



Public Health
England

NHS Fetal Anomaly Screening Programme Handbook

Valid from August 2018

Public Health England leads the NHS Screening Programmes



About Public Health England

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About PHE screening

Screening identifies apparently healthy people who may be at increased risk of a disease or condition, enabling earlier treatment or better informed decisions. National population screening programmes are implemented in the NHS on the advice of the UK National Screening Committee (UK NSC), which makes independent, evidence-based recommendations to ministers in the four UK countries. The Screening Quality Assurance Service ensures programmes are safe and effective by checking that national standards are met. PHE leads the NHS Screening Programmes and hosts the UK NSC secretariat.

www.gov.uk/topic/population-screening-programmes

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Contents

About Public Health England	2
About PHE screening	2
Executive summary	5
Introduction	6
The NHS Fetal Anomaly Screening Programme	7
General principles of screening	7
Background	7
Screening policy	8
Markers used in screening tests	9
Biochemical markers	10
Effect of vaginal bleeding on biochemical markers	10
Ultrasound markers	10
Down's syndrome, Edwards' syndrome and Patau's syndromes	11
Down's syndrome	11
Edwards' syndrome	11
Patau's syndrome	12
Screening tests	13
The early pregnancy scan	13
First trimester combined test	13
Second trimester quadruple test	14
Screening in twin pregnancies	15
National standards for T21/T18/T13 screening	15
The 18 ⁺⁰ to 20 ⁺⁶ week fetal anomaly ultrasound scan	16
18 ⁺⁰ to 20 ⁺⁶ NHS FASP ultrasound scan base menu	18
Fetal cardiac protocol	18
Normal variant	19
Image capture, storage and archiving	20
Training and professional competence	20
Safety of ultrasound	21
Diagnostic testing	22
Results of diagnostic testing	23
Audit	23
Non-invasive prenatal testing (NIPT)	23
Quality assurance	25
Key performance indicators	26
Screening safety incidents	26
Glossary	27

Appendix 1	32
18 ⁺⁰ to 20 ⁺⁶ FASP ultrasound scan base menu	32
Appendix 2 – Ultrasound images and schematics	34

Executive summary

The NHS Fetal Anomaly Screening Programme (FASP) has produced this handbook with support from the members of the programme advisory and task groups and in collaboration with health care professionals from around England.

This practical guidance supports healthcare professionals and stakeholders in the operational delivery of the screening pathway. New screening co-ordinators will find the handbook a source of information to support their induction and practice.

The handbook provides an update of recent changes to the programme. It refers to supporting documents and clinical guidance that providers should take into account to deliver a high quality screening programme.

Commissioners and screening and immunisation teams will find the handbook puts these documents into the context of the day to day work of the screening coordinators, midwives, sonographers, screening laboratory teams and obstetricians and fetal medicine specialists who make up the FASP multi-disciplinary team (MDT).

The handbook provides:

- structure and governance of the NHS FASP programme
- a comprehensive outline of each of the conditions
- clarity on the screening test and terminology
- hyperlinks to information and supporting documents
- detail on the delivery of each step of the screening pathway
- key practice points to consider
- updates on current quality assurance, data collection and audit processes

The handbook will be updated regularly to ensure it continues to be a valid reference document.

Introduction

The purpose of this handbook is to bring together in one publication the NHS Fetal Anomaly Screening Programme's (FASP) guidelines and recommendations that relate to the screening pathway and are not covered in detail in the other handbooks.

Throughout the document the following are used interchangeably:

- Down's syndrome is referred to as T21
- Edwards' syndrome as T18
- Patau's syndrome as T13

This handbook is part of a suite of documents that are reviewed and updated annually.

