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NATIONAL PROTOCOLS

Programme: Fetal Anomaly and Down's syndrome screening

Release: Version 1.0

Date: April 2015

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Foreword

Screening for Down's syndrome and fetal anomalies has been undertaken in Scotland for many years, however it was recognised that there were variations in the programmes offered within Health Boards. In 2001 following guidance from the United Kingdom (UK) National Screening Committee (NSC), Health Department Letter HDL(2001)34 was issued which set out the steps required to standardise screening for Down's syndrome. There was no HDL detailing policy regarding the offer of second trimester fetal ultrasound examination released at that time, however in the Scottish Executive Health Department 2002 report A Framework for Maternity Services in Scotland[1] and the 2003 follow up report of the Expert Working Group on Maternity Services that was concerned with implementation of the Framework[2] it was recommended that there should be a comprehensive antenatal diagnostic and screening service which would include a dating ultrasound examination, blood serum screening for Down's syndrome and Neural Tube screening as well as a detailed second trimester ultrasound examination. In addition to this the 2004 NHS Quality Improvement Scotland (NHS QIS) Health Technology Assessment (HTA) report 5 on Routine ultrasound scanning before 24 weeks of pregnancy[3] recommended that all boards should offer a first trimester nuchal translucency (NT) ultrasound examination combined with first trimester serum screening and a second trimester fetal anomaly ultrasound examination.

Whilst HDL (2001)34 was fully implemented, the ultrasound screening services offered in Scotland continued to be varied across the Health Boards. In July 2008 Chief Executive Letter CEL31(2008) was issued taking account of the updated advice from the UK NSC and the recommendations of the NHS QIS Health Technology Assessment Report 5 – Routine Ultrasound Scanning before 24 Weeks of Pregnancy. This set out a number of changes and developments to strengthen and extend the pregnancy and newborn screening programmes. This included the introduction of a combined first trimester screen for Down's syndrome, in which measurement of biochemical markers in the mother's blood is combined with the ultrasound measurement of nuchal translucency in the fetus; strengthening the second trimester screen to quadruple markers, for those women who do not present early enough in their pregnancy to take advantage of first trimester screening. Additionally, all Board areas were to universally offer a second trimester fetal anomaly ultrasound examination.

Subsequently CMO (2011)6 was issued on the 19 May 2011 setting out further developments to enhance the Down's syndrome screening programme. In order to meet national performance standards the cut off point to be adopted for a high chance result was changed to 1 in 150 or greater for both first and second trimester Down's syndrome screening and the gestational range for second trimester Down's syndrome screening was revised to 14 weeks + 2 days to 20 weeks + 0 days of pregnancy.

[1] Scottish Executive Health Department. 2002a. *A framework for Maternity Services in Scotland*. Edinburgh: SEHD

[2] Scottish Executive Health Department. 2003. *Implementing "a framework for Maternity Services in Scotland".* Overview report of the expert group on acute maternity services. Edinburgh: SEHD.

[3] NHS Quality Improvement Scotland. 2004. Health Technology Assessment report 5. *Routine ultrasound scanning before 24 weeks of pregnancy.* Edinburgh. NHS QIS

Key Clinical Issues Summary

Part 1 Down's syndrome Screening

a) Nuchal Translucency (NT) as part of Combined Ultrasound and Biochemical Down's syndrome Screening

1. Qualifications

Any health professional carrying out a nuchal translucency measurement for the purpose of screening and diagnosis of a related condition should have had specific training in undertaking this role, participate in ongoing quality control and hold, as a minimum, one of the following:

- Certificate/Diploma (as appropriate) in Medical Ultrasound (CMU/DMU) of the Society and College of Radiographers (SCoR) with evidence of appropriate continuous professional development (CPD).
- Post Graduate Certificate in Medical Ultrasound (PGCert.MU) approved and validated by a Higher Institute of Education (HIE) and accredited by the 'Consortium for Sonographic Education' (CASE) with evidence of appropriate CPD. The qualification should be relevant to obstetric ultrasound practise.
- Royal College of Obstetricians and Gynaecologists (RCOG), Royal College of Radiologists (RCR) Diploma in Obstetric Ultrasound , RCOG Immediate Ultrasound of Normal Fetal Anatomy Training Programme (Module 3) and Advanced Training Speciality Module (ATSM) in Fetal Medicine. Evidence of appropriate CPD should also be provided[*].
- Sonographers, who do not have a UK recognised ultrasound qualification i.e. those trained overseas, should be registered under the Voluntary Register of Sonographers[#].
- Written evidence or certification for obstetricians or radiologists detailing previous obstetric ultrasound training and experience in this or another country.

Additional in house training and ongoing supervision and support will be provided by a Screening Support Sonographer (SSS) who has undergone the NE of England training programme.

2. Equipment *(See appendix 1)*

The following is recommended

- A room based wheeled B mode scan machine, with adequate monitor size and the ability to adjust monitor and control consol heights with warranty and quality control;
- Colour, power and spectral Doppler should be available;
- A convex abdominal and transvaginal (TV) ultrasound transducer should be available;
- An obstetric measurement package with 0.1mm minimum precision, variable size and shape continuous motion calipers. Ellipse circumference area measurement;
- Cineloop.

3. The Appointment

- Appointments for a booking ultrasound examination including NT measurement should be 20 minutes as a minimum. This should include time to get on and off the couch, time to perform the ultrasound examination and to write the report;
- If there is no visible heartbeat demonstrated during an ultrasound examination NICE Guideline 154 - *Diagnosis and initial management in early pregnancy of ectopic pregnancy and miscarriage* should be followed. http://www.nice.org.uk/nicemedia/live/14000/61854/61854.pdf
- The decision whether the woman has chosen to have a NT measurement performed as part of a combined Down's syndrome screening programme should be clearly indicated to the sonographer before the start of the ultrasound examination. This should involve the referring practitioner obtaining signed consent from the woman prior to the ultrasound examination. The

[*] It is recognised that many senior experienced scanning professionals (obstetricians, radiologists and sonographers) may not have a formal qualification and would no want to prevent these very important providers of the service to be excluded from practicing. These individuals should continue scanning under a 'grandfather clause' whilst applying the above qualification parameters to new practitioners to first trimester screening.

[#] The database is kept and controlled by the Society and College of Radiographers (SCoR) in association with the UK Association OF Sonographers (UKAS).

woman has the option to change her mind and decline NT measurement, even after the ultrasound examination has started;

- NT measurements should be performed when the CRL is greater than or equal to 45mm – less than or equal to 84mm (see appendix 2); using the current BMUS 2009 dating formula, the gestational age range for NT measurement is from 11 weeks + 2 days to 14 weeks + 1 day. Further information regarding the criteria for measurement of fetal crown rump length (CRL) as part of combined screening for Trisomy 21 can be found in appendix 3;
- Where both dating and Down's syndrome screening are requested, and the CRL is greater than or equal to 45.0 and less than or equal to 84.0mm the pregnancy should be dated by CRL and combined screening performed.
- Where both dating and Down's syndrome screening are requested and the CRL is ≥ 84.1mm, the pregnancy should be dated by HC.
- If the HC is ≥101.0mm and the gestational age is ≥14 weeks + 2 days, date by HC. The CRL should be ignored as it is ›84.0mm. Quadruple screening should be offered.
- If the HC is ‹101 mm and the CRL is ›84mm, date by HC. If the gestational age as calculated from the HC is ≤ 14 weeks + 1 day, the woman should be informed that the NT risk cannot be calculated from a CRL ›84mm, even though the gestational age of her pregnancy as estimated by the HC, lies within the gestational age window for combined screening. Combined screening is not an option but quadruple screening can be offered from 14 weeks + 2 days gestation.
- Transvaginal ultrasound examination should be available if required although the majority of measurements will be achieved with a transabdominal approach;
- If it is not possible to obtain a measurement during the first appointment, at least one other attempt should be offered, whether on the same day or at a later date. If it is not possible to obtain an accurate NT measurement despite 'twice on the couch' then further attempts do not have to be offered but mid trimester serum screening should be offered. Further information regarding vaginal bleeding and vanishing twin can be found in appendix 4;
- It is best practice to take the blood for the serum component of the test on the same day.
- If there is a history of significant maternal vaginal bleeding at the time of the first trimester screening for Down's syndrome, combined screening can still be undertaken (see Appendix 4).
- When ultrasound shows that there is an empty second pregnancy sac, combined screening is still appropriate. If the ultrasound shows that there is a second sac containing a fetal pole with no heart activity (sometimes called 'vanished' twin a risk calculation based on the maternal age and nuchal translucency only should be utilised (see Appendix 4).
- During the scan, ideally, more than one measurement should be taken and **the maximum measurement that meets all the criteria** recorded as that which to use in the calculation of risk (see appendix 5);
- If the NT is >3.5mm all women (even those with very large NT's) should be encouraged to have bloods taken for Pregnancy-associated plasma protein A (PAPP-A) and Human Chorionic Gonadotropin (HCG) estimated;
- Women of any age or gestation with an NT over 4mm can be offered invasive testing without waiting for the risk calculation. Even if the other risk factors are as low as possible, the risk is very likely to be no better than 1 in 30;
- Women with NT measurements of between 3.5-4mm can be offered invasive testing without waiting for the risk calculation as it is likely that the risk will be much increased. There is a chance that the risk may be much lower than expected so waiting for the combined risk calculation before proceeding to invasive testing is also reasonable;
- Women with an NT of 3-3.5mm should ideally wait for the risk calculation before considering invasive testing unless at the extremes of age.
- Where the NT is greater than or equal to 3.5mm referral for discussion with midwives or doctors with up to date expertise in managing fetal anomaly is recommended and considered good clinical practice.

4. Quality Assurance of NT/CRL Measurement

- Each operator should have a unique identifier;
- There should be a statistically validated system that enables comparison of the median NT deviation (mm) from target value as set by the FMF regression curve for CRL v NT. This

requirement will be met by a DQASS assessment, provided at 6 monthly intervals. A local interim report will be prepared and distributed 3 months into each 6 month DQASS period. Target performance figures for each report will differ;

- There should be systems to notify individual operators shown to return measurements (over a 6 month period) with median deviations greater than 0.1 mm from the gestation corrected target.
- There should be a system in place to support operators failing to achieve the target performance. This will include the review of images using a structured scoring system. Performance should be closely monitored for the subsequent 3 months;
- All practitioners should be subject to monthly audit and image review
- Any practitioner undertaking less than 25 NT assessments in a 6 month period should not be performing NT (unless this was due to illness or maternity leave or similar temporary issue)
- Review of images should be undertaken by a Screening Support Sonographer (SSS). Where any department is itself too small to be performing sufficient examinations for statistical analysis there should be established links with their nearest larger centre;
- The SSS should not audit their own work and if there is no suitable colleague or deputy SSS then the audit should be undertaken by another SSS from another health board area
- Each unit should participate in Regional and National audit of the screening and diagnostic processes.

b) Maternity services and Down's Screening Laboratories

1. Qualifications[4]

- All professionals involved in the provision and delivery of antenatal screening for Down's syndrome should undergo education which is recognised by the UK National Screening Committee. This includes training offered by: Professional Colleges, Institutes of Higher Education, allied institutions and national/local support organisations;
- All staff involved in the provision and delivery of screening for Down's syndrome, should participate in an ongoing educational programme that is an integral part of their CPD.

2. Equipment

- The computer software used to calculate the Down's syndrome risk must comply with the current national specification for risk calculation software. It must also be CE marked and comply with European Union directives[5];
- All analysers used require to be covered by a maintenance contract renewed annually.

3. Quality Assurance[6]

- The laboratory must be accredited by an appropriate body e.g. Clinical Pathology Accreditation UK (Ltd)/UKAS ISO 15189;
- The laboratory must participate in an accredited external quality assessment scheme e.g. UK National External Quality Assessment Scheme (NEQAS) and the Down's syndrome screening Quality Assurance Support Service (DQASS), and be able to demonstrate satisfactory performance;
- Appropriate internal quality assurance procedures must be undertaken and documented, e.g. weekly or monthly checks of screen positive rates, results of the analysis of internal Quality Control (QC) specimens documented and regular checks of MOM's (multiples of the median) marker values;
- A stand alone screening laboratory must have a workload of at least 10,000 Down's syndrome screening samples per annum to have sufficient confidence in the quoted annual screen positive rates, and to have sufficient specimens to calculate reliable, monthly median values for the biochemical markers;

[4] NHS Fetal Anomaly screening programme Antenatal Screening - Working Standards for Down's syndrome Screening 2007, Published April 2007.
[5] UK National Screening Committee National Down's Syndrome Screening Programme for England: National Specification for Risk Calculation Software and Guidance on Implementation October 2004
[6] NHS Fetal Anomaly screening programme Antenatal Screening - Working Standards for Down's syndrome Screening 2007, Published April 2007.

- Laboratories undertaking Down's syndrome serum screening must comply with the national standards in force at any particular time, regarding detection rates, screen positive rates and the cut off used to define the higher risk population;
- A protocol should be in place in the maternity service to allow women who have opted out of screening to be screened or referred for an appropriate assessment at a later date if wished;
- Clear systems must be in place for tracking samples, i.e. from the test being taken to the reporting of the result; it is the responsibility of the referring health professional or their deputy to ensure that a result is obtained for every woman who has accepted and undergone screening.
- For first trimester biochemical screening venous blood should be collected not earlier than 11 weeks + 2 days and not later than 14 weeks + 1 day;
- For second trimester biochemical screening venous blood should be collected not earlier than 14 weeks + 2 days and not later than 20 weeks + 0 days;
- Specimens should be kept at room or refrigerator temperature (but not frozen) and arrive in the laboratory as soon as possible but within 72 hours for first trimester samples and 5 days for second trimester samples of venepuncture.

4. Reporting results

- It is desirable that laboratories should return any higher chance results to the referring unit within 1 working day of the receipt of the bloods and essential that this should be within 3 working days;
- Where the screening test has identified a higher chance of Down's syndrome, an attempt to communicate the result to the woman should be made within 3 days of its receipt by the referring centre;
- There should be a timely opportunity for the woman to discuss the implications of a higher chance result with doctors and midwives with specific expertise in this area;
- There should be an established system of referral to diagnostic services that should provide medical and midwifery input with specific and up to date experience in diagnostic procedures and the management of fetal anomaly;
- An appointment to consider and if chosen, have performed a diagnostic test should be made available within 2 working days of the woman receiving the higher chance result.

