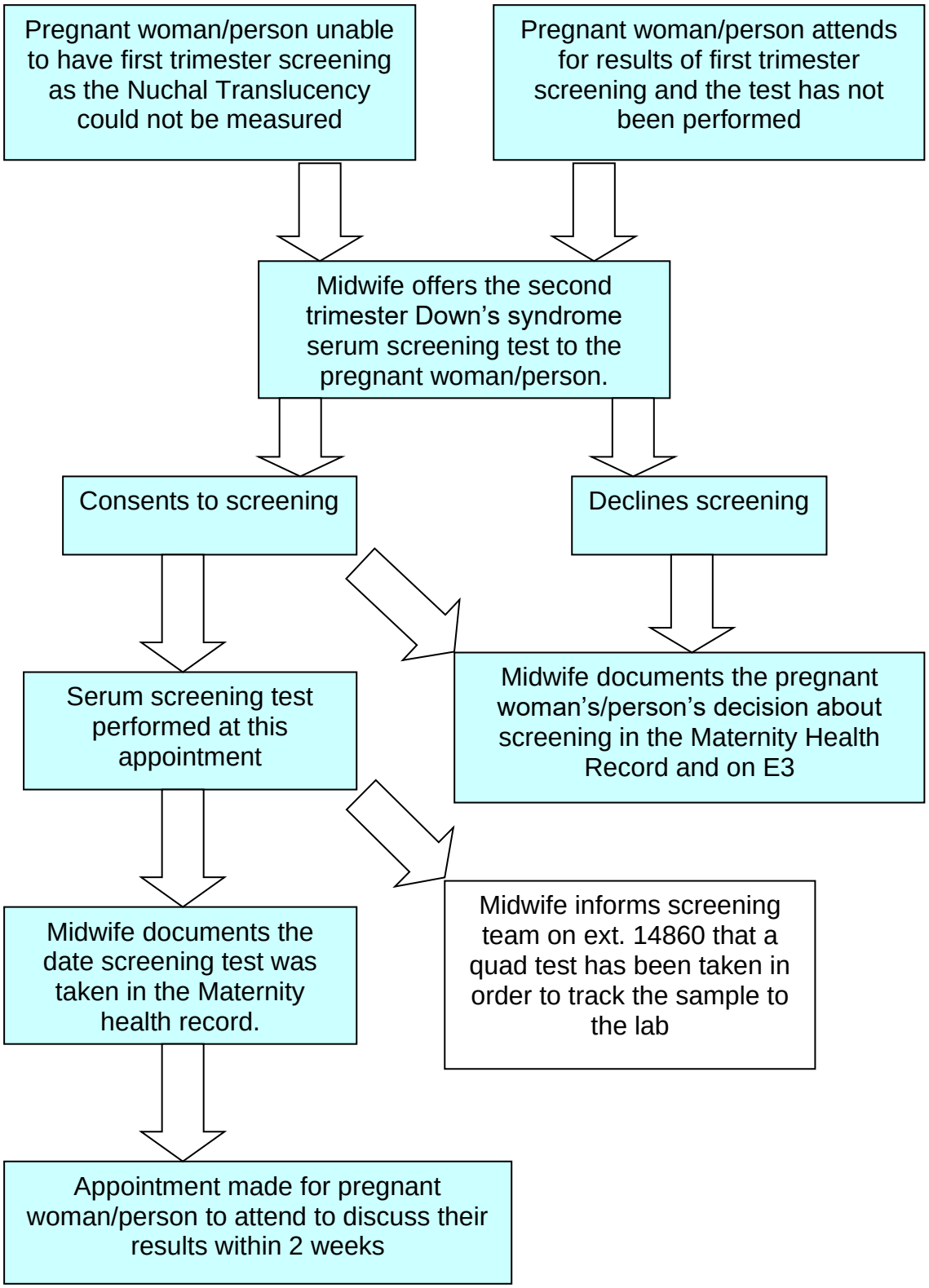
Date	Issue Number	Reviewed By	Description Of Changes (If Any)
June 2014	V2	H Ulyett, L Matthews and the Fetal Medicine Team	NIPE info inserted. Management of twins clarification, extra info on NT and completion of forms
October 2015	V2	As above	Lab changed to Nottingham Quad testing to be performed by the Antenatal Services Midwives when scan shows woman is too late for first trimester
February 2016	V3	As above	Addition of Quad testing for Twins if first trimester combined screening is not possible. Addition of screening for Edward's and Patau's syndrome to Down's screening guideline
October 2016	V4	As above	FISH only now for increased screening test result. Full culture for increased risk combined with NT of >3.5mm and / or a fetus with suspected structural abnormality

July 2018	V5	As above and L. Payne	Change of terminology from Down's Edwards & Patau's screening to Trisomy 13/18 & 21 screening Change of terminology from risk to chance throughout the document. Information for women about NIPT Change from full karyotype to micro-array for diagnostic testing
January 2020	V6	As for V5	Updated use of emails instead of fax for missing information requests and high chance results from the Lab. Terminology changed to Trisomy screening Lab instead of the hospital of Laboratory provider in case it changes again. Removal of NIPT from high chance pathway. Changes to the Trisomy screening failsafe SOP.
June 2021	V7	H. Ulyett, M.Bodley, L.Payne and Fetal medicine team	Addition of NIPT as part of the high chance results pathway on the NHS.
October 2023	V8	L.Payne H.Ulyett	Clearer documentation of failsafe for high chance results. High chance results discussion with woman/birthing person to be recorded on electronic maternity system Non-viable twin can have screening with the quad test QF-PCR for prenatal diagnosis added Reformatted
March 2024	V9	H. Ulyett	Addition of R445 for women with a previous history of a full Trisomy 13/18 or 21.
July 2024	V10	H. Ulyett	Addition of SOP for the management of Trisomy screening bloods when extra scan lists are running out of hours