1. **Service specifications.** Down's syndrome, Edwards' syndrome and Patau's syndrome (No. 16) and 18⁺⁰ to 20⁺⁶ (18 weeks and 0 days of pregnancy to 20 weeks and 6 days of pregnancy) fetal anomaly scan (No.17). These documents outline the service and quality indicators expected by NHS England and the recommendations and standards of the UK National Screening Committee (UK NSC).
2. **Programme Standards.** These define a set of standards relating to screening for Down's syndrome, Edwards' syndrome and Patau's syndrome and the 18⁺⁰ to 20⁺⁶ week fetal anomaly scan.
3. **Handbook for laboratories.** This sets out the requirements for laboratory staff involved in the pathways for first trimester screening for Down's syndrome, Edwards' syndrome and Patau's syndrome and second trimester biochemical screening for Down's syndrome.
4. **Ultrasound Practitioner's Handbook.** This sets out the requirements for ultrasound practitioners involved in the pathway for first trimester screening for Down's syndrome, Edwards' syndrome and Patau's syndrome.

The NHS Fetal Anomaly Screening Programme

General principles of 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. Further information regarding the general principles of screening can be found on [GOV.UK](https://www.gov.uk)

Background

NHS FASP, based in Public Health England, is an expert team that ensures national consistency and provides expertise. They support and manage ongoing roll out, technical and professional development of the programme and ensure quality and safety standards are maintained and continuously improved.

The screening programme has evolved from its establishment in 2001 when the majority of screening was performed using maternal biochemistry in the second trimester. The recommended method of screening is now first trimester screening, combining maternal age, biochemistry and ultrasound measurement of fetal nuchal translucency to provide a pregnant woman with her chance of having a baby with Down's syndrome, or Edwards' syndrome and Patau's syndrome.

The offer of a fetal anomaly scan is recommended and where accepted should be undertaken between 18⁺⁰ to 20⁺⁶ weeks of pregnancy. The fetal anomaly scan base menu sets out the fetal anatomy to be examined. The fetal anomaly scan screens for 11 conditions. NHS FASP has established standardisation of the following to enable commissioners and quality assurance teams to assess the quality of the service provided. These include:

- national standards, guidance and chance cut-off for Down's syndrome, Edwards' syndrome and Patau's syndrome screening programme in the light of emerging evidence
- a statistical service to ensure that laboratories have access to the best advice in maintaining their population medians
- specifications for chance calculation software to make sure that all laboratories calculate chance in a uniform way

- use of a base menu and fetal cardiac protocol to enable consistency in the structures examined as part of the 18⁺⁰ to 20⁺⁶ week fetal anomaly scan

Screening policy

NHS FASP offers screening to all eligible pregnant women in England to assess the chance of the baby being born with Down's syndrome, or Edwards' syndrome and Patau's syndrome or a number of fetal anomalies (structural abnormalities of the developing fetus).

NHS FASP requires that there is equal access to uniform and quality-assured screening across England and women are provided with high quality information. So they can make an informed choice about their screening and pregnancy options. Education and training resources are available for staff covering all stages of the process, from informing women of test availability, through to understanding and supporting their decisions.

The screening policy is to offer screening to assess the chance of the baby being born with Down's syndrome or Edwards' syndrome and Patau's syndrome. The test of choice for both singleton and twin pregnancies is first trimester combined screening. Women can choose:

- not to have screening
- to have screening for T21 and T18/T13
- to have screening for T21 only
- to have screening for T18/T13 only

The first scan usually takes place between 10 to 14 weeks and includes a blood sample taken to test for Down's syndrome, Edwards' syndrome and Patau's syndrome, with a second scan for fetal anomalies between 18⁺⁰ to 20⁺⁶ weeks. The timing of the scans allows for further diagnostic tests if required and ensures women have time to consider decisions about continuing their pregnancy. The second scan is designed to identify abnormalities which indicate:

- the baby may die shortly after birth
- conditions that may benefit from treatment before birth
- to plan delivery in an appropriate hospital/centre and/or to optimise treatment after the baby is born mean that the baby may die shortly after birth, conditions that may benefit treatment before birth, to plan delivery in an appropriate hospital/centre and/or to optimise treatment after the baby is born

Some women may choose not to be screened at all. It is important that this choice is respected.