Part 2 Fetal Anomaly Screening by ultrasound: Screening women who are at low risk of fetal anomaly

1. Qualifications[7]

All healthcare professionals undertaking a fetal anomaly ultrasound examination for the purpose of screening and diagnosis of a related condition should hold, as a minimum, one of the following qualifications:

- Certificate/Diploma (as appropriate) in Medical Ultrasound (CMU/DMU) of the Society and College of Radiographers (SCoR) with evidence of appropriate continuous professional development (CPD);
- Post Graduate Certificate in Medical Ultrasound (PGCert.MU) approved and validated by a Higher Institute of Education (HIE) and accredited by the Consortium for Sonographic Education (CASE) with evidence of appropriate CPD. The qualification should be relevant to obstetric ultrasound practise;
- Royal College of Obstetricians and Gynaecologists (RCOG), Royal College of Radiologists (RCR) Diploma in Obstetric Ultrasound, RCOG Intermediate Ultrasound of Normal Fetal Anatomy Training Programme (Module 3) and Advanced Training Speciality Module (ATSM) in Fetal Medicine. Evidence of appropriate CPD should also be provided[8];
- Doctors without a formal ultrasound qualification but who can demonstrate a large amount of experience in obstetric ultrasound with regular CPD;

[7] NHS Fetal Anomaly Screening Programme, 18^{+0} to 20^{+6} Weeks Fetal Anomaly Scan National Standards and Guidance for England; Published January 2010

[8] The NSC recognises that many senior experienced scanning professionals (obstetricians, radiologists and sonographers) may not have a formal qualification and would not wish to prevent these very important providers of the service to be excluded from practising. For these individuals NUSCoRG suggests that they should continue scanning under a 'grandfather clause' while applying the above qualification parameters to new practitioners to fetal anomaly scanning.

- Sonographers, who do not have an ultrasound qualification, should be registered under the Voluntary Register of Sonographers[9];
- Written evidence or certification for obstetricians or radiologists detailing previous obstetric ultrasound training and experience in this or another country;
- Practitioners should undertake a minimum of a half day obstetric fetal anomaly screening ultrasound session per week to maintain skills.

2. Equipment

As before ***(see appendix 1)***.

3. The Appointment

- Appointments should be at least 30 minutes which should include time getting on and off the couch, time to perform the ultrasound examination and to write the report (45 minutes for multiple pregnancies);
- Ultrasound examinations screening for fetal anomaly in low risk women should normally be performed between 18 and 20+6 weeks gestation;
- The base menu represents the minimum required to complete the screen. If it is not possible to complete the full base screen menu during the first appointment, at least one more attempt should be offered, whether on the same day or at a later date. In the absence of any suspicion of abnormality, if it is not possible to complete a full screen despite twice on the couch then further attempts do not have to be offered and the woman should be counselled accordingly. The base screening menu has been developed over 4 years with evidence taken from a number of publications; including those referenced in the National Institute for Clinical Excellence (NICE) 2008 guideline[10] (the full base menu is included as appendix 6 and the fetal cardiac protocol as appendix 7);
- Reports should be structured and ideally electronic. All required images should be electronically stored and archived to fulfil medico-legal requirements;
- The following images and measurements are required to be stored
    - Head Circumference (HC) demonstrating HC measurement and measurement of the atrium of the lateral ventricle
    - Suboccipito-bregmatic demonstrating measurement of the transcerebellar diameter
    - Coronal view of lips with nasal tip
    - Chest, 4 chamber view, both outflow tracts, lungs
    - Abdominal Circumference (AC) demonstrating AC measurement, cord insertion, kidneys, bladder
    - Femur Length (FL) demonstrating (FL) measurement, metacarpals & metatarsals x 2
    - Sagital and transverse view of spine including sacrum and skin covering
    - Amniotic fluid, placenta position noted
- A Down's syndrome risk generated by a nationally accepted screening method should not be recalculated up or down by the presence or absence of an ultrasound marker of less predictive power than increased nuchal fold;
- It is recommended that the term ultrasound "Down's syndrome soft marker" is no longer used and is now referred to as a 'normal variant'[11].

4. Quality Assurance

Ideally there should be a national congenital anomalies audit in order to determine:

- How many and which anomalies are identified by screening;
- How many and which anomalies are not identified by screening;

[9] The database is kept and monitored by the Society and College of Radiographers (SCoR).
[10] National Collaborating Centre for Women's and Children's Health. *Antenatal Care: Routine Care for the Healthy Pregnant Woman*, 2nd edn. RCOG Press, London, March 2008.
[11] RCOG (2000) '*Routine Ultrasound Screening in Pregnancy: Protocol, Standards and Training'*

- Which anomalies not identified by screening were potentially identifiable using the base menu;
- Outcome of pregnancies where an anomaly is identified by screening;
- Once an audit becomes established and the process of fetal anomaly screening analysed, it would be possible to identify Key Performance Indicators (KPIs), however the prevalence of individual anomalies may be too low for meaningful comparisons.

Until such data is available from a national congenital anomalies audit it is suggested that the following data be collected and collated:

- Number of women undergoing fetal anomaly screening within unit 18 – 20+6 weeks as a percentage of the number of women booking in that unit;
- Number of women in which a fetal anomaly is suspected and the nature of that anomaly;
- Number of women in whom fetal anomaly (and nature of that anomaly) was suspected but no fetal anomaly is identified at birth.

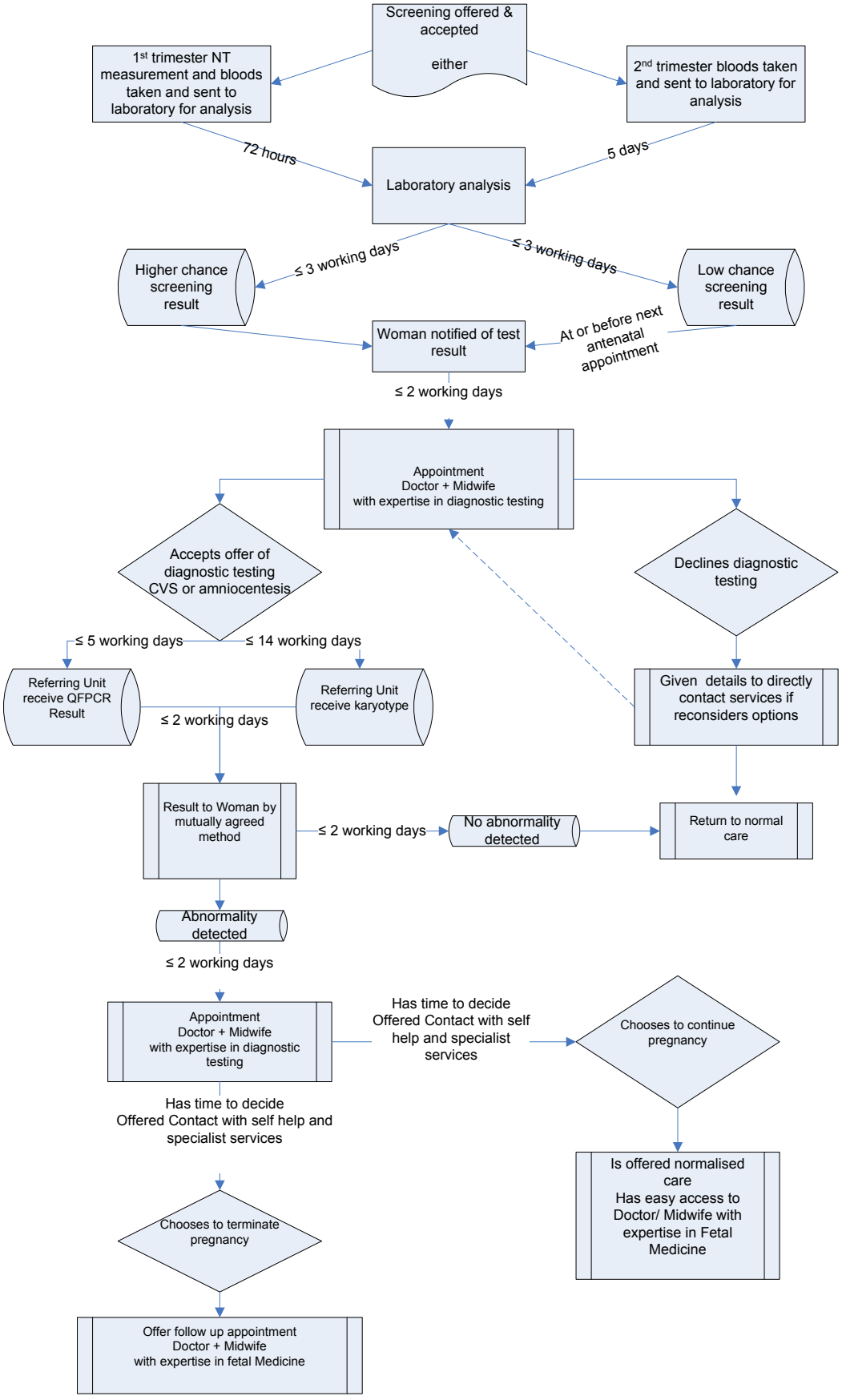
In an attempt to be able to benchmark nationally against the NHS Fetal Anomaly Screening Programme (NHS FASP) standards, on an annual basis each unit should provide (as a minimum) the screen positive rate (SPR) and detection rate (DR) for the following 11 conditions. It is hoped that the development of a Scottish national congenital anomaly audit will provide an evidence base for the usefulness of these conditions and highlight other parameters that may provide useful as performance indicators.

Conditions	Recommended Detection rate (%)
Anencephaly	98
Open spina bifida	90
Cleft lip	75
Diaphragmatic hernia	60
Gastroschisis	98
Exomphalos	80
Serious Cardiac abnormalities[12]	50
Bilateral renal agenesis	84
Lethal skeletal dysplasia	60
Edwards' syndrome (Trisomy 18)	95
Patau's syndrome (Trisomy 13)	95

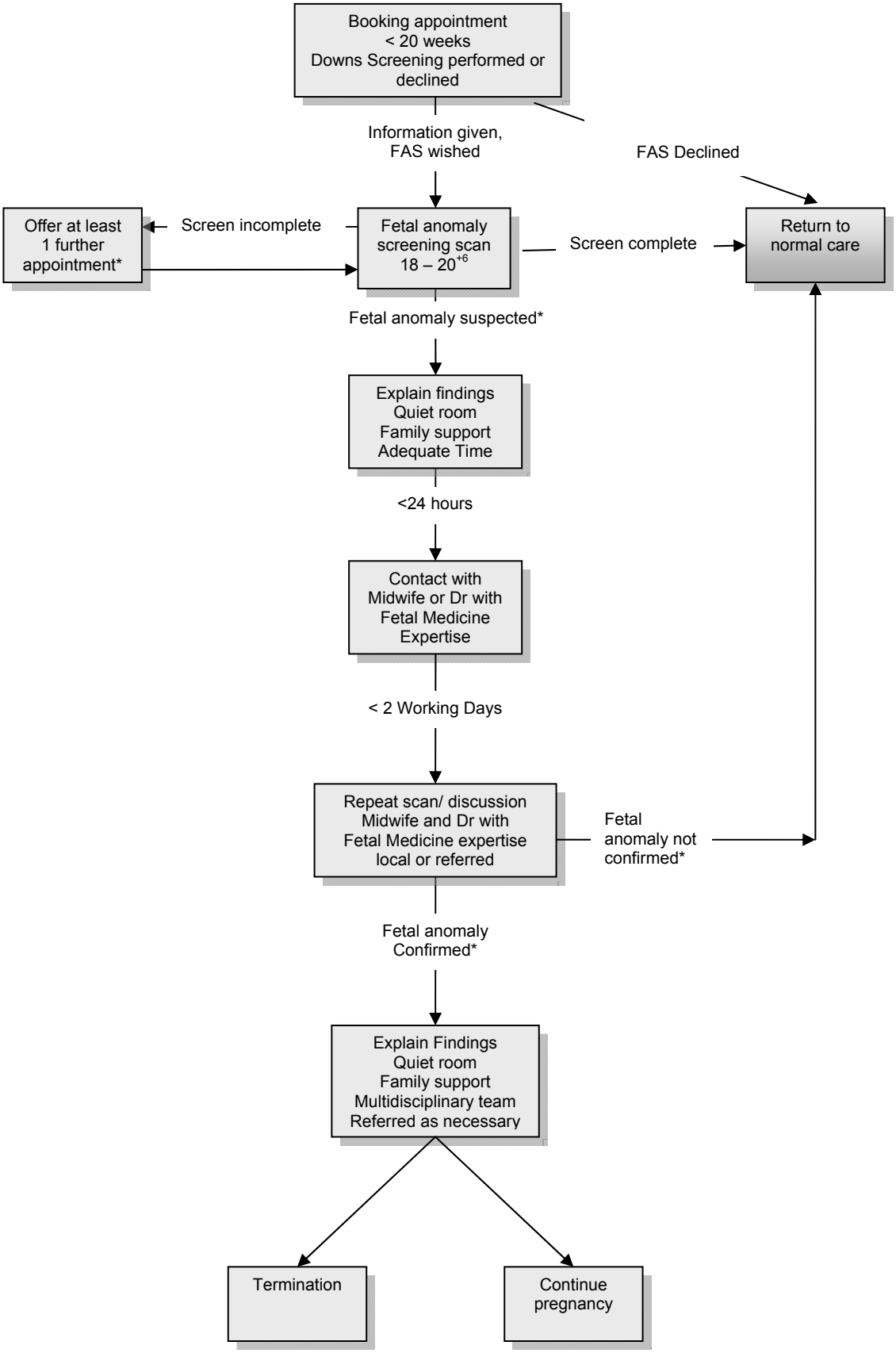
[12] *ICD10 code(s) used*
Q20.0 Common arterial trunk
Persistent truncus arteriosus
Q20.3 Discordant ventriculoarterial connection
Dextrotransposition of aorta
Transposition of great vessels (complete)
Q21.3 Tetralogy of Fallot
Q22.5 Ebstein's anomaly
Q23.4 Hypoplastic left heart syndrome
Q25.1 Coarctation of aorta

Patient Pathways

Down's syndrome screening pathway for women accepting screening



Low risk fetal anomaly screening pathway



1. Introduction

This document contains standard national protocols for all healthcare professionals involved in the NHS Scotland Down's and fetal Anomaly screening programme. In order to ensure equity of service across Scotland, NHS Boards are required to ensure that the screening service provided locally adheres to these protocols.

Standard operating procedures and local protocols are not included in the document; these need to reflect specific local arrangements and therefore need to be produced and maintained locally.

This document is intended to refer predominantly to low risk women. Women who have had a previously affected baby and those who have other medical conditions or family history of fetal anomaly may require specialised care.