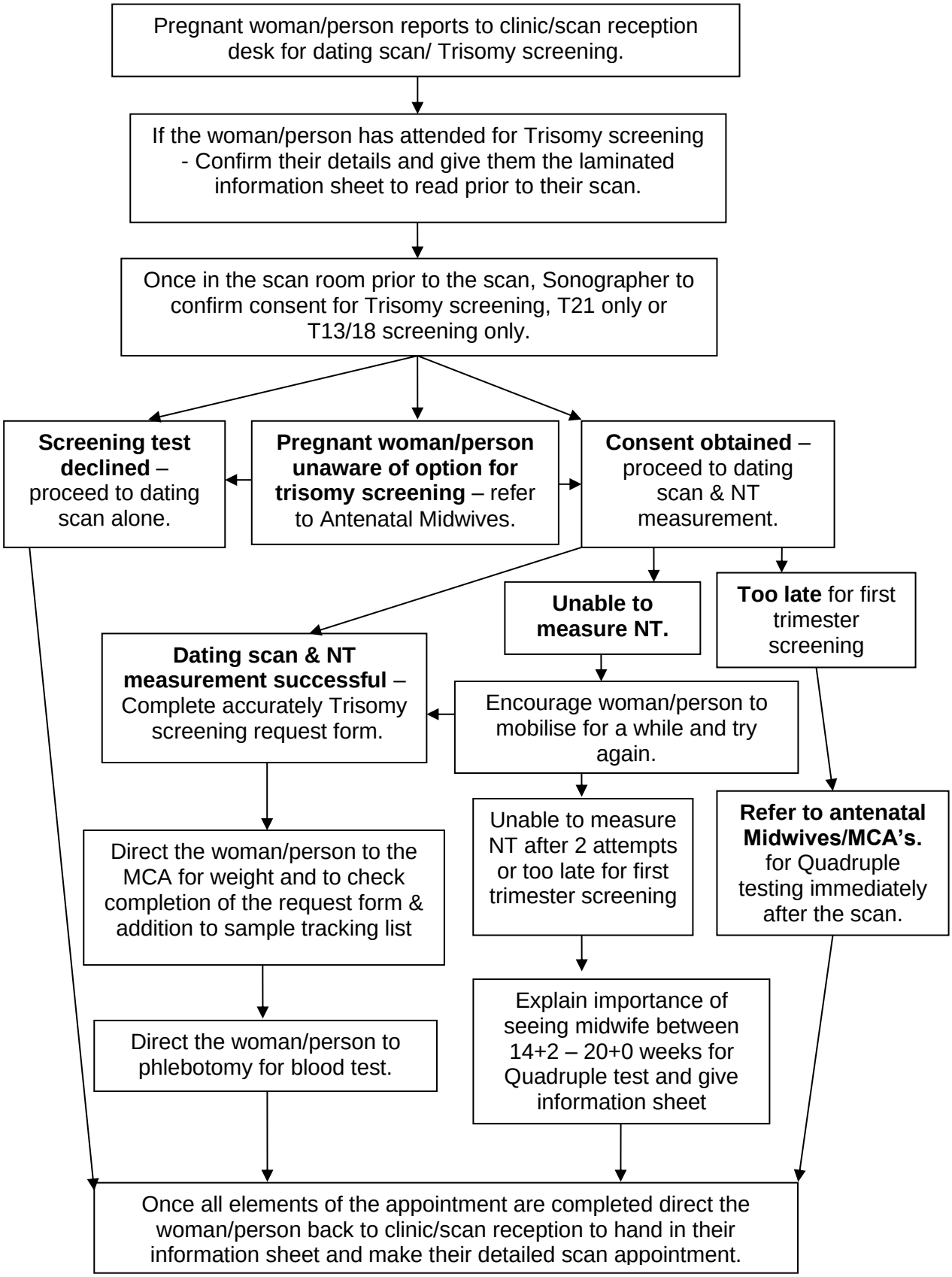
Appendix 1 – Process for the first trimester Trisomy 21/13/18 screening appointment.



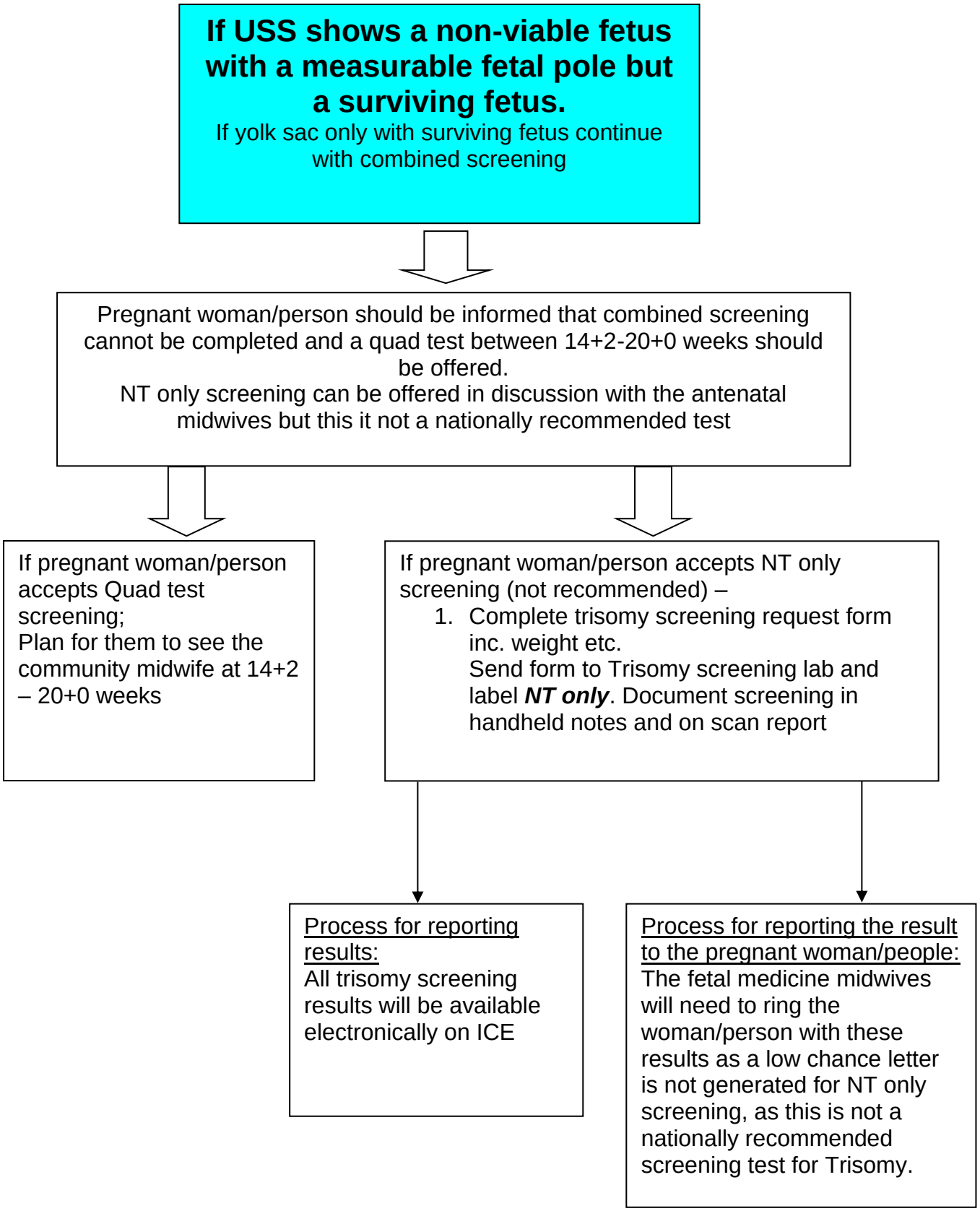
Appendix 2 – Process for the second trimester Down's syndrome screening appointment within the Community setting.