Markers used in screening tests

All women have a chance of having a baby with Down’s syndrome, Edwards’ syndrome or Patau’s syndrome and this chance increases with age. The older a mother, the more chance she has of having a baby with the one of these conditions

Table 1: Example for a woman who is 16 weeks pregnant

Maternal age	Chances of having a pregnancy affected by Down’s syndrome	Probability
20 years	1 in 1500	0.07%
30 years	1 in 900	0.1%
40 years	1 in 100	1%

Figure 1: Graph to illustrate the likelihood of a pregnancy affected by Down’s syndrome, Edwards’ syndrome or Patau’s syndrome according to maternal age showing T21 (black line), T18 (blue line), T13 (green line). The chance is at the time of the 12 weeks scan

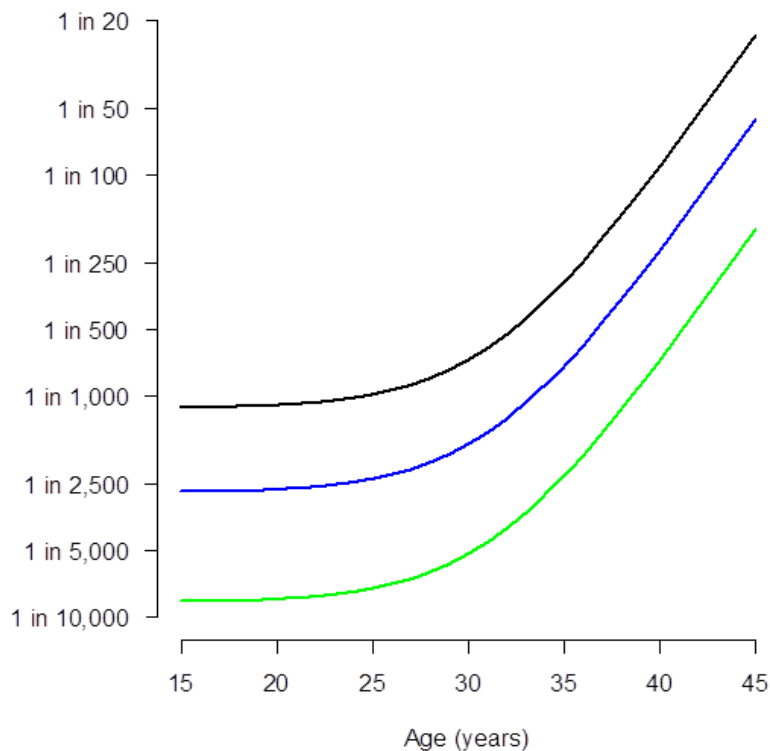


Table 2: Reframing chance

Chance of an affected pregnancy		Chance of an unaffected pregnancy	
1 in 4	25%	3 in 4	75%
1 in 5	20%	4 in 5	80%
1 in 10	10%	9 in 10	90%
1 in 20	5%	19 in 20	95%
1 in 30	3%	29 in 30	97%
1 in 50	2%	49 in 50	98%
1 in 100	1%	99 in 100	99%
1 in 200	0.5%	199 in 200	99.5%

Can be applied to any screening test where the result is reported as a probability.

Biochemical markers

There are 5 analytes (commonly referred to as markers) measured by the laboratory that are used to calculate the likelihood of a pregnancy being affected by Down’s syndrome, Edwards’ syndrome or Patau’s syndrome – 6 if human chorionic gonadotropin (hCG) and its free beta subunit are considered as 2 separate analytes. Refer to the [laboratory handbook](#) for more information on biochemical markers.

Effect of vaginal bleeding on biochemical markers

There are concerns that a history of significant maternal vaginal bleeding might change the levels of biochemical markers used in the combined test. NHS FASP recommends women are offered the combined test in the normal way (calculating the chance based on maternal age, NT, free beta hCG and PAPP-A levels). Current evidence suggests that the biochemical marker levels are not significantly different in women with this history.

Ultrasound markers

Please refer to the [laboratory](#) and [ultrasound practitioner’s](#) handbooks for more detailed information on ultrasound markers.