2. Offer of Screening

The objective of these screening programmes is to offer fetal anomaly screening to all pregnant women in Scotland to provide them with information, which allows them to make informed choices. The programmes are in 2 main parts with screening and diagnostic components. Firstly, there is screening for chromosomal abnormalities, mainly Down's syndrome. Secondly there is screening for a variety of structural but also chromosomal and genetic fetal anomalies, by ultrasound at 18 - 20+6 weeks.

There are similarities but also marked differences in these 2 screening programmes. What both have in common is that they identify women who are at a higher risk of fetal anomaly. If there is an increased chance of fetal anomaly then referral into a diagnostic pathway should be offered. This document is primarily concerned with the processes of the screening rather than of the diagnostic services.

The majority of women identified as having an increased chance of a baby affected by Down's syndrome will have an unaffected pregnancy. Women should be offered a diagnostic test that if accepted, should, in the majority of situations, provide a definite result – the baby does or does not have Down's syndrome.

Women who have a suspicion of fetal anomaly following ultrasound screening should also be referred into a diagnostic process. Unlike Down's syndrome screening, a higher proportion will turn out to have a true fetal anomaly but unlike Down's syndrome screening, the diagnostic process frequently results in less certainty as to the nature and implications of a prenatal diagnosis.

Both screening techniques are aimed at informing women and offering choice: the choice whether to undergo diagnostic tests which may carry risks; the choice whether to seek more certainty in diagnosis by referral for specialist opinion and further imaging; occasionally the choice whether to undergo prenatal intervention and sometimes the choice whether or not to continue the pregnancy.

There are 2 opportunities for Down's syndrome screening: the preferred option that is more sensitive and specific is first trimester screening which provides an estimate of the chance of Down's syndrome between 11^{+2} and 14^{+1} week's gestation using a combination of maternal age; nuchal translucency and biochemical markers. For those women who do not present in time for first trimester screening, there is also a midtrimester screening option available from 14^{+2} to 20^{+0} weeks that provides an estimate of the chance of Down's syndrome using a combination of maternal age and a further set of biochemical markers. If a woman undergoes the earlier screening, with the current screening design the mid-trimester biochemical screen should **not also** be offered.

Fetal anomaly screening by ultrasound is usually offered between 18 and 20^{+6} weeks gestation. Women should be informed that the purpose of the ultrasound examination is to screen for fetal anomaly, and any who do not wish to be informed if abnormalities are found, should be advised that all the significant findings seen on the ultrasound examination will be reported and that therefore they should consider not having the screening test.

2.1 Responsibilities

NHS Scotland is responsible for ensuring that all pregnant women known to the service are provided with clear information in an appropriate format to help them make an informed choice about whether to take up any offer of screening. They should be offered appropriate screening for their gestation. If the offer of screening is accepted women should have the opportunity to become aware of the results of that screening as per NHS QIS standards.

There are some responsibilities which rest with the woman herself including: the registration of pregnancy in time to access pregnancy screening and testing; making the decision whether to undergo screening and testing; notifying the NHS if no result is provided within the agreed timeframe; and where offered, attending appointments for onward care.

Each Board should have in place a *Multi-Disciplinary Clinical Steering Group* to oversee the clinical management, governance and quality of the Boards pregnancy and newborn screening programmes.

The Multi-Disciplinary Steering Group should set out a comprehensive strategic plan for improving quality in accordance with the Board's overall service developments; develop policies aimed at managing and reducing clinical risk and ensure inter-agency arrangements are in place to support women through the screening and diagnostic pathways

The group also has a responsibility for:

- Contributing to the development and implementation of screening and diagnostic care pathways in line with national standards and policies;
- Ensuring that all care pathways are regularly reviewed and modified in line with the national programme's changing standards and policy;
- Ensuring arrangements are in place for the audit of Down's syndrome and ultrasound fetal anomaly screening programmes and linking to an agreed quality assurance framework;
- Providing a supportive framework for women and their families who have higher risk of, or are found to have a pregnancy with an abnormality;
- Advising and supporting staff on antenatal screening and diagnostic issues;
- Communicating with primary care services;
- Providing an ongoing education and training programme for staff offering screening and diagnostic testing to improve awareness and skills and reduce risk of serious untoward incidences;
- Providing an annual screening report which reflects the national minimum audit criteria for Down's syndrome and ultrasound fetal anomaly screening programmes.

2.2 Process

All pregnant women attending an antenatal booking clinic, or being seen in the community, should be given written information on the screening tests available in time to seek more information and make a decision whether to undergo testing.

All women who present before 20^{+0} weeks of pregnancy should be offered Down's syndrome screening appropriate to their gestation. Those under 20^{+6} weeks should also be offered screening for fetal anomaly by ultrasound examination. Women first attending at 21 weeks or over should be offered an ultrasound at which there may be the opportunity to screen for fetal anomaly although it should be explained that the screening is likely to be less complete at later gestations. It should also be explained that screening that will provide a calculation of the chances of Down's syndrome is not available.

Boards should identify a designated senior midwife who is responsible for ensuring that:

- Every eligible woman is given the opportunity to be screened;
- Both a primary and failsafe mechanism is in place to ensure that results (both higher chance and lower chance) are received for all women screened;
- Women have the opportunity to receive the results in writing with the offer of appropriate counselling and onward care into diagnostic pathways.

The host Board of the antenatal clinic/community maternity service is responsible for the clinical governance of the service and for ensuring that:

- Every health professional involved in offering/performing a screening test is suitably qualified and trained;
- Every woman who attends an antenatal clinic before 20+0 weeks is offered screening for Down's syndrome;
- Every woman who attends an antenatal clinic before 20+6 weeks is offered fetal anomaly screening by ultrasound;
- Every woman who books for delivery at hospital or at home is offered screening appropriate to her gestation;
- Information on the offer made (and whether or not it is accepted) is recorded in case notes or equivalent;
- Where gestationally appropriate, two screening ultrasound examinations are offered to all pregnant women presenting for maternity care; a dating ultrasound examination ±NT measurement from the equivalent CRL of greater than or equal to 45mm – less than or equal to 84mm (around 11^{+2} -14^{+1} weeks of pregnancy) and a fetal anomaly ultrasound examination between 18^{+0} to 20^{+6} weeks of pregnancy.

3. Consent for Screening

Information should be made available taking into account of the woman's physical, cultural, ethical educational and mental health needs preferably 48 hours in advance of the screening tests, unless precluded by late presentation.

Women and their partners should be provided with information about the implications of the screening tests. For Down's syndrome screening, this should include: implications of receiving high or low risk result; information on the false positive rates of the screening test; the techniques involved and risks that may be associated with any diagnostic tests and also information about the conditions themselves. For fetal anomaly screening by ultrasound there should be information about the range of anomalies that might be suspected with examples of the more common conditions and rates of their detection, the degrees of certainty that may follow an abnormal screening ultrasound examination and the chances of a false positive result.

Both screening systems should be discussed as 'options' rather than an inevitable aspect of routine maternity care.

Women must be given sufficient time to make decisions whenever options are presented.

Where maternal or fetal samples are being obtained, there should be information available about their storage and disposal. Should there be an interest in studying any excess material there should be a process for the woman to decline to give consent and for the laboratory to be notified of this decision.

It is the responsibility of the health professional to ensure that the correct information is entered into all fields when completing a screening request card.

If a woman declines a screening or diagnostic test, this should be recorded in the notes by the responsible healthcare professional. A protocol should be in place to allow women who have opted out of screening or diagnostic testing to change their mind and still undergo screening or testing at a later date, provided it is still possible at that gestation.

Supplementary information, including relevant informative/supportive websites or details of support organisations, should be offered to all women receiving a positive screening or diagnostic test result.

Professionals involved in screening for Down's syndrome and fetal anomalies should work collaboratively with appropriate agencies such as: social services, voluntary sector support groups, religious bodies and bereavement services; in order to provide a comprehensive support network that is centred on the woman's needs and requests.

4. Down's syndrome Screening

4.1 The screening process

4.1.1 Organisation

There must be an agreed local written policy which adheres to national standards to define the purpose of radiological and laboratory based assessment of risk of Downs and other chromosomal syndromes. Laboratories must be accredited by an appropriate body e.g. Clinical Pathology Accreditation (CPA) UK (Ltd)/UKAS, participate in an accredited external quality assessment scheme e.g. UK National External Quality Assessment Service (NEQAS), and be able to demonstrate satisfactory performance. Additionally the laboratory must submit screening data to Down's syndrome screening Quality Assurance Support Service (DQASS) at least twice a year.

Appropriate internal quality assurance procedures must be undertaken and documented, e.g. weekly or monthly checks of screen positive rates, results of the analysis of internal QC specimens and regular checks of median MoM marker values. The laboratory must participate in audit of the screening service at local and national level and provide an annual report, or the necessary data for the preparation of an annual report.

A stand-alone screening laboratory must have a workload of at least 10,000 Down's syndrome screening specimens per annum to have sufficient confidence in the quoted annual screen positive rates, and to have sufficient specimens to calculate reliable, monthly median values for the biochemical markers. Laboratories with a workload of less than 10,000 specimens a year must be part of a 'managed network' of no less than 3 laboratories, with each having a minimum workload of 5,000 specimens per year and identical screening policies and analytical procedures in force.

Software used to calculate the Down's syndrome risk must comply with the current national specification for risk calculation software. It must also be CE marked and comply with EU directives. Laboratories must comply with the national standards in force at any particular time, regarding detection rates, screen positive rates and the cut-off used to define the higher risk population. In the absence of universal or standardised maternity information systems, the laboratory information management system will be used as a repository of national aggregated data on the screening programme.

95% of pregnancy screening reports must be issued to an appropriate healthcare professional within 3 working days of receipt of the specimen and where appropriate, ultrasound examination measurements at the laboratory.

4.1.2 Multiple pregnancies

In Scotland at the current time, the risk of Down's syndrome can be estimated for each fetus based on a combination of maternal age and the NT measurement only. It should be explained to the woman beforehand that this provides a less accurate estimate of risk than that used for singleton pregnancies. In addition, the particular diagnostic and therapeutic implications and their variation with different chorionicities should be explained to all women with multiple pregnancies before undergoing screening.

Mid-trimester serum screening for Down's syndrome should not be offered for multiple pregnancies at this time.

4.1.3 Request form

The request form must contain fields which conform to the minimum dataset as advised by the laboratory which currently includes:

- Patient details (forename, surname, date of birth, Community Health Index (CHI) number)
- Hospital attended or other referral source
- The date the sample was taken

- Information on the pregnancy needed to interpret the screening results (date of USS, gestation, maternal weight, smoking status, previous affected pregnancy, ethnicity, diabetic status, assisted conception details)

4.2 Blood Specimens for Screening - first and mid-trimester Down's syndrome screening

All pregnant women undergoing screening for Down's syndrome must have a dating ultrasound examination before blood is taken. To establish gestation accurately, the British Medical Ultrasound Society (BMUS) recommends the use of CRL up to 14wks + 1day (equivalent to CRL = 84mm) and the use of HC after this gestation (*Loughna P, Chitty L, Evans T, Chudleigh T (2009). Fetal size and dating: charts recommended for clinical obstetric practice. Ultrasound, 17,161-7).* If there is no visible heartbeat demonstrated during an ultrasound examination NICE Guideline 154 - Diagnosis and initial management in early pregnancy of ectopic pregnancy and miscarriage, should be followed. http://www.nice.org.uk/nicemedia/live/14000/61854/61854.pdf

It is the referring maternity units' responsibility to ensure clear systems must be in place for tracking samples, i.e. from the test being taken to the reporting of the result.

Clotted blood samples are required for serum testing and should be the first sample taken at any one time of blood withdrawal. The local laboratory will advise the type of specimen tube should be used. Samples must not be collected in ethylenediaminetetraacetic acid (EDTA), and specimens should not be poured from one tube into another as this will interfere with the tests and invalidate results therefore necessitating a repeat blood sample. It is best practice to take the blood for the serum component of the combined first trimester screen on the same day as the NT measurement.

4.3 First trimester combined nuchal translucency (NT) and biochemical screening

If there is a history of vaginal bleeding at the time of the first trimester screening for Down's syndrome, the combined test can still be undertaken as the biochemical marker levels are not significantly altered.

If at the time of undertaking NT measurement an empty second pregnancy sac is identified, the associated levels of biochemical markers appear to be unaffected and no different to those in a singleton pregnancy. As such combined first trimester screening is still appropriate. If the ultrasound shows that there is a second sac containing a fetal pole but with no heart activity (sometimes called 'vanished' twin), it is possible that there could be a contribution to the maternal biochemical markers for many weeks after the fetal demise and in this event risk calculation should be based on the maternal age and nuchal translucency only. It should be explained to women (as for multiple pregnancies) that this provides a less robust estimate of a pregnancy being affected. Further information regarding vaginal bleeding and vanishing twin can be found in appendix 4.

4.3.1 Nuchal Translucency measurement

Nuchal Translucency ultrasound should be performed in a room designed for the purpose and the time allowed should be at least 20 minutes. This should include time to get on and off the couch, time to perform the ultrasound examination and to write the report. Imaging should be predominantly transabdominal but there should be the option of transvaginal imaging should be available for use as a last resort at the sonographer's discretion.

- NT measurements should be performed when the CRL is ≥45mm – ≤ 84mm; using the current BMUS 2009 dating formula, the gestational age range for NT measurement is from 11 weeks + 2 days to 14 weeks + 1 day. Where both dating and Down's syndrome screening are requested, and the CRL is greater than or equal to 45.0 and less than or equal to 84.0mm the pregnancy should be dated by CRL and combined screening performed.

- Where both dating and Down's syndrome screening are requested and the CRL is ≥ 84.1mm, the pregnancy should be dated by HC.
- If the HC is ≥101.0mm and the gestational age is ≥14 weeks + 2 days, date by HC. The CRL should be ignored as it is >84.0mm. Quadruple screening should be offered.
- If the HC is <101 mm and the CRL is >84mm, date by HC. If the gestational age as calculated from the HC is ≤ 14 weeks + 1 day, the woman should be informed that the NT risk cannot be calculated from a CRL >84mm, even though the gestational age of her pregnancy as estimated by the HC, lies within the gestational age window for combined screening. Combined screening is not an option but quadruple screening can be offered from 14 weeks + 2 days gestation.

It is essential that the NT measurement is as accurate as possible[13]. The ultrasound imaging equipment used should have a cineloop function and callipers that have a precision to one decimal point, i.e. 0.1 mm. Operators should be aware of and adhere to BMUS (British Medical Ultrasound Society) guidelines for safe use of ultrasound including exposure time limits. The algorithm within the ultrasound equipment for calculating gestation from CRL/HC must be the same as that in use by the laboratory risk calculation software.