Appendix 3. Map of care for pregnant women/people attending for first trimester Trisomy screening through the Antenatal department.



Appendix 4 - UHL process for Trisomy Screening following diagnosis of a "Demised Twin".



Appendix 5 – Community letter

University Hospitals of Leicester NHS
NHS Trust

Date ……

Dear ……

I have tried to visit you at home today to give you some test results.

Could you please contact the Antenatal Midwives for further information as soon as possible?

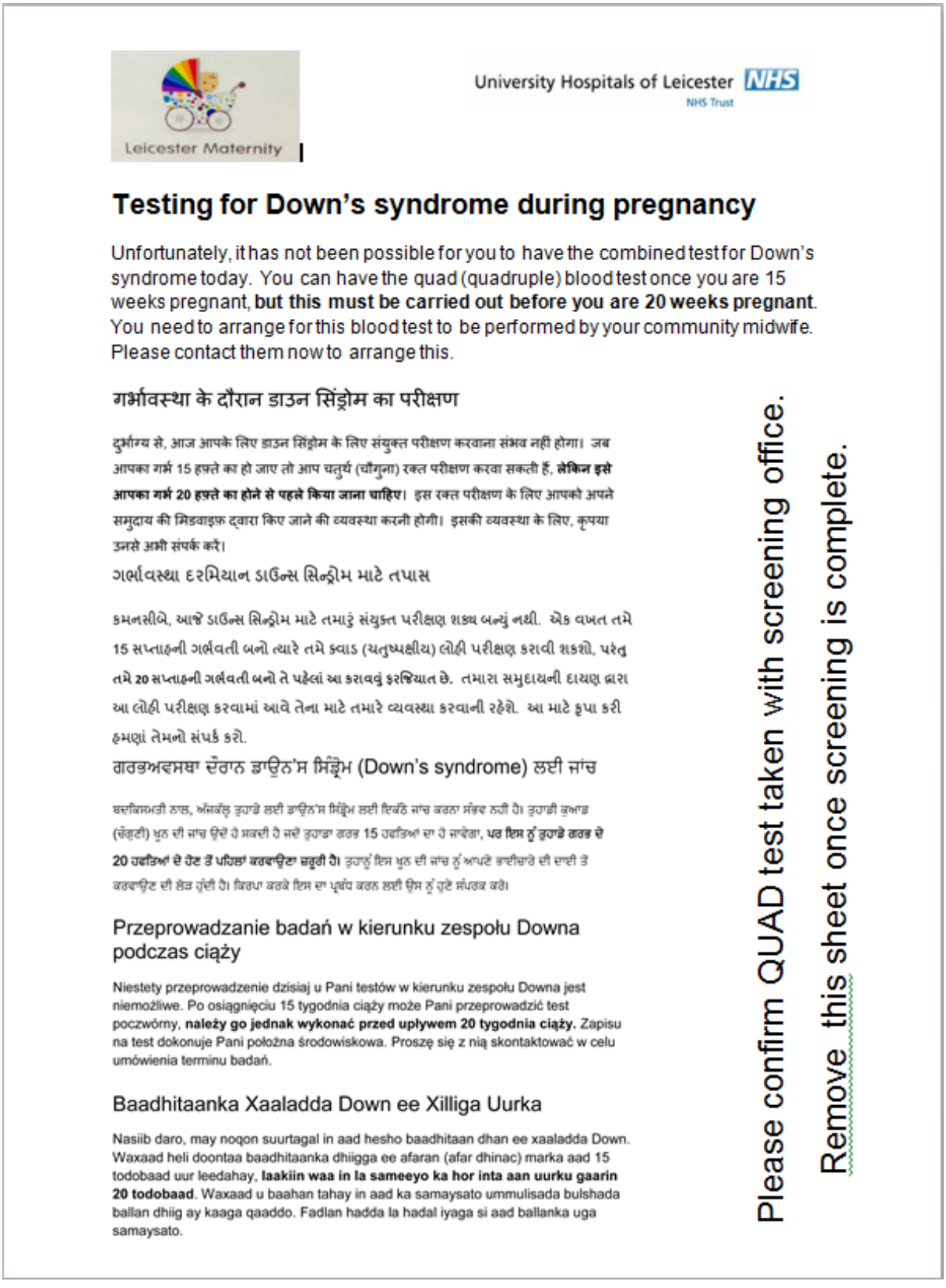
If you are booked to have your baby at the LRI telephone 0116 2586106 (between 8.30am and 4pm)

If you are booked to have your baby at the LGH telephone 0116 2584829 (between 8.30am and 4pm)

Yours sincerely

Community Midwife

Appendix 6 – patient letter for incomplete screen



Leicester Maternity

University Hospitals of Leicester NHS
NHS Trust

Testing for Down's syndrome during pregnancy

Unfortunately, it has not been possible for you to have the combined test for Down's syndrome today. You can have the quad (quadruple) blood test once you are 15 weeks pregnant, **but this must be carried out before you are 20 weeks pregnant**. You need to arrange for this blood test to be performed by your community midwife. Please contact them now to arrange this.