Down's syndrome, Edwards' syndrome and Patau's syndromes

Inside the cells of our bodies there are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. There are 23 pairs of chromosomes in each cell. Changes can occur in the sperm or egg cells, which can lead to a baby having an extra chromosome.

Down's syndrome

People with Down's syndrome (T21) have extra chromosome 21 in the cells of their body.

A person with Down's syndrome will have some level of learning disability. This means they will find it harder than most people to understand and to learn new things. They may have communication challenges and difficulty managing some everyday tasks. People with Down's syndrome have distinctive facial features but they do not all look the same. Most children with Down's syndrome attend mainstream schools but will require additional support.

Some health problems are more common in people with Down's syndrome. These include heart conditions and problems with hearing and vision. Many health problems can be treated but unfortunately around 5% of babies will not live past their first birthday.

For babies without serious health problems survival is similar to that of other children and most people with Down's syndrome will live into their 60s or longer. People with Down's syndrome can have a good quality of life and most say they enjoy their lives. With support, many more people with Down's syndrome are able to get jobs, have relationships and live semi-independently in adulthood.

Edwards' syndrome

Babies with Edwards' syndrome have an extra copy of chromosome 18 in all or some cells. A condition caused by the presence of an extra copy (3 instead of 2) of chromosome 18.

Sadly the survival rates are low and of those babies born alive only around 10% live past their first birthday. Some babies may survive to adulthood but this is rare. All babies born with Edwards' syndrome will have a learning disability and a wide range of

physical challenges, which can be extremely serious. They may have problems with their heart, limbs, kidneys and digestive system. Babies with Edwards' syndrome may have a low birthweight.

Patau's syndrome

Babies with Patau's syndrome have an extra copy of chromosome 13 in all or some cells.

Sadly the survival rates are low and of those babies born alive only around 10% live past their first birthday. Some babies may survive to adulthood but this is rare. All babies born with Patau's syndrome will have a learning disability and a wide range of physical challenges, which can be extremely serious. They may have problems with their heart, limbs, kidneys and digestive system.

Around half of babies with Patau's syndrome will have a cleft lip and palate. Babies with Edwards' syndrome and Patau's syndrome may have a low birthweight. Despite their difficulties children can slowly make progress in their development.

Older children with either Edwards' syndrome or Patau's syndrome are likely to attend a specialist school.

Screening tests

The early pregnancy scan

The scan has several purposes. It is to:

- confirm viability
- ascertain if it is a singleton or multiple pregnancy
- estimate gestational age
- detect major structural anomalies that may be identified in early pregnancy eg anencephaly

If the woman accepts screening for T21, T18/T13 syndromes, the scan is one component of the screening test. Ultrasound scanning in pregnancy should, in the first instance, be performed transabdominally.

Assessment techniques and biometric charts used for fetal measurements must meet nationally agreed standards. [BMUS – Loughna et al 2009](#).

First trimester combined test

The combined test uses maternal age, the nuchal translucency measurement (NT) and 2 biochemical markers, free beta hCG and PAPP-A, together with the gestational age calculated from the crown rump length (CRL) measurement, to calculate the chance of the pregnancy being affected by T21 or T18/T13. The optimal time to perform the combined test is between 11⁺¹ weeks to 14⁺¹ weeks of gestation, which corresponds to a CRL of 45.0 mm to 84.0 mm.

In cases where screening is accepted but it is not possible to obtain the NT measurement at the first appointment, at least one other attempt should be offered. This may be on the same day or at a later date. If it is not possible to obtain an accurate NT measurement despite 'twice on the couch' then further attempts do not have to be offered and the woman should be referred in to the second trimester screening pathway.

If the ultrasound measurement shows that the CRL is less than 45.0 mm, the woman should be recalled for a further scan to measure the NT. If the CRL is greater than 84.0 mm, the second trimester quadruple test should be offered.