A midline sagittal section of the fetus should be obtained with the fetus horizontal on the screen, either supine or prone (see image 1)[14]. The fetus should be in the neutral position with the head in line with the spine, neither hyper extended nor flexed. The NT should be measured once the magnification of the fetus is made as large as possible before the image is frozen, such that, only the fetal head and shoulders are visible on screen (see image 2). Small movement of the callipers on the magnified image must produce only a 0.1 mm change in the measurement. It may be helpful to reduce the gain to improve the image quality.

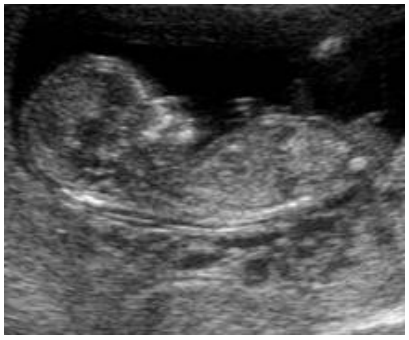


Image 1

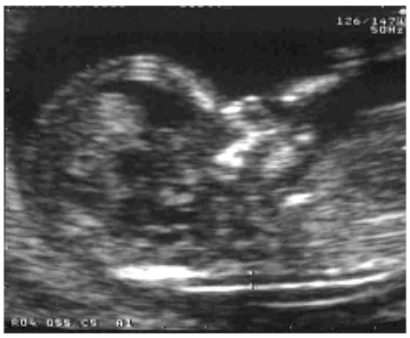
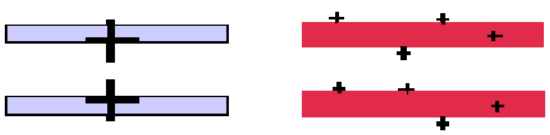


Image 2

Care must be taken to distinguish between fetal skin and amnion so that the amnion is not included in the measurement because both structures may appear as thin membranes. This is achieved by waiting for spontaneous fetal movement away from the amniotic membrane;

Diagram 1



Correct *Incorrect*

The maximum thickness of the subcutaneous translucency between the skin and the soft tissue overlying the cervical spine should be measured (see Image 2). The calliper selected should be a

[13] NHS Fetal Anomaly screening programme Antenatal Screening - Working Standards for Down's syndrome Screening 2007, Published April 2007.
[14] FASP (2010) Recommended criteria for measurement of fetal nuchal translucency (NT) as part of combined screening for Trisomy 21 within the NHS in England

vertical cross. Measurements should be taken with the horizontal lines of the callipers placed 'on' the lines that define the nuchal translucency thickness, not in the line and not in the translucency (see Diagram 1). The image should be unfrozen and the measurement repeated until the optimum image, meeting the above criteria has been obtained. This image must be retained in accordance with medico-legal requirements for 25 years.

It is best practice to take a measurement from more than one image. The maximum NT measurement that meets **all** criteria should be used for Down's syndrome calculation. Images should be stored of all measurements. It is the sonographer's responsibility to produce an NT result in mm that can be sent to the laboratory for analysis (the requirement is to send **one** measurement to lab).

Where it is not possible to obtain an NT measurement at the first clinic appointment, women should be offered a return appointment within the required gestational range. If unable to obtain a NT measurement at the subsequent appointment or it is not possible to offer a return appointment due to gestational age of pregnancy then the woman should be offered a mid-trimester biochemical screening test.

A NT greater than or equal to 3.5mm is a significant early pregnancy scan finding. It is associated with an increased risk of fetal cardiac, genetic and chromosomal anomalies. Referral to doctors/midwives with specific and up to date experience in diagnostic procedures and the management of fetal anomaly is considered as good clinical management even when screening for Down's syndrome has been declined.

If the NT is >3.5mm all women (even those with very large NT's) should be encouraged to have Pregnancy-associated plasma protein A (PAPP-A) and Human Chorionic Gonadotropin (HCG) estimated. Women of any age or gestation with an NT over 4mm can be offered invasive testing without waiting for the risk calculation. Even if the other risk factors are as low as possible, the risk is very likely to be no better than 1 in 30. Women with NT measurements of between 3.5-4mm can be offered invasive testing without waiting for the risk calculation as it is likely that the risk will be much increased. There is a chance that the risk may be much lower than expected so waiting for the combined risk calculation before proceeding to invasive testing is also reasonable. Women with an NT of 3-3.5mm should ideally wait for the risk calculation before considering invasive testing unless at the extremes of age.

4.4 Training and Education

Any health professional carrying out a nuchal translucency measurement for the purpose of screening and diagnosis of a related condition should have had specific training in undertaking this role, participate in ongoing quality control and hold, as a minimum, one of the following:

- Certificate/Diploma (as appropriate) in Medical Ultrasound (CMU/DMU) of the Society and College of Radiographers (SCoR) with evidence of appropriate continuous professional development (CPD).
- Post Graduate Certificate in Medical Ultrasound (PGCert.MU) approved and validated by a Higher Institute of Education (HIE) and accredited by the 'Consortium for Sonographic Education' (CASE) with evidence of appropriate CPD. The qualification should be relevant to obstetric ultrasound practise.
- Royal College of Obstetricians and Gynaecologists (RCOG), Royal College of Radiologists (RCR) Diploma in Obstetric Ultrasound , RCOG Immediate Ultrasound of Normal Fetal Anatomy Training Programme (Module 3) and Advanced Training Speciality Module (ATSM) in Fetal Medicine. Evidence of appropriate CPD should also be provided[*].

[*] It is recognised that many senior experienced scanning professionals (obstetricians, radiologists and sonographers) may not have a formal qualification and would no want to prevent these very important providers of the service to be excluded from practicing. These individuals should continue scanning under a 'grandfather clause' whilst applying the above qualification parameters to new practitioners to first trimester screening.

- Sonographers, who do not have a UK recognised ultrasound qualification i.e. those trained overseas, should be registered under the Voluntary Register of Sonographers[#].
- Written evidence or certification for obstetricians or radiologists detailing previous obstetric ultrasound training and experience in this or another country.

Additional in house training and ongoing supervision and support will be provided by a Screening Support Sonographer (SSS) who has undergone the NE of England training programme.

4.5 Blood tests

It is best practice that blood samples should be collected at the same appointment as that of the NT measurement being successfully obtained.

Specimens as whole blood should be kept at room or refrigerator temperature (but not frozen) and arrive in the laboratory as soon as possible but within 72 hours of venepuncture.

Samples and forms should be given a number and the details on the form entered into the laboratory computer system/screening database. The assigned number should be used to link sample, patient details and analytical results and appears on the final report.

Where blood samples are obtained ahead of the NT measurements, blood samples should be numbered and analysed immediately and the patient ID entered into the database. When the NT measurements are subsequently received and patient demographic details entered into the database, the computer should link the NT data with the blood sample and the same accession number should be applied to the NT form.

The first trimester markers, free beta subunit of hCG (FβhCG) and pregnancy associated plasma protein A (PAPP-A) are measured in serum by immunoassay. Results should be converted to MoM corrected for co-variables such as gestational age, maternal weight, smoking status, any previous affected pregnancies and assisted conception. A risk of Down's syndrome should be estimated from the corrected FβhCG and PAPP-A MoMs, NT MoM and maternal age.

After testing, any leftover serum is stored in numbered tubes in a freezer bank to allow repeat analysis in the event of an analytical problem. (These stored samples may also be used for quality control and anonymised research for development of new tests for prenatal screening as detailed in the patient information leaflet. Women can choose to opt out of their sample being used in this way).

[#] The database is kept and controlled by the Society and College of Radiographers (SCoR) in association with the UK Association OF Sonographers (UKAS).

4.6 Mid-trimester serum screening

Women who have undergone first trimester screening should **not** undergo mid-trimester screening.

Venous blood should be collected not earlier than 14 weeks^{+2} days and not later than 20 weeks^{+0} days. Specimens as whole blood should be kept at room or refrigerator temperature but not frozen and arrive in the laboratory as soon as possible but within 5 days of venepuncture.

Blood samples arriving in the laboratory must be correctly labelled and accompanied by the appropriate fully completed request form. Sample and form should be given an accession number and the details on the form entered into the laboratory computer/screening database. The assigned laboratory number should be used to link sample, patient details and analytical results and appears on the final report.

Serum is separated from the clotted blood specimen and the concentrations of Alpha-fetoprotein (AFP), intact human Chorionic Gonadotrophin (hCG), unconjugated oestradiol (UE3) and inhibin A should be measured using specific immunoassay methods. The concentrations are then converted to multiples of the median (MoM) and corrected for co-variables including gestational age, maternal age and weight, smoking status, ethnicity, diabetic status and if applicable, previous affected pregnancy and assisted conception to provide a risk of Down's syndrome.

All laboratories currently use similar methods for calculating risks for Down's syndrome (trisomy 21) from a combination of maternal age and additional factors, and biochemical results. When mid-trimester screening for Down's syndrome is performed AFP is one of the panel of markers used. When the AFP level is elevated, in view of the increased chance of NTD and other structural anomalies, the laboratory report will display a comment to this effect.

In normal circumstances women who have been successfully screened for Down's syndrome with combined ultrasound and biochemical markers in the first trimester (whether privately or through the NHS) should not normally undergo screening using AFP alone in the 2nd trimester. The recognised screening modality for NTD and Abdominal Wall Defects is the fetal anomaly scan at 18 – 20^{+6} weeks.

4.7 Laboratory Reports

The report issued from the laboratory must contain information which conforms to the minimum data set which should include:

- The patient (name, date of birth, CHI number)
- Hospital or other referral source
- The date the sample was taken
- The screening results expressed as 'The chances of …. syndrome are 1 in xxx'
- The information used to determine these results: gestation, Marker MoM; NT measurement (if first trimester screen); maternal weight; smoking status; family origin; previous affected pregnancy, etc) and maternal age.
- For AFP results greater than 2.0MoM the report should state that there is an increased chance of neural tube defect.

The report should also comment that the interpretation of the results is only correct provided the relevant information is correct.

Computer generated reports conforming to the agreed minimum dataset should be issued by each laboratory. Over 95% of results should be reported within 3 working days of receipt of the sample by the laboratory. First trimester laboratory reports should be communicated within 3 working days of receipt, if the sample and nuchal translucency measurements are received together. If the nuchal translucency measurements are taken later than the sample then the report should be issued within 2 days of receiving the nuchal translucency measurement.

All reports should be communicated to the referrer and on receipt; the details on the report should be checked. If any information is inaccurate, the amended information should be sent to the laboratory by e-mail using the proforma supplied before the woman is notified. The laboratory should recalculate the risk result and issue an amended report.

All women who are identified as having a lower chance of Down's syndrome should have the opportunity to receive the result in writing and for the report to be filed in the hand held notes.

For those women who are identified as having a higher chance of Down's syndrome, the results should be given priority and securely emailed and telephoned to the referrer depending on prior arrangement between referrer and screening laboratory. There should be a robust system in place so that any results arriving at the referral source can be identified immediately on their receipt. Email addresses used should be within the nhs.net domain and only accessible to the relevant staff.

Women who have undergone Down's syndrome screening should have previously indicated how they would like to receive any higher chance result and had their preference documented in the hospital notes.

Attempts should be made to communicate to the woman any higher chance result within 3 working days of the result being received from the laboratory and an opportunity to attend for a diagnostic appointment given within a further 2 working days.

On request, copies of all reports may be provided to the General Practitioner (GP) or to the original referral source if that differs from the unit where antenatal care is being provided.

All reports should be retained in electronic format by each laboratory. It should be possible for information and results relating to individual pregnancies held on the laboratories' screening database to be accessed by telephone enquiry from an identifiable and verifiable source.

All laboratory documentation should be retained for appropriate periods of time and disposed of as specified in the laboratory Standard Operating Procedure.

4.8 Failsafe

All results should be sent by the screening laboratory to the referral source unless indicated otherwise.

There should be a system in place to ensure that a result has been received for every woman who has been screened.

If no result has been received within the timeframe agreed with the pregnancy screening laboratory, the laboratory should be contacted and should provide a report as soon as possible. If no sample or request was received at the laboratory a repeat sample should be taken and sent to the laboratory as soon as possible.

For first trimester screening, provided the gestation is still less than 14^{+1} weeks, the original NT measurement can still be used but this should be expressed clearly on the repeat form with the gestation and CRL measurement at the time the NT measurement was obtained specified. Should repeat bloods be required for first trimester screening but by the time this has become apparent, the gestation exceeds 14^{+1} weeks, the woman should have the situation explained and quadruple serum screening then offered.

If further bloods for mid-trimester screening were taken but were not able to be processed by the laboratory and the gestation exceeds 20^{+0} weeks by the time this becomes apparent, the woman should be offered an appointment to discuss the issues with senior medical staff and other options including diagnostic testing considered.

4.9 The Diagnostic Process

4.9.1 Organisation

It is the responsibility of each Health board to ensure that there is a diagnostic pathway for all women identified as having a higher chance of Down's syndrome. Women should be counselled by medical and also by midwifery staff who have specific and recent experience in the diagnostic tests available.

The decision whether to have a diagnostic test should be the woman's choice. Women should be given time to make that choice even if it involves further appointments. Written information about the diagnostic tests, their techniques and risks should be made available at the time the woman is informed that there was a higher chance of Down's syndrome from the screening tests. Further information about the conditions themselves with local figures about techniques and number of tests performed by operators (but not necessarily about local miscarriage rates – see RCOG guideline: Amniocentesis and Chorionic Villus sampling, Greentop Guideline No 8, 2010) should be available at the time of the diagnostic appointment. The option to have a diagnostic test should not be dependent on the intention to terminate if an anomaly is identified but information about the techniques and processes involved in termination should be available if requested.

The invasive tests of chorionic villus sampling and amniocentesis should be conducted according to the standards laid down in the RCOG guideline: Amniocentesis and Chorionic Villus sampling, Greentop Guideline No 8, 2010. The auditable standards of this guideline should be monitored locally but also be available for national audit subject to the Board's policy on confidentiality.

4.9.2 Diagnostic results

Results by Quantitative Fluorescent-Polymerase Chain Reaction (QF PCR), karyotyping or other molecular genetic equivalent should be made available to the referring unit within agreed time frames.

Reporting times for molecular cytogenetics are incorporated in the cytogenetic service level agreement as they are dependent on the sample type which is reported through the cytogenetic service.

On receipt of the result by the referring unit, women should have the opportunity to receive the result within 2 days of its receipt by whichever method was previously agreed and documented in the notes at the time that the diagnostic test was performed. There should be the opportunity for all women to receive written confirmation of the result if they wish to.