गर्भावस्था के दौरान डाउन सिंड्रोम का परीक्षण

दुर्भाग्य से, आज आपके लिए डाउन सिंड्रोम के लिए संयुक्त परीक्षण करवाना संभव नहीं होगा। जब आपका गर्भ 15 हफ़्ते का हो जाए तो आप चतुर्थ (चौगुना) रक्त परीक्षण करवा सकती हैं, **लेकिन इसे आपका गर्भ 20 हफ़्ते का होने से पहले किया जाना चाहिए**। इस रक्त परीक्षण के लिए आपको अपने समुदाय की मिडवाइफ द्वारा किए जाने की व्यवस्था करनी होगी। इसकी व्यवस्था के लिए, कृपया उनसे अभी संपर्क करें।

ગર્ભાવસ્થા દરમિયાન ડાઉન્સ સિન્ડ્રોમ માટે તપાસ

કમનસીબે, આજે ડાઉન્સ સિન્ડ્રોમ માટે તમારું સંયુક્ત પરીક્ષણ શક્ય બન્યું નથી. એક વખત તમે 15 સપ્તાહની ગર્ભવતી બનો ત્યારે તમે ક્વાડ (ચતુષ્પ્રકીય) લોહી પરીક્ષણ કરાવી શકશો, પરંતુ **તમે 20 સપ્તાહની ગર્ભવતી બનો તે પહેલાં આ કરાવવું ફરજિયાત છે.** તમારા સમુદાયની દાયણ દ્વારા આ લોહી પરીક્ષણ કરવામાં આવે તેના માટે તમારે વ્યવસ્થા કરવાની રહેશે. આ માટે કૃપા કરી હમણાં તેમનો સંપર્ક કરો.

ਗਰਭਅਵਸਥਾ ਦੌਰਾਨ ਡਾਉਨ'ਸ ਸਿੰਡ੍ਰੋਮ (Down's syndrome) ਲਈ ਜਾਂਚ

ਬਦਕਿਸਮਤੀ ਨਾਲ, ਅੱਜਕੱਲ੍ਹ ਤੁਹਾਡੇ ਲਈ ਡਾਉਨ'ਸ ਸਿੰਡ੍ਰੋਮ ਲਈ ਇਕੱਠੇ ਜਾਂਚ ਕਰਨਾ ਸੰਭਵ ਨਹੀਂ ਹੈ। ਤੁਹਾਡੀ ਕੁਆਡ (ਚੌਗੁਣੀ) ਖੂਨ ਦੀ ਜਾਂਚ ਉਦੋਂ ਹੋ ਸਕਦੀ ਹੈ ਜਦੋਂ ਤੁਹਾਡਾ ਗਰਭ 15 ਹਫ਼ਤਿਆਂ ਦਾ ਹੋ ਜਾਵੇਗਾ, **ਪਰ ਇਸ ਨੂੰ ਤੁਹਾਡੇ ਗਰਭ ਦੇ 20 ਹਫ਼ਤਿਆਂ ਦੇ ਹੋਣ ਤੋਂ ਪਹਿਲਾਂ ਕਰਵਾਉਣਾ ਜ਼ਰੂਰੀ ਹੈ।** ਤੁਹਾਨੂੰ ਇਸ ਖੂਨ ਦੀ ਜਾਂਚ ਨੂੰ ਆਪਣੇ ਕਾਈਚਾਰੇ ਦੀ ਦਾਈ ਤੋਂ ਕਰਵਾਉਣ ਦੀ ਲੋੜ ਹੁੰਦੀ ਹੈ। ਕਿਰਪਾ ਕਰਕੇ ਇਸ ਦਾ ਪ੍ਰਬੰਧ ਕਰਨ ਲਈ ਉਸ ਨੂੰ ਹੁਣੇ ਸੰਪਰਕ ਕਰੋ।

Przeprowadzanie badań w kierunku zespołu Downa podczas ciąży

Niestety przeprowadzenie dzisiaj u Pani testów w kierunku zespołu Downa jest niemożliwe. Po osiągnięciu 15 tygodnia ciąży może Pani przeprowadzić test poczwórny, **należy go jednak wykonać przed upływem 20 tygodnia ciąży.** Zapisu na test dokonuje Pani położna środowiskowa. Proszę się z nią skontaktować w celu umówienia terminu badań.

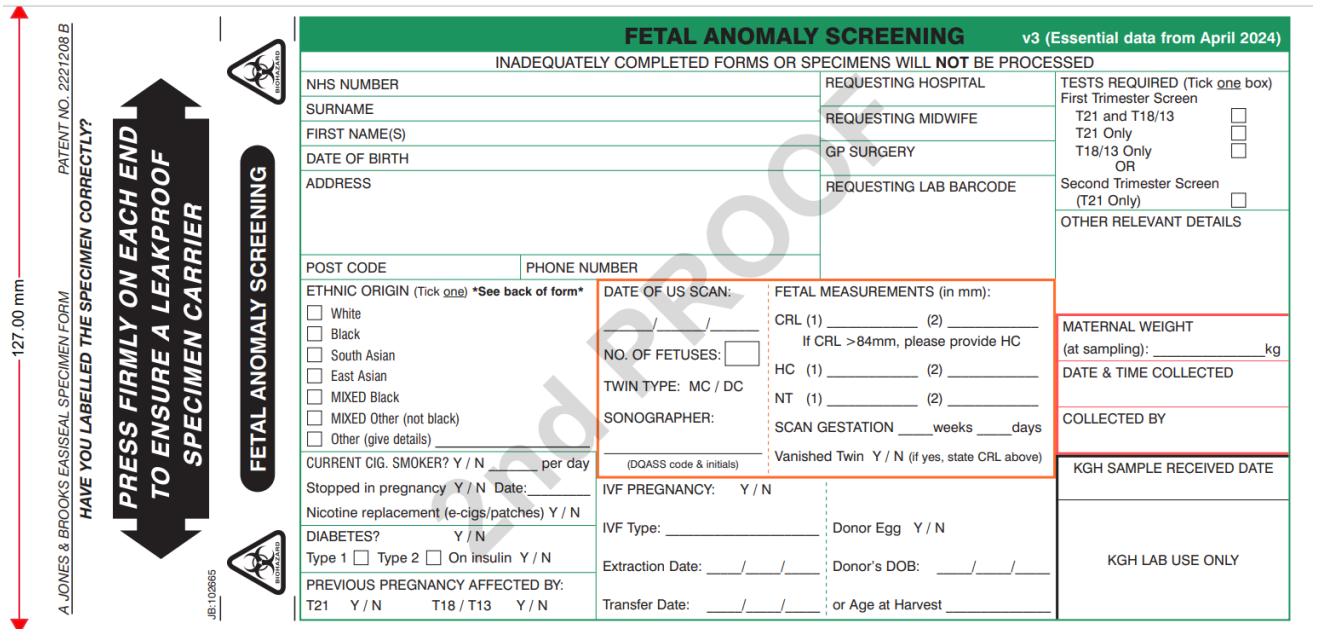
Baadhitaanka Xaaladda Down ee Xilliga Uurka