Cytogenetic and molecular genetic departments must have a system to feedback pregnancy outcomes to the screening laboratory of any affected pregnancy whether or not a diagnostic test was performed during the pregnancy. This should be subject to the Board's policy on confidentiality and data protection and included in the annual report that should include data from a regional level and also by unit within that region.

5. Fetal Anomaly Screening by Ultrasound

5.1 The screening process

Fetal anomaly screening by ultrasound examination for women with a low risk of fetal abnormality should be performed between 18 and 20+6 weeks. To establish gestation, a first trimester ultrasound examination should be offered beforehand. The decision whether to undergo screening by ultrasound should not be dependent on the decision whether to undergo Down's syndrome screening: the two screening systems should be considered separately.

Some fetal anomalies will be identified at ultrasound examinations performed both earlier and later than 18-20+6 weeks and women should be made aware of these possibilities before each ultrasound examination is requested or performed.

5.2 Gestation calculation

Crown-rump length (CRL) should be used for estimating fetal gestational age up to 84mm. The head circumference (HC) and femur length (FL) can be used for estimating fetal gestational age after 13^{+0} weeks (see appendix 2 for dating table).

5.3 General principles

Consent should be obtained and documented in any combination of the following formats before a fetal anomaly ultrasound examination is arranged.

- In the woman's hand held notes (SWHMR)
- Ultrasound clinical information storage system
- Data entry on the hospital clinical information system
- Ultrasound request/report form

There should be information available about the range of abnormalities (with examples) that might be suspected or identified. This information should be available at the time the appointment is made but also at the time of the ultrasound examination appointment itself. Before each fetal anomaly examination, it is good practise for the sonographer to explain the purpose of the examination and obtain further verbal consent. Women who request an ultrasound examination at 18+0 – 20+6 weeks of pregnancy, but who express a wish not to be informed if abnormalities are found, should be advised that all the significant findings seen on the examination will be reported and that therefore they should consider not having ultrasound screening.

It is also good practice to ensure that women have the opportunity to view the image on a monitor throughout the examination if they wish to, whether or not a fetal anomaly is suspected.

Women with a multiple pregnancy must receive appropriate information prior to being screened; this information must include the implications and limitations of the ultrasound examination.

5.4 Organisation

The fetal anomaly screening examination must be performed in a room specifically designed for that purpose. A low risk singleton fetal anomaly screening appointment ultrasound examination should be no less than 30 minutes (45 minutes if a multiple pregnancy) which should include 'on and off the couch time' as well as time to complete the report. All women should have the opportunity to have one other person present during the fetal anomaly screening ultrasound examination; beyond this, the number of individuals allowed at the examination should be in accordance with the local policy.

All ultrasound departments must have an agreed written policy which adheres to national standards and defines the purpose of antenatal screening using ultrasound examinations. Assessment techniques and biometric charts used for fetal measurements must meet nationally

agreed standards with clear policies and procedures in place for the communication of normal and abnormal ultrasound examination results.

Ultrasound imaging machines used for antenatal screening must adhere to National and European standards for their specifications, maintenance schedules and upgrading.

The ultrasound examination requests should include all relevant medical and social issues, which may affect the risk of abnormality, and be made available to the person performing the ultrasound examination.

Ideally a structured electronic report should be generated and stored on a local clinical information system. Whatever system is used, a copy of the report should be made available for the woman's maternity hand held notes and a further copy kept either electronically or where no such system exists, on paper in the hospital notes.

Any health professional carrying out ultrasound screening for fetal anomaly must have a postgraduate certificate of competency to practise obstetric ultrasound or equivalent. An average of at least one ultrasound examination session per week, that includes screening for or diagnosis of fetal anomaly is considered the minimum required to be involved in screening. There should also be ongoing accreditation processes in place such as a CPD programme or DQASS.

5.5 Fetal Anatomy – low risk screening ultrasound examination

Each ultrasound service should adopt the 18^{+0} to 20^{+6} week fetal anomaly screening ultrasound base menu as recommended by the NSC Fetal Anomaly Screening Programme (FASP) when performing a low risk fetal anomaly screening ultrasound examination. As a minimum, the following anatomical structures as detailed in Table 1[15] below are the least that should be checked for the anomaly screen to be considered completed.

[15] FASP (2009) Base Screening Ultrasound Menu During Pregnancy, National Screening Committee, May 2009

Table 1: 18^{+0} - 20^{+6} Fetal Anomaly Screening Base Menu[16]

No.	Area	Structure	View
1.	Head and neck	Skull Neck: *-Skin fold (NF)* Brain: *-Cavum septum pellucidum* *-Ventricular atrium* *-Cerebellum*	Shape Subjective - measure NF if looks increased
2.	Face	Lips	Coronal view
3.	Chest	Heart: *-Four-chamber view* *-Outflow tracts* Lungs	Refer to Fetal Cardiac Protocol (Appendix 8)
4.	Abdomen	Stomach: *-Stomach and short intra-hepatic section of umbilical vein* Abdominal wall Bowel Renal pelvis Bladder	Transverse, sagittal Transverse Transverse - measure AP if looks increased Sagittal and transverse
5.	Spine	Vertebrae Skin covering	Sagittal and transverse Sagittal and transverse
6	Limbs (a) Limbs (b)	Femur Hands: *-Metacarpals (right and left)* Feet: *-Metatarsals (right and left*)	Length (one leg only) Visible (not counted) Visible (not counted)
7.	Uterine cavity	Amniotic fluid Placenta*	Subjective volume Visible and position noted

Whilst it is not national policy to screen for placenta praevia, placental site may be useful for future management and it is therefore best practice to note placental site.

[16] Second-trimester ultrasound to detect fetuses with Down syndrome: a meta-analysis. Smith-Bindman R, Hosmer W, Feldstein VA, Deeks JJ, Goldberg JD. JAMA. 2001 Feb 28; 285(8):1044-55.

Fetal Anomaly Ultrasound Screening Programme Study: Literature Survey June 2007 Lyn Bryant, Adam Fisher & Fiona Vicente Social Research & Regeneration Unit A University of Plymouth Centre

Ultrasound screening for fetal abnormalities. Report of the working party. Royal College of Obstetricians and Gynaecologists 2000.

Six specific fetal anatomical sections should be identified at examination. An electronic image and report should be stored and archived fulfilling medico-legal requirements of storage.

- Head Circumference (HC) demonstrating HC measurement and measurement of the atrium of the lateral ventricle
- Suboccipito-bregmatic demonstrating measurement of the transcerebellar diameter
- Coronal view of lips with nasal tip
- Abdominal Circumference (AC) demonstrating AC measurement
- Femur Length (FL) demonstrating (FL) measurement
- Sagital view of spine including sacrum and skin covering

A Down's syndrome risk generated by a nationally accepted screening method **should not be recalculated, up or down**, due to the presence or absence of an ultrasound marker of less predictive power than an increased nuchal fold[17].

Women who are found to be **low risk** through testing in either the first or second trimesters or have declined screening for Down's syndrome**,** should **not be referred for further assessment of chromosomal abnormality** if normal variants such as the examples below (whether single or multiple) are seen at the 18^{+0} to 20^{+6} weeks' fetal anomaly screening ultrasound examination[18]. It is **encouraged that the term ultrasound "soft marker" is no longer used.[19]**

1. Choroid plexus cyst
2. Dilated cisterna magna
3. Echogenic foci in the heart
4. Two-vessel cord

However, the appearances listed below (previously classified as "markers") are examples of findings which **should be reported and the woman referred for further assessment** by doctors and midwives with particular and up to date experience in the management of fetal abnormality i.e. treated as for any other suspected fetal anomaly:

1. Nuchal fold greater than 6mm
2. Ventriculomegaly (atrium greater than 10mm)
3. Echogenic bowel (with density equivalent to bone)
4. Renal pelvic dilatation (greater than 7 mm)

All women should be offered a further ultrasound examination (up to 23 weeks of pregnancy) to complete the screening examination if the image quality of the first examination is compromised by one or more of the following:

- Increased maternal body mass index (BMI)
- Uterine fibroids
- Abdominal scarring
- Sub-optimal fetal position

[17] UK NSC FASP Programme Statement: Recalculation of Down's syndrome risk following ultrasound examination at the mid-trimester ultrasound scan
[18] Women who have not had Down's screening (booked to late or somewhere in which this programme was not available) should have counselling based on maternal age and/or family history, not on whether normal variants are found during scanning
[19] RCOG (2000) '*Routine Ultrasound Screening in Pregnancy: Protocol, Standards and Training*'

Any subsequent attempts at screening may be, at the sonographers' discretion, later the same day or at a later date. Where an adequate assessment of the fetal anatomy remains compromised after the second ultrasound examination, the woman should be told that the screening is incomplete and documented in all formats.

The local policy regarding fetal sexing should be made available before the ultrasound examination appointment is made and supported by information available at the time of the ultrasound examination. If offered it should include information about success rates from published and local figures.

5.6 Informing women about ultrasound findings

Sonographers should be encouraged to communicate the results of the ultrasound examination verbally with the woman at the time the ultrasound examination is being performed, whether or not a fetal anomaly is suspected.

Women should have the opportunity to discuss the results of their ultrasound examination with or without their partner present.

5.7 The diagnostic process

If a fetal anomaly is suspected during an anomaly screening ultrasound examination then the woman should be informed at the time of the ultrasound examination or shortly afterwards. Some indication of the nature of the suspected problem should ideally be outlined by a sonographer who performed the examination. This should occur in privacy and ideally, if the woman wishes, in the company of a partner, friend or relative.

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It is good practice for the sonographer to seek a second opinion. This may be either by another sonographer or other professional with expertise in fetal anomaly ultrasound who may repeat the ultrasound examination or review images taken at the time of the initial ultrasound examination.

It is the responsibility of the Health board to have a clear diagnostic pathway for the ongoing referral of women in whom a fetal anomaly has been suspected. If a cardiac abnormality is suspected (unless associated with more serious anomalies) there should be an agreed pathway for interaction with paediatric cardiology services and a discussion initiated concerning timing and location of delivery.

The woman should receive further counselling by a health professional with particular and up to date experience in the management of fetal anomaly within 1 working day of the fetal anomaly being first suspected. This may be a midwife, sonographer, an obstetrician or radiologist and should normally be in person although on some occasions may be by telephone or other remote means.

An appointment to discuss the implications of the suspicion of a fetal anomaly and further ultrasound examinations intending to make a diagnosis should ideally be offered within 2 working days, and no later than 5 working days. This appointment should involve referral to a multidisciplinary team including medical staff and midwifery staff with current expertise in the care of women with fetal anomaly which may be available locally or involve referral to another centre.

The woman must be given an explanation about why the referral is necessary.

If the presence of a fetal anomaly is confirmed there should be the opportunity for referral to other specialist services such as clinical genetics, neonatology, paediatric cardiology and paediatric surgery where necessary. The diagnostic certainty, implications and choices should be discussed and further appointments or referral considered.

Women in whom fetal anomaly is either suspected or confirmed, should be given the opportunity to receive written information about the anomalies; including information about the degree of certainty of diagnosis and be given time to reflect on the implications and further diagnostic options.

In cases where termination of pregnancy might be considered, this should be entirely the choice of the woman. The option to continue a pregnancy even in the face of confirmed lethal or severe fetal anomaly should be available. Conversely, for conditions that satisfy the requirements of Section E of the 1967 Abortion Act (as amended in 1990) and where women request this option, the provision of termination of pregnancy with or without feticide, should be available either locally or by referral to another unit.

5.8 Audit, Quality Control and Performance indicators

There should be a local audit of the effectiveness of the screening of fetal anomalies by ultrasound. Data should be available for regional and national audit subject to the Board's policy on confidentiality. These data should include, for example: the number of low risk women who undergo fetal anomaly screening; the number in whom a fetal anomaly is suspected; the number in whom a prenatal diagnosis was made; the outcome of that pregnancy with an assessment of the accuracy of the prenatal diagnosis and the number of false positives, where a fetal anomaly was either suspected or diagnosed but where no anomaly was present at delivery. The gestation at which the anomaly was first suspected and the process by which the investigation was performed should also be recorded: whether through booking ultrasound; as a result of serum screening; following a low risk anomaly screening ultrasound or at other opportunity. It is intended that data will form part of a Scottish national congenital anomaly audit or register.

In addition in order to be able to benchmark nationally against the NHS FASP standards, on an annual basis each Unit should provide (as a minimum) the screen positive rate (SPR) and detection rates (DR) for the following 11 conditions.

Conditions	Recommended Detection rate (%)
Anencephaly	98
Open spina bifida	90
Cleft lip	75
Diaphragmatic hernia	60
Gastroschisis	98
Exomphalos	80
Serious Cardiac abnormalities[20]	50
Bilateral renal agenesis	84
Lethal skeletal dysplasia	60
Edward's syndrome (Trisomy 18)	95
Patau's syndrome (Trisomy 13)	95

It is hoped that the development of a Scottish national congenital anomaly audit will provide an evidence base for the usefulness of these conditions and highlight other parameters that may provide useful as performance indicators

20

ICD10 code(s) used
Q20.0 Common arterial trunk
Persistent truncus arteriosus
Q20.3 Discordant ventriculoarterial connection
Dextrotransposition of aorta
Transposition of great vessels (complete)
Q21.3 Tetralogy of Fallot
Q22.5 Ebstein's anomaly
Q23.4 Hypoplastic left heart syndrome
Q25.1 Coarctation of aorta

5.9 Training and Education

All professionals involved in the provision and delivery of pregnancy and newborn screening should undergo education to ensure that consistent, up-to-date information is being given to women as they make decisions along the screening pathway. All new staff involved in screening should work through an appropriate induction programme.

Any health professional carrying out fetal anomaly ultrasound examination for the purpose of screening for fetal anomaly should hold, as a minimum, one of the following:

a. Certificate/Diploma (as appropriate) in Medical Ultrasound (CMU/DMU) of the Society and College of Radiographers (SCoR) with evidence of appropriate continuous professional development (CPD).
b. Post Graduate Certificate in Medical Ultrasound (PGCert.MU) approved and validated by a Higher Institute of Education (HIE) and accredited by the 'Consortium for Sonographic Education' (CASE) with evidence of appropriate CPD. The qualification should be relevant to obstetric ultrasound practice.
c. Royal College of Obstetricians and Gynaecologists (RCOG), Royal College of Radiologists (RCR) Diploma in Obstetric Ultrasound, RCOG Intermediate Ultrasound of Normal Fetal Anatomy Training Programme (Module 3) and Advanced Training Speciality Module (ATSM) in Fetal Medicine. Evidence of appropriate CPD should also be provided[*].
d. Sonographers, who do not have a UK recognised ultrasound qualification i.e. those trained overseas, should be registered under the Voluntary Register of Sonographers[#].
e. Written evidence or certification for obstetricians or radiologists detailing previous obstetric ultrasound training and experience in this or another country.

Diagnostic ultrasound procedures should only be carried out by staff fully trained in image acquisition and interpretation, the use of the equipment and in the safe use of ultrasound.

Regular appraisals should be carried out in order to set objectives and assess performance.

A programme of education and training for practitioners should incorporate aspects of: breaking bad news, audit and report writing, clinical governance and facilitation of learning events.

Boards should encourage a multidisciplinary approach between all health professionals providing pregnancy screening in the provision of local education and training programmes. Provision of education and training for interpreters and advocacy workers should be included.

5.10 Equipment

A formal review of equipment should be between four to six years following installation. High priority must be given to the replacement of equipment which relies on obsolete or redundant technology, where this compromises image quality.

The equipment should be capable of producing images of diagnostic quality. An image archiving system is recommended as this will allow rapid recall and review of images by those responsible for pregnancy care and which fulfills the medico-legal requirements e.g. PACS.

[*] It is recognised that many senior experienced scanning professionals (obstetricians, radiologists and sonographers) may not have a formal qualification and it is not intended to prevent these very important providers of the service to be excluded from practicing. These individuals should continue scanning under a 'grandfather clause' whilst applying the above qualification parameters to new practitioners to new practitioners to fetal anomaly scanning.
[#] The database is kept and controlled by the Society and College of Radiographers (SCoR) in association with the UK Association OF Sonographers (UKAS).

Equipment size should take account of prospective location, available space, and the need for portability.

Display size should be sufficient for clear visualisation. Equipment should be able to reproduce accurate measurements.

Machine functions should include:

a. Magnification facility
b. Cineloop function
c. Calipers that have a precision to one decimal point, i.e. 0.1 mm
d. Adjustable signal processing facilities
e. Tissue specific pre-sets for individual clinical applications

In order to obtain the best image quality, sonographers and clinicians must select the most appropriate probe (curvi-linear or vaginal probe) and frequency setting relevant to gestational age.

5.11 Ultrasound Safety

Each ultrasound screening department should ensure that a health and safety risk assessment is carried out at least once a year.

Ultrasound departments must ensure that quality control procedures are carried out on an annual basis ideally using a tissue mimicking phantom. All ultrasound equipment must be cleaned in guidance with local policy to minimise risk of infection with particular attention given to the machine keyboard, hand held probes and flexes.

At least annual servicing and maintenance should be undertaken. Any cracks in the probe should be reported immediately. Electric safety testing is also required annually by qualified personnel.

To avoid the use of excessive and inappropriate exposure levels, each practitioner must use output (transmit) power control settings set at the minimum possible to achieve diagnostic images. Machines should be set up so that the default (switch on) setting of the acoustic output power control is low. A low setting should be set for each new examination. Output power should only be increased during the examination when this is necessary.

The probe should not be held in a fixed position over the fetus for any longer than is necessary and should be removed from the woman whenever there is no need for real-time imaging. The freeze-frame and cine loop facilities should be used, with the probe removed from the woman's abdomen when considering or discussing an image feature.

Women should not have prolonged unnecessary ultrasound investigations. Exposure time must be kept to a minimum. Attention should be paid to check for probe overheating in intra-cavity probes and should it be suspected, the probe not used until checked by the manufacturer or other suitably qualified person.

Sensitive tissues such as an embryo less than 8 weeks after conception, or the head brain or spine of the fetus, must be treated with particular care and efforts made to reduce potential thermal effects when exposed to diagnostic ultrasound.

Detailed BMUS safety statements are available at www.bmus.org

6. Evaluation of the Down's syndrome and Fetal Anomaly Screening programme

Audit and monitoring of the screening programme should be performance managed at all health service levels (national and local).

All screening programmes are expected to have the appropriate tools to support the minimum criteria for the audit process. This must include clerical support, information technology (IT) equipment/software and networks that link with appropriate data collection systems within the Board.

All abnormal findings subsequently proved to be normal should be kept on a database for the purposes of quality control; confirmed fetal abnormalities should be recorded on/in the:

- Board's clinical information system
- Woman's maternity hand held notes
- Woman's hospital notes
- Fetal anomaly database e.g. Viewpoint/ CRIS

A high standard of cytogenetic and perinatal pathology with feedback to the ultrasound and pregnancy laboratory departments are an essential element for a screening service.

6.1 Quality control

Laboratory and ultrasound services must be able to provide (as a minimum) from the proportion of the pregnant population that had screening,

- Detection rate (DR)[21]:
- Screen positive rate (SPR)

Each Board's ultrasound service must regularly submit fetal biometry data (including NT measurements) to the National Down's Syndrome Quality Assurance Screening Service (DQASS), so that performance activity can be regularly audited and monitored. This is normally undertaken by the laboratories from the laboratory database.

All sonographers/clinicians performing nuchal translucency measurements must have their results subjected to rigorous, valid audit and to external evaluation. Ultrasound services must be able to monitor the reproducibility of fetal biometric measurements and be able to provide data of ultrasound activity. All ultrasound examinations should be documented and archived and hard copy of all abnormal findings made.

Each Board should audit women's views and experiences following ultrasound examinations as part of a screening programme.

All ultrasound images should be carefully archived. In respect of abnormal ultrasound examination findings, consideration should be given to store them as a thermal image, video, DVD or in the Boards Picture Archiving Communication System (PACS).

All ultrasound screening reports and images must be captured, stored and archived for the purposes of a complete maternal record and to fulfil medico-legal requirements.

[21] This is the number of observed 'suspected' anomalies which are later confirmed either by karyotyping, following termination of pregnancy or in the postnatal period.

7. Adverse Incidents

As with any screening programme, there is potential for significant adverse incidents. All adverse incidents should be managed appropriately to minimise the risks to, and effects on the woman and participating Boards.

An adverse incident can be any of the following:

Administrative

- Failsafe procedures not instigated
- Woman/requesting midwife/ GP not notified of result

Laboratory

- Assay errors
- Interpretation errors

Clinical

- Misdiagnosis
- Long waiting times through the pathway from positive screening test to confirmed diagnosis

7.1 Procedure

Any healthcare professional involved in the NHS Scotland pregnancy screening programme who becomes aware of a suspected problem should follow agreed local Board clinical governance procedures.

Local clinical governance procedures may vary from one Board to another but commonly involve an initial period of local investigation and establishment of extent of problem followed by external independent peer review when appropriate.

In all cases associated with the screening programme, there will be a thorough investigation and National Services Division (NSD) will be notified early in the process – at the time of internal investigation. In view of the sensitivities of national screening programmes and the public interest in them, NSD may require an external peer review even if local Board management decide not to invoke this.

If necessary NSD and the Board will meet to discuss and agree what action, if any, is required.

NSD will notify the Scottish Government Health Directorates (SGHD) and decide if action is needed in other NHS Board areas.

Note:
These protocols are to be used in addition to, and do not replace, the Boards' Clinical / Adverse Incident Reporting Procedures.

8. Confidentiality

Professional staff involved in the screening programme will comply with the provisions of the Caldicott Report. In particular, patient-identifiable information will only be used in clearly defined and monitored circumstances, only when absolutely necessary and should entail the use of the minimum necessary patient-identifiable information.

Access to patient identifiable information will be on a strict need to know basis, everyone in the organisation will be aware of their responsibilities with respect to patient confidentiality and the organisation will ensure that its use of patient-identifiable information is lawful.

National Services Scotland (ISD and NSD) does not require aggregated information returns on the performance of the screening programme to include patient-identifiable information; information on clinical activity for national data sets and monitoring must be submitted in anonymised format.

Appendix 1 - Specification for Diagnostic Ultrasound Equipment for NT / Fetal Anomaly Screening

Clinical Requirements
Suppliers - The equipment provided must be capable of allowing the user to perform Nuchal translucency measurements.

Users/Purchasers – The checked boxes indicate the minimum required tasks for an ultrasound scanner for NT / Fetal Anomaly Screening. Please add any additional requirements as appropriate to your department.

Clinical Task	1st Trimester	2nd Trimester
Fetal Measurement	✓	✓
Visualisation of Fetal Anatomy	✓	✓
OTHER (please describe)		

Technical Requirements

Feature	
Physical and Ergonomic Features The system offered should meet the recommendations outlined in the Society and College of Radiographers Publication "Prevention of Work Related Musculoskeletal Disorders in Sonography", SoR, 2007.	
*Room based wheeled unit	✓
Screen with ultrasound image area f.o.v. 12cm x 15 cm with a matrix of 512 x 512 and effective pixel size of no less than 500. See Royal college of Radiologist specification guidelines.	✓
Slave Monitor	✓
Flexible Position Monitor (height and angle)	✓
Flexible position control console (height and angle)	✓
Subdued console lighting	✓
OTHER (please describe)	

Feature	
Scan Modes	
B-Mode	✓
Tissue Harmonic Imaging	✓
Spatial Image Compounding	
M-Mode	
Colour & Power Doppler	✓
Spectral Doppler	✓
3-D Imaging	
Real-Time 3-D	
Split Screen Imaging	✓
OTHER (please describe)	

Feature	
Transducers – sufficient transducers should be supplied to meet the clinical requirements outlined in section 1	
Convex	✓
Trans Vaginal	✓
OTHER (please describe)	

Feature	
Measurement / Calculation Tables – these should be sufficient to meet the clinical requirements outlined in section 1	
Multiple shaped callipers of minimum precision 0.1 mm with continuous motion	✓
Callipers of dynamically varying contrast compared to background.	✓
Small sized callipers for measurements < 5mm	✓
Ellipse Circumference / Area Measurement	✓
Freehand Circumference / Area Measurement	
Measurement on real time (non-frozen) images	
Measurement on frozen images	✓
Off-line measurement (on saved images)	✓
Obstetric Calculation and measurement Package	✓
User definable tables	✓
Fetal Cardiac Measurement Package	
Fetal vascular Measurement Package	✓
OTHER (please describe)	

Feature	
Controls and Other Features	
Freeze image facility Read and Write Zoom	✓
Read and Write Scroll	✓
Cine-frame review	✓
Footswitch control of freeze / zoom / store / print* *Delete as applicable	✓
2.5.5 Where air filters are fitted to the equipment these must be easily removable for cleaning and the frequency and method of cleaning specified	✓
Other (please describe)	

Feature	
Image Storage and Output Options	
Network port with a minimum capability if 100 MBits /s	✓
Large capacity on-board image management system (storage > 40Gb)	✓
***USB / CD / DVD *Delete as applicable**	✓
Thermal Paper Printer	✓
Confirmed Dicom compatibility for Print, Store, Work list, Retrieve, Display, and Presentation.	✓
OTHER (please describe) **TEST pattern, such as SMPTE as part of the spec** **Include a DVD player so that images can be played back at the bedside.**	

General Requirements

Environmental / Room Conditions
The Machine must have the ability to be safely used in rooms and to be safely stored in areas with a minimum temperature of 21ºC ± 1ºC.

Safety and standards
All systems (including any peripheral/auxiliary equipment supplied) must be CE Medical marked and comply with current European and UK specifications for medical equipment including IEC60601-1-1, IEC60601-1-2 and IEC60601-2-37.

Acoustic power outputs must meet national and international standards set down by AIUM/NEMA ("Acoustic output Measurement Standard for diagnostic ultrasound equipment"). NEMA Standards Publication UD2-2004. Published by National Equipment Manufacturer's Association, 1300 North 17th Street, Suite 1847, Rosslyn, Virginia 22209-3806 USA. (www.nema.org)

Manufacturer and user defined system presets must include an option to default to low acoustic output power in all modes.

Biological safety – Details must be given of the recommended methods of cleaning and, where appropriate, sterilisation of all transducers and other parts of the system. A detailed protocol and list of approved cleaning materials should be provided.

Training, Documentation and Support
A full set of operators' manuals for the system and all ancillary equipment must be provided. Additional "quick guide" booklets or cards should also be provided.

The manufacturers must provide adequate training and support to ensure that all primary users of the equipment are familiarised with all aspects of the system operation within a reasonable period of time following installation. The proposal for meeting this requirement should be outlined.

The numbers and base locations of applications staff available to the users should be provided. Guaranteed post-sales user support is essential.

Warranty and Maintenance
A statement of the warranty period for the equipment and specifically for the probes should be included with the supplier response. Extended warranty options should be quoted to include first-line in-house service and maintenance by hospital staff (including training as described above) if available.

Full details of each maintenance contract should be supplied including response time, hours of availability and numbers and availability of appropriately trained engineering staff. Also the options for transducer cover (accidental and wear-and-tear) and software upgrades should be given.

Dr Tony Evans et al
The Machine Specification Project
NHS Fetal Anomaly Screening Programme

Appendix 2 – Crown Rump Length (CRL) and Head Circumference (HC) dating tables.