Nasiib daro, may noqon suurtagal in aad hesho baadhitaan dhan ee xaaladda Down. Waxaad heli doontaa baadhitaanka dhiigga ee afaran (afar dhinac) marka aad 15 todobaad uur leedahay, **laakiin waa in la sameeyo ka hor inta aan uurku gaarin 20 todobaad**. Waxaad u baahan tahay in aad ka samaysato ummulisada bulshada ballan dhiig ay kaaga qaaddo. Fadlan hadda la hadal iyaga si aad ballanka uga samaysato.

Please confirm QUAD test taken with screening office.

Remove this sheet once screening is complete.

Appendix 7 – Kettering Trisomy screening request form.



127.00 mm

A JONES & BROOKS EASISEAL SPECIMEN FORM PATENT NO. 2221208 B

HAVE YOU LABELLED THE SPECIMEN CORRECTLY?

PRESS FIRMLY ON EACH END TO ENSURE A LEAKPROOF SPECIMEN CARRIER

FETAL ANOMALY SCREENING

JB:102665

FETAL ANOMALY SCREENING v3 (Essential data from April 2024)

INADEQUATELY COMPLETED FORMS OR SPECIMENS WILL **NOT** BE PROCESSED

NHS NUMBER

SURNAME

FIRST NAME(S)

DATE OF BIRTH

ADDRESS

POST CODE

PHONE NUMBER

REQUESTING HOSPITAL

REQUESTING MIDWIFE

GP SURGERY

REQUESTING LAB BARCODE

TESTS REQUIRED (Tick one box)
First Trimester Screen
T21 and T18/13 ☐
T21 Only ☐
T18/13 Only ☐
OR
Second Trimester Screen (T21 Only) ☐

OTHER RELEVANT DETAILS

ETHNIC ORIGIN (Tick one) *See back of form*
☐ White
☐ Black
☐ South Asian
☐ East Asian
☐ MIXED Black
☐ MIXED Other (not black)
☐ Other (give details) _____

CURRENT CIG. SMOKER? Y / N _____ per day
Stopped in pregnancy Y / N Date: _____
Nicotine replacement (e-cigs/patches) Y / N

DIABETES? Y / N
Type 1 ☐ Type 2 ☐ On insulin Y / N

PREVIOUS PREGNANCY AFFECTED BY:
T21 Y / N T18 / T13 Y / N

DATE OF US SCAN: ____/____/____

NO. OF FETUSES: ☐

TWIN TYPE: MC / DC

SONOGRAPHER: _____ (DQASS code & initials)

FETAL MEASUREMENTS (in mm):
CRL (1) _____ (2) _____
If CRL >84mm, please provide HC
HC (1) _____ (2) _____
NT (1) _____ (2) _____
SCAN GESTATION ____weeks ____days
Vanished Twin Y / N (if yes, state CRL above)

IVF PREGNANCY: Y / N

IVF Type: _____

Extraction Date: ____/____/____

Transfer Date: ____/____/____

Donor Egg Y / N

Donor's DOB: ____/____/____

or Age at Harvest _____

MATERNAL WEIGHT (at sampling): _____kg

DATE & TIME COLLECTED

COLLECTED BY

KGH SAMPLE RECEIVED DATE

KGH LAB USE ONLY

2nd PROOF

Appendix 8 - Standard Operating procedure to failsafe Trisomy Screening Programme.

- Community Midwives document at booking on E3 the women's choice of screening
- Antenatal midwives review print out of the booking notes and select the timeframe for the screening scan
- Clinic coordinators create and send the scan appointment to the pregnant woman/person.
- When the pregnant woman/person attends the scan, the sonographer confirms their screening choice and documents this on the scan report. Where possible completes the NT measurement and records this on the scan report.
- If the screening cannot be completed because the NT could not be measured, the pregnant woman/person is told they need to make an appointment with the community midwife for Quad testing between 15- and 20-weeks' gestation. A printed information sheet is given to the woman/person explaining this in 6 different languages and is attached to the notes.
- If the pregnant woman/person is too late for 1st trimester screening, quad testing is offered and performed in the hospital.
- Pregnant women/people are told they will receive a written letter detailing the result within 2 weeks and are asked to ensure they have a follow up CMW appointment to discuss the results.
- Pregnant women/people who have had trisomy screening test within the hospital have their demographic details recorded on a list and this is forwarded daily to the screening team.
- A 'shipping manifest list', which contains all screening samples received by UHL laboratory, is emailed daily to the screening team (Mon-Friday). This manifest is then cross referenced with the patient demographic list and any samples not received by the laboratory are then identified and followed up to ensure a result is received by the pregnant woman/person.
- 5 weeks after the booking is entered onto E3, an automated weekly list of the bookings uploaded into the screening access database and weekly results for first trimester and second

trimester are added to the database using excel and data matches are electronic using NHS number.

- The screening team then follow up any missing results to ensure that all pregnant women/people who request to have Trisomy screening, receive a conclusive result.