Crown Rump Length (CRL) dating table

GA (days) = 8.052√ (CRL X 1.037) + 23.73

CRL (mm)	GA (wks + days) 50th centile	5th centile	95th centile
43	11+0	10+3	11+5
44	11+1	10+3	11+6
45	11+2	10+4	11+6
46	11+2	10+5	12+0
47	11+3	10+5	12+1
48	11+4	10+6	12+1
49	11+4	10+6	12+2
50	11+5	11+0	12+2
51	11+5	11+1	12+3
52	11+6	11+1	12+4
53	11+6	11+2	12+4
54	12+0	11+2	12+5
55	12+1	11+3	12+5
56	12+1	11+3	12+6
57	12+2	11+4	12+6
58	12+2	11+4	13+0
59	12+3	11+5	13+0
60	12+3	11+6	13+1
61	12+4	11+6	13+1
62	12+4	12+0	13+2
63	12+5	12+0	13+3
64	12+5	12+1	13+3
65	12+6	12+1	13+4
66	12+6	12+2	13+4
67	13+0	12+2	13+5
68	13+0	12+3	13+5
69	13+1	12+3	13+6
70	13+1	12+4	13+6
71	13+2	12+4	14+0
72	13+2	12+5	14+0
73	13+3	12+5	14+0
74	13+3	12+6	14+1
75	13+4	12+6	14+1
76	13+4	13+0	14+2
77	13+5	13+0	14+2
78	13+5	13+0	14+3
79	13+6	13+1	14+3
80	13+6	13+1	14+4
81	14+0	13+1	14+4
82	14+0	13+2	14+5
83	14+0	13+3	14+5
84	14+1	13+4	14+6

Head Circumference (HC) dating table *

Log_e (GA weeks) = 0.010611 X HC – 0.000030321 X HC^2 + 0.43498 X 10^{-7} X HC^3 + 1.848

HC (mm)	Gestation (wks+days)
80	12+3
81	12+4
82	12+5
83	12+5
84	12+6
85	12+6
86	13+0
87	13+0
88	13+1
89	13+2
90	13+2
91	13+3
92	13+3
93	13+4
94	13+5
95	13+5
96	13+6
97	13+6
98	14+0
99	14+0
100	14+1
101	14+2
102	14+2
103	14+3
104	14+3
105	14+4
106	14+4
107	14+5
108	14+6
109	14+6
110	15+0
111	15+0
112	15+1
113	15+2
114	15+2

HC (mm)	Gestation (wks+days)
115	15+3
116	15+3
117	15+4
118	15+4
119	15+5
120	15+6
121	15+6
122	16+0
123	16+0
124	16+1
125	16+1
126	16+2
127	16+3
128	16+3
129	16+4
130	16+4
131	16+5
132	16+5
133	16+6
134	17+0
135	17+0
136	17+1
137	17+1
138	17+2
139	17+2
140	17+3
141	17+4
142	17+4
143	17+5
144	17+5
145	17+6
146	17+6
147	18+0
148	18+1
149	18+1

HC (mm)	Gestation (wks+days)
150	18+2
151	18+2
152	18+3
153	18+3
154	18+4
155	18+5
156	18+5
157	18+6
158	18+6
159	19+0
160	19+0
161	19+1
162	19+2
163	19+2
164	19+3
165	19+3
166	19+4
167	19+4
168	19+5
169	19+6
170	19+6
171	20+0
172	20+0
173	20+1
174	20+1
175	20+2
176	20+3
177	20+3
178	20+4
179	20+4
180	20+5
181	20+5
182	20+6
183	20+6

*Loughna P, Chitty L, Evans T, Chudleigh T (2009). Fetal size and dating: charts recommended for clinical obstetric practice. Ultrasound, 17: 161-167

- The Nuchal Translucency (NT) measurement should be made when the Crown Rump Length (CRL) is between 45mm and 84mm.
- Using the current BMUS 2009 dating formula, the gestational age range for NT measurement is from 11 weeks + 2 days to 14 weeks + 1 day.

- From 14 weeks + 2 days to 20 weeks + 0 days, the quadruple test should be offered.

- Where both dating and Down's syndrome screening are requested, and the CRL is between 45.0 and 84.0mm the pregnancy should be dated by CRL and combined screening performed.

- Where both dating and Down's syndrome screening are requested and the CRL is ≥ 84.1mm, the pregnancy should be dated by HC.

- If the HC is ≥101.0mm and the gestational age is ≥14 weeks + 2 days, date by HC. The CRL should be ignored as it is ›84.0mm. Quadruple screening should be offered.

- If the HC is ‹101 mm and the CRL is ›84mm, date by HC. If the gestational age as calculated from the HC is ≤ 14 weeks + 1 day, the woman should be informed that the NT risk cannot be calculated from a CRL ›84mm, even though the gestational age of her pregnancy as estimated by the HC, lies within the gestational age window for combined screening. Combined screening is not an option but quadruple screening can be offered from 14 weeks + 2 days gestation.

Appendix 3 - Recommended criteria for measurement of fetal crown rump length (CRL) as part of combined screening for Trisomy 21

NHS Scotland endorses the criteria for measurement of fetal crown rump length (CRL) as part of combined screening for Trisomy 21 as advocated by the NSC Fetal Anomaly Screening Programme.

Criteria for CRL	Detail	References
CRL range in mm	45–84mm	FMF 11-13 weeks scan online (2010)[22]
Image magnification	The magnification of the fetus is made as large as possible before the image is frozen, to clearly demonstrate the entire crown rump length	NHS FASP Condensed Education Module for Trisomy 21 resource, and the NHS FASP NT resource[23]
Fetal position	A midline sagittal section of the whole fetus should be obtained, ideally with the fetus horizontal on the screen so that the line between crown or rump is at 90° to the ultrasound beam	Fetal size and dating: charts recommended for clinical obstetric practice (2009)[24]
Fetal attitude	Neutral position with fluid visible between fetal chin and chest. Neither hyper-extended nor flexed	NHS FASP Condensed Education Module for Trisomy 21 resource, and the NHS FASP NT resource2
Three measurements	The best of three measurements should be taken3. Record/archive the image that meets all the criteria	Fetal size and dating: charts recommended for clinical obstetric practice (2009)3
Linear CRL measurement	Linear callipers should be used to measure the maximum un-flexed length, in which the end points of crown and rump are clearly defined	Fetal size and dating: charts recommended for clinical obstetric practice (2009)3
Calliper placement	Intersection of callipers (+) placed on the outer margin of skin borders of the fetal crown and rump	NHS FASP Condensed Education Module for Trisomy 21 resource, and the NHS FASP NT resource2

[22] http://www.fetalmedicine.com/fmf/online-education/01-11-136-week-scan/
[23] NHS FASP Condensed Education Module for Trisomy 21 resource, and the NHS FASP NT resource www.fetalanomaly.screening.nhs.uk
[24] Loughna P, Chitty L, Evans T, Chudleigh T (2009) *Ultrasound Fetal Size and Dating: Charts Recommended for Clinical Obstetric Practice,* Volume 17:3. Pub: British Medical Ultrasound Society

Appendix 4 - Considerations relating to the biochemical component of the combined screening test for Down's syndrome in the first trimester

NHS Scotland endorses the programme statement issued by the NSC Fetal Anomaly Screening Programme regarding vaginal bleeding and vanishing twin.

There are two different clinical events that have raised questions about the reliability of the biochemical component of the first trimester combined screening test for Down's syndrome[25]:

1. **Maternal vaginal bleeding**

If there is a history of significant maternal vaginal bleeding at the time of the first trimester screening for Down's syndrome, there have been concerns that this might change maternal blood levels of the biochemical markers used in the combined test, perhaps secondary to placental disruption. However we suggest that services continue to use the combined test in the normal way (calculating the risk based on maternal age, nuchal translucency, fb-HCG and PAPP-A levels), because current data[26] suggest that the biochemical marker levels are not significantly different in women with this history.

2. **'Vanished' twin**

When ultrasound shows that there is an empty second pregnancy sac, the biochemical markers appear no different to those in a singleton pregnancy and the combined test of NT, PAPP-A and free beta HCG can be used to calculate the risk. If ultrasound shows that there is a second sac containing a fetal pole with no heart activity (sometimes called 'vanished' twin), it is possible that there could be a contribution to the maternal biochemical markers for many weeks[27] [28]. We recommend that in this event services undertake the risk calculation based on the maternal age and nuchal translucency only (i.e. without biochemistry).

[25] NHS FASP 2008. NHS Fetal Anomaly Screening Programme – Screening for Down's syndrome: UK NSC Policy recommendations 2007–2010.

[26] Spencer K, Spencer CE, Stamatopoulou A, Staboulidou I, Nicolaides KH. Early vaginal bleeding has no impact on biochemical markers in first trimester aneuploidy screening. *Prenatal Diagnosis*, in press.

[27] Gjerris AC, et al. The effect of a 'vanishing twin' on biochemical and ultrasound first trimester screening markers for Down's syndrome in pregnancies conceived by assisted reproductive technology. *Hum Reprod*. 2009 Jan, 24(1): 55–62.

[28] K Spencer, I Staboulidou, K H Nicolaides (2010) First trimester aneuploidy screening in the presence of a vanishing twin: implications for maternal serum markers. Prenat Diagn Jan. DOI: 10.1002/pd.2445, Published Online: 11 Jan 2010

Appendix 5 - Recommended criteria for measurement of fetal nuchal translucency (NT) as part of combined screening for Trisomy 21

NHS Scotland endorses the recommended criteria for measurement of fetal NT as part of combined T21 screening as advocated by the NSC Fetal Anomaly Screening Programme.

Criteria for NT	Detail	References
Image magnification	1. Image magnification should be such that the fetal head and upper throrax occupy the whole screen	FMF 11–13 weeks scan online resource (2010)
Fetal position	1. A midline sagittal section of the fetus should be obtained 2. The fetus should be horizontal on the screen, either supine or prone*	1 FMF 11–13 weeks scan online resource (2010) 2 Down's Syndrome Screening Programme standards (2004)
Distinguishing between fetal skin and amnion	1. Care must be taken to distinguish between fetal skin and amnion	FMF 11–13 weeks scan online resource (2010)
Fetal attitude	1. The fetus should be in a neutral position, with the head in line with the spine	FMF 11–13 weeks scan online resource (2010)
Maximum nuchal thickness	1. The widest part of the translucency must always be measured 2. The umbilical cord may be around the fetal neck in about 5% of cases. In such cases it is more appropriate to use the average of the two measurements	1,2 FMF 11–13 weeks scan online resource (2010)
Calliper placement	1. Measurements should be taken with the inner border of the horizontal line of the callipers placed ON the line that defines the NT thickness. The crossbar of the calliper should be such that it is hardly visible as it merges with the white line of the border, not in the nuchal fluid 2. In magnifying the image (pre- or post-freeze zoom) it is important to turn the gain down 3. During the scan more than one measurement must be taken and the maximum one that meets all the criteria should be recorded 4. This image must be retained in the patient record	1,2,3 FMF 11–13 weeks scan online resource (2010) 4 Down's Syndrome Screening Programme standards (2004)

**This differs from FMF criteria as NHS FASP does not recommend screening for nasal bone absence or hypoplasia, thus allowing measurement of the NT with the fetus in the prone position.*

References

1. 11–13 weeks scan online resource (2010) www.fetalmedicine.com/fmf/online-education/01-11-136-week-scan/
2. The National Screening Committee: National Down's Syndrome Screening Programme (DoSySP) (2004), Standards for fetal nuchal translucency (NT) measurement for Down's syndrome Screening

Appendix 6 – Fetal Anomaly Base Menu

NHS Scotland endorses the fetal anomaly base menu as advocated by the NSC Fetal Anomaly Screening Programme.

Fetal Anomaly Base Menu[29]

No.	Area	Structure	View
1.	Head and neck	Skull Neck: *-Skin fold (NF)* Brain: *-Cavum septum pellucidum* *-Ventricular atrium* *-Cerebellum*	Shape Subjective - measure NF if looks increased
2.	Face	Lips	Coronal view
3.	Chest	Heart: *-Four-chamber view* *-Outflow tracts* Lungs	Refer to Fetal Cardiac Protocol (Appendix 9)
4.	Abdomen	Stomach: *-Stomach and short intra-hepatic section of umbilical vein* Abdominal wall Bowel Renal pelvis Bladder	Transverse, sagittal Transverse Transverse - measure AP if looks increased Sagittal and transverse
5.	Spine	Vertebrae Skin covering	Sagittal and transverse Sagittal and transverse
6	Limbs (a) Limbs (b)	Femur Hands: *-Metacarpals (right and left)* Feet: *-Metatarsals (right and left*)	Length (one leg only) Visible (not counted) Visible (not counted)
7.	Uterine cavity	Amniotic fluid Placenta	Subjective volume Visible and position noted

[29] Second-trimester ultrasound to detect fetuses with Down syndrome: a meta-analysis. Smith-Bindman R, Hosmer W, Feldstein VA, Deeks JJ, Goldberg JD. JAMA. 2001 Feb 28; 285(8):1044-55.

Fetal Anomaly Ultrasound Screening Programme Study: Literature Survey June 2007 Lyn Bryant, Adam Fisher & Fiona Vicente Social Research & Regeneration Unit A University of Plymouth Centre

Ultrasound screening for fetal abnormalities. Report of the working party. Royal College of Obstetricians and Gynaecologists 2000.

Appendix 7 – Fetal Cardiac Protocol

NHS Scotland endorses the fetal cardiac protocol as advocated by the NSC Fetal Anomaly Screening Programme.

Definition of major congenital heart Disease (CHD)
CHD is a condition that will require immediate cardiac assessment and/or treatment within the first year of a child's life.

Fetal echocardiography screening
Fetal echocardiography involving the four-chamber view of the heart and the outflow tracts forms part of the 'Fetal anomaly base menu'. As a minimum, four basic intracardiac views are required they are: laterality, the four-chamber view, the left ventricular outflow tract and the right ventricular outflow tract. A detailed description of all the structures that require assessment is outlined in detail below. The use of colour flow Doppler is not a requirement, but it should be encouraged as it may help provide additional information and improve detection of CHD. It is likely that the use of colour flow Doppler will be incorporated into the assessment of fetal echocardiography in 2013.

View	Size	Position	Structure	Function
Situs/Laterality Determine left and right side of the fetus from position in uterus		Stomach and heart on the left		
Four-Chamber Transverse section of the thorax including a complete rib and crux of the heart	*Normal cardiac size occupies 1/3 of area of the thorax (measurement not required) *X2 atria of equal size *X2 ventricles of equal size *X2 patent atrioventricular valves of equal size	*Mostly in the left chest *Apex points towards the left	*Left and right side of the heart are symmetrical *Moderator band at right ventricle apex *Crux - point at which lower part of atrial septum meet upper part ventricular septum and where both atrioventricular valves are inserted * Differential offsetting of valves, the tricuspid valve inserts more apically than the mitral valve *Ventricular septum intact from apex to crux *Foramen ovale flap in left atrium	*Rhythm - synchronous atrial and ventricular contractions *Two ventricles contract equally *Mitral and tricuspid valves open freely

Aorta/Left Ventricular Outflow Tract This view shows the outflow tract of the left ventricle		Aorta arises from the left ventricle & sweeps out towards the right shoulder	The anterior wall of the aorta is continuous with the ventricular septum	Aortic valve opens freely
***Pulmonary/Right Ventricular Outflow Tract**[30] This view shows the outflow tract of the right ventricle **Or Three-Vessel View (3VV)** This view shows the outflow tract of the right ventricle including the pulmonary artery, the aorta and the superior vena cava.	The diameter of the pulmonary artery is slightly greater than the diameter of the aorta which is slightly greater than the diameter of the superior vena cava	Main pulmonary artery arises from the right ventricle and is directed backwards towards the spine *The pulmonary artery lies to the left with the superior vena cava to the right and aorta in the middle *Pulmonary artery continues as the arterial duct	The main pulmonary artery bifurcates	Pulmonary valve opens freely

[30] * The outflow tract of the right ventricle can be visualised in either the right ventricular outflow tract view or the three-vessel view. Depending on fetal position and lie one view may be easier to obtain than the other. **Either view when normal implies normal crossover**. **Only one view is required**.