High chance results

- The screening team will check on several occasions throughout the week with the fetal medicine midwives to ensure that all high chance results are actioned within the required timeframe and forwarded to the Screening Team. (The weekly list then contains pregnant women/people who to date have not had trisomy screening).
- The screening team then follow up, using this list, all pregnant women/people who accept screening to ensure they receive a conclusive result within the screening timeline.

Standard operating procedure for the daily checking of the Down's shipping manifest

- Check sticker list against shipping manifest
- Check for late bookers on Failsafe Officer's calendar against shipping manifest and E3/HISS
- Check 'Chase Quads' on Failsafe Officer's calendar against ilab and Downs cohort.
- Any not ticked on sticker sheet: Check E3 to see if bloods should have been taken & check iLab for different names. If blood should have been taken, check list at blood room and check with pregnant woman/person if they had the bloods taken. Email Downs labs to see if the samples arrived after the manifest was generated. If time allows, wait a day for bloods to arrive. If not arrived, arrange a retest. If the pregnant woman/person had declined at their scan, then cross them off the sticker list and write 'declined' through their name.
- Any not ticked on shipping manifest, will most probably be a quad test. Highlight these, and check S number against Downs cohort. If pregnant woman/person is on the cohort, then write 'on manifest (enter date)' in comments section. If not on cohort, then write 'no cohort' on the manifest next to the woman's/person's name.
- Check 'Quads taken in the community' worksheet to see if any Quads have been taken, check they are on the manifest.
- Any NT could not be measured on the sticker list in error, put woman/person in the Failsafe Officer's calendar when they will be 17/40.
- Go through the Downs cohort: Underneath the Downs buttons section, starting with 'Show 17+weeks (still open)' button. Chase Quads from this section, or later bookers/DNAs
- Next click on 'Scans booked but not yet attended' button. Work your way through any scans from previous day.
- Next, check 'Approaching 14 weeks (still open)'
- Then check 'Between 11+1 and 13+2 weeks (still open)'
- Make sure all 'gestation at booking' are filled in on the cohort and updated whilst going through the cohort. Click 'refresh gestations' at the end of daily checks, then 'refresh this form'.

Appendix – 9 NUH non-completed screening document

Nottingham University Hospitals NHS Trust

Title of Standard Operating Procedure	**Management of Trisomy screening failsafe for pregnant women/people being cared for by Leicester area midwives**
Author ***(include email and role)***	**Angie Godfrey** (*Screening Coordinator-* angie.godfrey@nuh.nhs.uk **Emma Haworth** (*Screening Coordinator-* emma.haworth@nuh.nhs.uk
Division & Specialty	Family Health - Obstetrics
Manager	Bodil Hamilton –Cody
Version	Version 1
Ratified by	Maternity Governance
Date ratified	02.09.2019
Date of review	01.09.2020
Audience	NUH and UHL Antenatal and Newborn screening Coordinators , Antenatal and newborn screening failsafe officer Leicester community matron Leicester community midwives

This procedure is to be followed each time combined screening cannot be completed at NUH

1 **Responsibility of NUH Antenatal clinic maternity support worker / midwife to failsafe the NT list**

- Document on NT list reason for combined screening not performed
- Document on NT list any actions, e.g. contacted pregnant woman/person, appts made
- If unable to contact a Nottingham pregnant woman/person. Telephone community coordinator to follow up (07812268458)
- Document actions on MEDWAY
- Write any follow up actions in the ANC communication diary where appropriate
- Sign, date and document number of pages on NT list
- Scan in the NT list to the Antenatal scan folder.
- This will be checked daily by the Antenatal and Newborn Screening Failsafe Officer /Screening Coordinator who will then add all pregnant women/people who have not had combined screening to the Trisomy failsafe spread sheet.
- Keep NT lists for 1 month. The failsafe officer / ANNBSC will check on the 5th of each month all sheets have been received for the previous month and confirm with each ANC they can be disposed of

2. Responsibility of NUH Failsafe officer / Screening Coordinator for pregnant women/people cared for by Leicestershire community midwives

- Every Tuesday email via **NUHNT.antenatal-empath@nhs.net** , (NUHANNBscreening@nhs.net details of pregnant women/people to UHL ANNBSC who are 18 week gestation
- UHL to return outcome / action by the Friday of the same week (where a bank holiday occurs clear timescales to be agreed by email)
- Monitor Failsafe until outcome obtained e.g. decline or result obtained

NUH Screening coordinators and Failsafe officer contact details

Screening team generic email address NUHANNBscreening@nhs.net

Antenatal and newborn screening coordinator

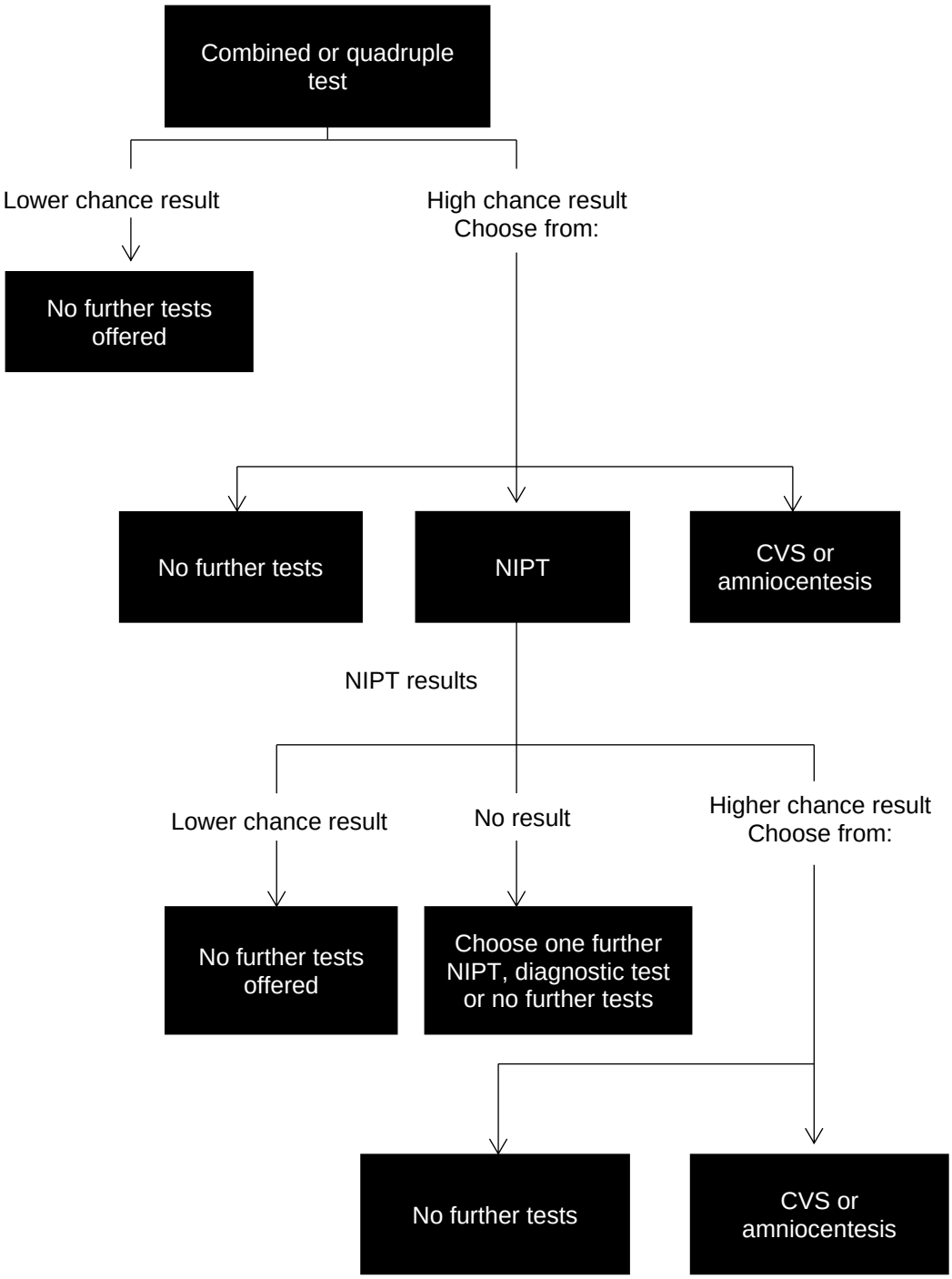
City Campus – 0115 9691169 ext. 57408 mobile 07812268657

QMC Campus – 0115 9249924 ext.63986 mobile 07812268656

Failsafe officer – mobile 07812270085

Appendix 10 – High chance trisomy screening result pathway inc. NIPT on the NHS

START HERE



This flowchart shows your options. A full text description of this pathway is also available at www.gov.uk/government/publications/fetal-anomaly-screening-care-pathways/downs-syndrome-edwards-syndrome-and-pataus-syndrome-screening-pathway-combined-or-quadruple-test-taken-on-or-after-1-june-2021

Appendix 11 – Discussion crib sheet

Crib sheet for discussion about a high chance Trisomy screening result including NIPT on the NHS

Following a high chance Trisomy 13/18 or 21 combined or quad test result all pregnant women/people continue to have the choice to:

1. Do nothing
2. Have a diagnostic test with associated risk of miscarriage of 1:100 results available with 1-3 days)

Once NIPT is available on the NHS also discuss option to:

3. Have a NIPT (if eligible). Refer to the following for help with this discussion:
    a. NIPT is still a screening test but is more accurate than the combined or quad test.
    b. Results available in 1-2 weeks
    c. **A high chance NIPT result for T21 or T13/18 means that more than 90% of babies will have the condition** and if this is the case, we would offer them a diagnostic test to confirm the result.
    d. **A low chance NIPT result means that it is unlikely that the baby has the condition, but** this is more likely if the result from combined/quad testing was really high (1:2-10).

This information is available to women/people electronically by scanning this QR code:

https://phescreening.blog.gov.uk/wp-content/uploads/sites/152/2021/06/FASP-DEP-higher-chance-leaflet-plain-A4-PDF-version.pdf

For pregnant women/people who want more information about the likelihood of a low chance result being incorrect and the baby is affected with the condition you can use the figures:

I. **T21**- Any high chance Combined/Quad result – low chance NIPT = 1:1100 that the baby is affected with Down's syndrome.

II. **T21** Combined/Quad result **1:2** – low chance NIPT = 1:100 that baby is affected with Down's syndrome. The pregnant woman/person should be aware that this result is much lower than their result from combined/quad test.