Abbreviations

AC	Abdominal Circumference
AFP	Alpha-fetoprotein
ATSM	Advanced Training Speciality Module
BMI	Body Mass Index
BMUS	British Medical Ultrasound Society
CASE	Consortium for the Accreditation of Sonographic Education
CEL	Chief Executive Letter
CHD	Congenital Heart Disease
CHI	Community Health Index
CI	Confidence Interval
CMU	Certificate in Medical Ultrasound
CPA UK (Ltd)	Clinical Pathology Accreditation United Kingdom Limited
CPD	Continuous Professional Development
CRL	Crown Rump length
DMU	Diploma in Medical Ultrasound
DR	Detection Rate
DQASS	Down's syndrome screening Quality Assurance Support Service
EDD	Estimated Date of Delivery
EDTA	Ethylenediaminetetraacetic acid
EU	European Union
FβhCG	Free-beta subunit human chorionic gonadotropin
FL	Femur Length
FMF	Fetal Medicine Foundation
GMC	General Medical Council
GP	General Practitioner
HC	Head Circumference
HCG	Human Chorionic Gonadotropin
HDL	Health Department Letter
HIE	Higher Institute of Education
HPC	Health Professional Council
HTA	Health Technology Assessment
IT	Information Technology
KPI	Key Performance Indicator
MoM	Multiples of the Median
NHS QIS	National Health Service Quality Improvement Scotland
NICE	National Institute for Clinical Excellence
NMC	Nursing and Midwifery Council
NSD	National Services Division
NSS	National Services Scotland
NT	Nuchal Translucency
NTD	Neural Tube Defect
PACS	Picture Archiving and Communication Systems
PAPP-A	Pregnancy-associated plasma protein A
PgCert	Postgraduate Certificate
PgCert.MU	Post Graduate Certificate in Medical Ultrasound
QC	Quality Control
QF PCR	Quantitative Fluorescent-Polymerase Chain Reaction
RCOG	Royal College of Obstetricians and Gynaecologists
RCR	Royal College of Radiologists
SCoR	Society and College of Radiographers
SGHD	Scottish Government Health Directorates
SPR	Screen Positive Rate
SWHMR	Scottish Woman Held Maternity Record

TV	Trans-Vaginal
UE3	Unconjugated Oestradiol
UK NEQAS	United Kingdom National External Quality Assessment Service
UK NSC	United Kingdom, National Screening Committee
UK NSC FASP	United Kingdom National Screening Committee Fetal Anomaly Screening Programme

Glossary

Affected pregnancies	Pregnancies in which the fetus has the target condition.
Amniocentesis	An invasive procedure undertaken from about 15 completed weeks (15+0) onwards to obtain a sample of amniotic fluid surrounding the fetus. A needle is passed through the mother's abdomen into the uterus, under continuous ultrasound guidance, and a sample of fluid is withdrawn. The fluid, and cells within it, can be tested for certain conditions such as Down's syndrome and other chromosomal and inherited disorders. Out of 100 women who have this test from 15 weeks it is likely that one will miscarry as a direct consequence of the procedure.
Amniotic fluid	The fluid surrounding the fetus in the uterus, which protects it during pregnancy and labour. It contains substances and cells from the fetus, which can be removed by amniocentesis and examined.
Antenatal	The period from conception to birth.
Chorionic villus sampling (CVS)	An invasive procedure performed under ultrasound guidance after 10 completed weeks of pregnancy to obtain a sample of placental tissue, which is taken through either the cervix or the abdomen. The range of chromosomal and genetic conditions that can be detected is similar to those for amniocentesis except that Neural Tube Defects cannot be diagnosed. For every 100 women who have this test from the 11th week in pregnancy one or two will miscarry.
Chromosome	Structures found in the nucleus of cells, composed of DNA and proteins. Normally humans have 46 chromosomes in each cell, 23 from each parent. Of these, 22 are autosomes and one is a sex chromosome.
Chromosome anomaly	A change in the number or arrangement of the normal 23 pairs of chromosomes.
Combined test	Between 11weeks $^{+2}$ days and 14 weeks $^{+1}$ days weeks of pregnancy, a combination of the Nuchal Translucency (NT) measurement and a blood sample from the mother which measures the concentration of pregnancy associated plasma protein-A (PAPP-A), and free beta human chorionic gonadotrophin (Free beta hCG). Together with the mother's age and the gestation of the pregnancy, these are used to estimate the chances that the fetus is affected Down's syndrome.
Confirmed result	The results of initial screening tests are not usually 100% certain, and are often called presumptive results. The results of screening tests are NOT confirmed results. They are often confirmed later, with further diagnostic tests.
Congenital	Present at or shortly after birth.
Congenital anomaly	An anomaly present at birth, although not necessarily hereditary.

Coverage	This is the proportion of people in the eligible group who actually undergo the screening.
Cut-off level	Screening tests divide people into a group at lower risk of the condition being screened for, and a group at higher risk who are then offered further investigations. Cut off level is a point defined by the programme used to distinguish higher and lower risk.
Detection rate	The proportion of affected individuals with a positive screening result
Diagnostic test	Refers to the analytical process involved in obtaining a result. For example the diagnostic test on an amniocentesis sample (invasive procedure) is the karyotype or PCR.
Disorder	Several words are used to describe illnesses. They are sometimes called diseases, disorders or conditions.
DNA (deoxyribonucleic acid)	The molecule that encodes genetic information. DNA is a double-stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C
Down's syndrome (trisomy 21)	A disorder caused by the presence of an extra copy (three instead of two) of chromosome 21. It affects all population groups and is distinguished by a number of features occurring together including low muscle tone, a face that appears flatter with eyes slanting upward, small ears and an unusually wide neck and a deep crease across the palm of the hand. Some may have heart problems or visual problems or may develop Alzheimer's disease. Although people with Down's syndrome have learning difficulties, these vary in severity.
Edwards' syndrome (trisomy 18)	A syndrome caused by the presence of an extra copy (three instead of two) of chromosome 18. The combination of features present in babies affected with trisomy 18 can lead to many different problems including growth deficiency, feeding and breathing difficulties, developmental delays, learning difficulties, undescended testes in males, kidney malformations, and heart defects. They may also have malformations in the bones. Survival of infants with trisomy 18 depends on how severely they are affected. Most do not survive the first year of life.
Effectiveness	The extent to which intervention results in the desired outcomes under everyday conditions.
Eligible group	Target group for offer of screening.
Embryo	A fertilised ovum (egg) in the early stage of development. In humans the term is reserved for the first eight weeks of development.
False-negative result	Screening tests divide people into lower and higher risk groups. Some people with a negative screening test result do actually have the condition being screened for. These people are said to have a 'false-negative' result.

False-positive result	Screening tests divide people into lower and higher risk groups. Some people with a positive screening test result do not actually have the condition being screened for. These people are said to have a 'false-positive' result.
Family history	History of a condition in at least one of the following family members: parent, sibling, grandparent, great-grandparent, aunt, uncle, nephew, niece or cousin or child.
Fetal Anomaly/ Abnormality	These terms are used interchangeably: are synonyms
Fetal Anomaly ultrasound scan	A screening test offered to pregnant women to monitor the growth and development of the fetus before birth by producing a real-time visual image. Scans before 16 weeks are useful for dating and assessing the viability of the pregnancy (and are able to detect some major malformations). Detailed scanning at 18–20^{+6} weeks should show up most malformations as well as some minor ones.
Fetus	In humans, the unborn child after the end of the eighth week of pregnancy to the moment of birth.
Gene	The unit of a chromosome through which particular characteristics are inherited from one or both parents.
Genetic counselling	Information and support provided by an appropriately trained health professional, to individuals who have known conditions in their families or who are concerned about the future possibility of genetically inherited conditions.
Genetic counsellor	A health professional with specialised training in genetics and counselling who can provide information and support for individuals or families with concerns about a genetic disorder that may run in the family.
Genetic testing	Examination of an individual's genetic material to identify alterations that may cause a disorder.
Genetics	1. The study of the structure and function of genes. 2. The genetic features which occur in individuals, families and populations.
Gestational age	The duration of an ongoing or completed pregnancy, measured from the first day of the last menstrual period (usually about two weeks longer than that measured from conception).
Incidence	The number of new instances of a specific condition occurring during a certain period in a specified population.
Inheritance	The passing of familial characteristics from one generation to the next.
Inherited	Having a hereditary characteristic; there are many inherited characteristics, including eye colour, hair colour and health disorders.
Invasive procedure	Invasive procedure - is a method used to obtain a sample, usually to aid diagnosis e.g. amniocentesis and chronic villi sampling are invasive procedures.
In vitro fertilisation (IVF)	The process whereby an egg is fertilised with sperm in the test tube and then transplanted into a woman's uterus.
Karyotype	A photomicrograph of an individual's chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type; used to correlate chromosomal anomalies with the characteristics of specific diseases. Karyotyping is often used for prenatal diagnosis of conditions such as Down's syndrome.

Marker	A protein or steroid hormone measured in maternal serum which shows altered levels in pregnancies affected by some fetal abnormalities such as Down's syndrome and NTDs.
Miscarriage	Loss of a fetus before the 24th week of pregnancy.
Morbidity	The extent of being affected by a disease or condition. In epidemiology, the morbidity rate is the prevalence of a disease within a particular number of the population.
Morbidity rate	In epidemiology, the prevalence of a disease within a population, usually expressed as cases per 100,000.
Mortality/mortality rate	The incidence of death in a population in a given period.
Mosaic	An individual who has some cells with an unusual genetic or chromosomal make-up while the rest of the cells in the body have the typical genetic or chromosomal constitution.
Mutation	A change in the gene resulting from an error made when the gene is being copied. It may result in altered gene function. 'Alteration' may be more acceptable to women and their families.
Neural tube	The embryonic structure that forms into the skull and spine.
Neural tube defect (NTD)	An anomaly where the spine has not closed over the central nervous tissue. If this lesion is at the head, the condition is called anencephaly and is incompatible with life. If it occurs lower down the spine it is called spina bifida and results in varying degrees of physical and learning disabilities. Detailed ultrasound scanning is the way in which these conditions are confirmed, often following a raised AFP blood test. Most NTDs are 'open', which means there is no skin over the lesion in the spine; about one in seven cases of spina bifida are 'closed', which means that although the spine has not covered the nervous tissue there is a covering of skin. Closed conditions are less likely to be detected antenatally by the AFP test.
Non-invasive	A procedure that does not require incision into the body or the removal of tissue.
Nuchal scan (Nuchal transluscency scan NT)	At 11–14 weeks $^{+1}$ day of pregnancy the thickness of fluid at the nape of the fetal neck, the nuchal translucency, is measured. An increased amount of fluid may indicate that the fetus has Down's syndrome, structural or genetic anomaly. By combining the mother's age and the gestation of the pregnancy with information from the scan an individual statistical chance of an anomaly can be given for that particular pregnancy. If the chance is one in 150 or higher a diagnostic test, such as CVS, will be offered.
Patau's syndrome (trisomy 13)	A disorder caused by the presence of an extra copy (three instead of two) of chromosome 13. The disorder is characterised by low birth weight, cleft lip or palate, defects of the heart, eye structure, spine, scalp and abdomen, abnormal genitalia, low set ears, abnormal palm pattern, extra digits and overlapping of fingers over thumb. Between 80 per cent and 90 per cent of babies do

	not survive infancy and those that do survive have learning disabilities.
Placenta	The structure that provides the fetus with nourishment during development. It is attached to the wall of the uterus and connects to the fetus through the umbilical cord.
Polymerase chain reaction (PCR)	A rapid diagnostic test for the most common chromosomal and genetic anomalies. Using a small amount of amniotic fluid, PCR amplifies and enables specific regions of the DNA molecule to be quantified from uncultured amniocytes. The test is used to provide a definitive diagnosis of Down's syndrome, haemoglobin disorders and other single-gene disorders.
Predisposition	A situation in which a person, due to their inherited genetic makeup, may have a particular susceptibility to a condition if exposed to the appropriate environmental triggers.
Prevalence	The proportion of people in a population who have a given disease or attribute.
Prevalence rate	The number of people with the condition or attribute, divided by the population at risk.
Prognosis	Predicted course and outcome of a disorder, based on all the knowledge related to a specific case, e.g., age, sex, the course of the disorder in other patients.
Quadruple test	Second trimester test to calculate the risk of Down's syndrome, usually based on the measurement of AFP, uE3, free b-hCG (or total hCG), and inhibin-A together with the woman's age.
Quality assurance	A system for monitoring and maintaining high standards in every aspect of a screening programme.
Risk	Risk is usually taken to mean the chance of an event happening. It can be expressed in a number of ways.
Screening	Screening is a process of identifying apparently healthy people who may be at increased risk of a disease or condition. They can then be offered information, further tests and appropriate treatment to reduce their risk and/or any complications arising from the disease or condition.
Screening programme	The whole system of activities needed to deliver high quality screening. It ranges from identifying and informing those to be offered screening through to the treatment and follow up of those found to have abnormality, and support for those who develop disease despite screening.
Screening test	A test or inquiry used on people who do not have or have not recognised the signs or symptoms of the condition being tested for. It divides people into low and higher risk groups.
Sensitivity	This is a measure of test performance. High sensitivity means that the test 'catches' as many people with the condition as possible. It is measured as the proportion of those with the condition, who have a positive test result. It is the same as the detection rate.
Specificity	This is a measure of test performance. High specificity means the test has as few false positives as possible. It is measured as the proportion of those without the condition, who have a negative test result.

Surveillance	Ongoing observation of the health of individuals or populations.
Syndrome	Combination of symptoms and signs grouped together to form a disorder.
Termination of pregnancy	The medical expulsion or extraction from the uterus of a fetus in the first, second or third trimester of pregnancy.
Trisomy	Three copies of a particular chromosome rather than the usual pair.
True-negative result	Screening tests divide people into low and higher risk groups. Most of the people with a negative screening test result do not have the condition being screened for. These people are said to have a ‘true-negative’ result.
True-positive result	Screening tests divide people into low and higher risk groups. Some people with a positive screening test result do have the condition being screened for. These people are said to have a ‘true-positive’ result.
Twins	May be genetically identical (monozygous) when they arise from a single fertilised egg or non-identical (dizygous) when they arise from two separate eggs.
Uptake	Is the proportion of people, who when offered a test, take it up.
Variant	A change for example in a gene or protein. For example, a variant in a haemoglobin gene resulting in a variant in the haemoglobin the body produces thus causing a sickle cell disease.