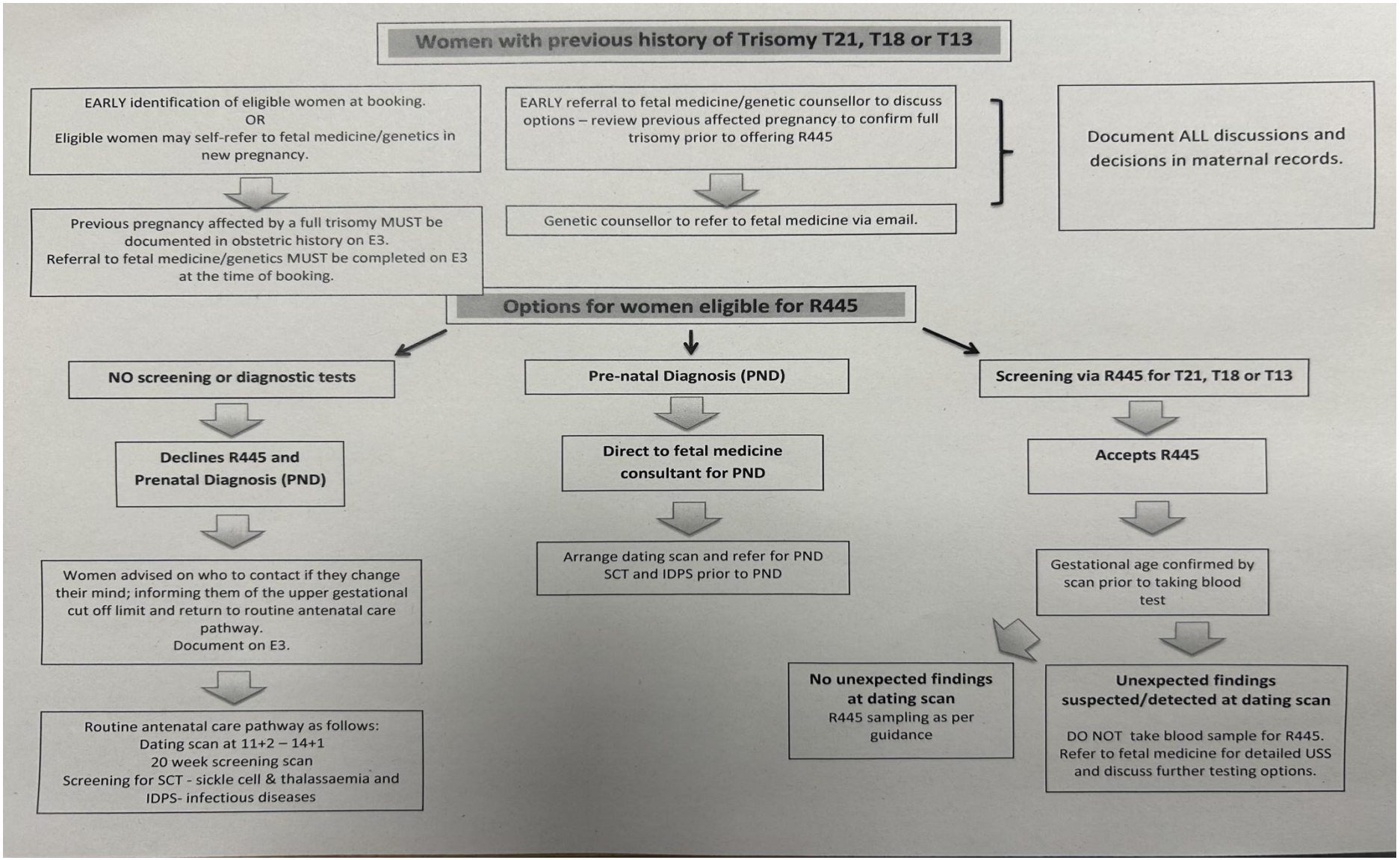
III. **T21** – 50% of babies with T21 will show an abnormality at the anomaly scan where further discussion would take place about the low chance NIPT result and the offer of diagnostic testing should be considered.

IV. **T13/18** – Any high chance Combined/Quad result – low chance NIPT = 1:480 that baby is affected with Edward's or Pataus syndrome.

V. **T13/18** Combined/Quad result **1:2** – low chance NIPT = 1:21 that baby is affected with Edward's or Pataus syndrome. The pregnant woman/person should be aware that this result is lower than their result from combined/quad test.

VI. **T13/18** – 95% of babies with T13/18 will show an abnormality at the anomaly scan where further discussion would take place about the low chance NIPT result and the offer of diagnostic testing should be considered. This may be a reason for pregnant women/people with a low chance NIPT result for T13/18 from NIPT to wait for the anomaly scan prior to making a decision about a diagnostic test.

Appendix 12: Pathway and information for the offer of R445 on the NHS for previous history of Trisomy 13/18 or 21.



R445 is Non-Invasive Prenatal Testing (NIPT) for women/people with a previous pregnancy with Down's (T21), Edwards' (T18) or Patau's (T13) syndrome. It provides an earlier opportunity to access and receive screening to make informed decisions about the current pregnancy.

Key Facts

- R445 ***replaces*** combined or quadruple screening for eligible women/people.
- R445 has greater sensitivity than combined or quadruple screening.
- R445 can be offered at later date if T21, T13 or T18 in previous pregnancy was disclosed after the booking appointment provided not ≥21+6*
- It cannot tell if their baby definitely has one of these conditions but may inform further decisions about the pregnancy.
- NIPT may not detect partial trisomies translocations or mosaicism.
- NIPT will not detect other chromosome conditions.

*discuss with NIPT lab prior to taking and sending sample to explain reasons for offering R445

Eligibility	Exclusion	Results
All women/people with history of previous pregnancy with full trisomy – T21, T18 or T13 From 10 weeks – 21+6 weeks confirmed by ultrasound but to be carried out in line with dating scan (11+2-14+1) as unexpected findings may help inform decisions about PND or R445 Two NIPT attempts – 2nd offered if 1st one fails Twin pregnancies can have R445	Other trisomy in previous pregnancy (trisomy other than T21, T18 or T13)* Not full trisomy in previous pregnancy (mosaicism, translocation, partial trisomy of T21, T18 or T13)* One of the parents has a Robertsonian or balanced translocation of T21, T18 or T13* Women/people using a donor egg in current pregnancy* Maternal cancer (unless in remission) Blood transfusion in last 4 months (whole blood or plasma) Bone marrow or organ transplant recipient Vanished twin pregnancy (empty 2nd pregnancy sac or sac containing non-viable fetus) Maternal T21 Maternal balanced translocation or mosaicism of T21, T13 or T18 Immunotherapy in current pregnancy excluding IVIg treatment Stem cell therapy **referral to genetic counselling and/or fetal medicine should be offered*	Reported same way as other NIPT samples taken for higher chance combined or quadruple screening test. Reported as higher chance, lower chance or no result. No numerical values are reported. **Samples with 'no results'** • No further tests • One further R445 NIPT test (repeat) • PND (CVS or amniocentesis) **Failed R445 – second attempt** • No further tests • PND (CVS or amniocentesis) *In these instances combined or quadruple tests should NOT be offered as these tests have lower sensitivity than R445.*

Appendix 13 - Standard Operating Procedure for Extra scan lists in Maternity which are outside of normal office hours – management of Trisomy screening blood samples.

- The team arranging the extra list should inform the labs that they should be expecting trisomy screening samples out of normal office hours.
- All women who have blood tests should be added to the sticker list and this should be sent to the screening team.
- All data on the request forms must be double checked for accuracy before sending to the laboratory.
- At the end of the list someone should be identified to be responsible for taking the samples to the laboratory.
- The screening team will check the sticker list against the shipping manifest to Kettering on the next working day.

NB - Samples must not be left unattended in blood rooms or MAU blood collection zones, as these areas may not be checked out of hours.