The first trimester combined test allows earlier decision making for parents. In practice, 2 models are available for performing the combined test:

1. A maternal blood specimen may be taken (from 10 weeks onwards) prior to the ultrasound scan. The biochemistry results can then be made available at the time of the NT scan and the combined test result can be calculated at the time of the appointment. Although the result may be calculated by the sonographer, the laboratory remains responsible for the software that calculates the screening result. Midwifery and/or ultrasound departments must have a process in place to share ultrasound measurements and final screening results with the laboratory to enable timely audit of all results.
2. A maternal blood specimen may be taken at the time of the ultrasound scan and the combined test result made available within a few days of the biochemistry results being authorised by the laboratory.

When calculating a chance for T21 and/or T18/T13 syndromes, the nuchal translucency measurement must be used in combination with a maternal serum screening test. The nuchal translucency measurement must not be used in isolation.

Where women have chosen not to accept screening for Down's syndrome, Edwards' syndrome and Patau's syndrome, but choose to accept an early pregnancy scan, structural anomalies may still be identified, including an NT of $\geq 3.5\text{mm}$. It is not within NHS FASP's remit to provide guidance regarding the clinical care of women who have declined screening. However, they should be aware that any such anomaly will be reported and signposted as per local clinical guidelines for care and management.

NHS FASP recommends that the Down's syndrome and/or Edwards' syndrome and Patau's syndrome screening chance generated from first trimester combined screening must not be recalculated up or down. Following the initial screening test or at the 18⁺⁰ to 20⁺⁶ fetal anomaly ultrasound scan due to the presence or absence of a single ultrasound marker of less predictive power than increased nuchal fold ([Smith-Bindman et al, 2001](#)).

For further information regarding the scan element of the combined screening test please see the [Ultrasound practitioner's handbook](#).

Second trimester quadruple test

The quadruple test uses maternal age and 4 biochemical markers measured from 14⁺² weeks until 20⁺⁰ weeks - AFP, hCG (total, intact or free beta subunit), uE3 and Inhibin-A. Although this combination of markers has a lower detection and a higher screen positive rate than the combined test, it is the nationally recommended screening strategy in the second trimester.

There will always be a need for a screening test in the second trimester for those women who book too late for first trimester testing or when an NT measurement cannot be obtained (despite twice on the couch) in the first trimester. Further information regarding the

practicalities of a solution to combining dating and screening requirements at the early pregnancy scan are explored in more detail in the following article: [Chudleigh et al \(2011\)](#)

Screening in twin pregnancies

Women with a twin pregnancy are eligible for combined screening or quadruple screening dependent on gestational age. For detailed information regarding screening in twin pregnancies please see section 6 of the [Laboratory Handbook](#).

National standards for T21/T18/ T13 screening

The national standards seen in Table 3 state the threshold for the national programme and will be reported on each year by the Down's syndrome screening Quality Assurance Support Service (DQASS).

Table 3. National Standards – Thresholds for performance

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

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

The 18⁺⁰ to 20⁺⁶ week fetal anomaly ultrasound scan

NHS FASP recommends the offer of a mid-pregnancy scan which is undertaken between 18⁺⁰ to 20⁺⁶ weeks of pregnancy to screen for major fetal anomalies. The examination should be undertaken in accordance with the 18⁺⁰ to 20⁺⁶ NHS FASP ultrasound scan base menu and fetal cardiac protocol.

Some providers are able to arrange the fetal anomaly scan later within the recommended window that is closer to 20 weeks as opposed to 18 weeks. Where this occurs, services must be able to facilitate referrals for further investigations and options for pregnancy choices in a timely manner and within the required national timeframes. Ongoing audit of practice should be in place to monitor conformity. The screening pathway must be completed by 23⁺⁰ weeks of pregnancy.

Women who wish to have a fetal anomaly ultrasound scan, but do not wish to be informed if abnormalities are found, should be advised that all significant findings seen on scan will be reported and, therefore, they should consider not having fetal anomaly ultrasound screening.

The main structures to be assessed at the 18⁺⁰ to 20⁺⁶ week scan are defined as shown in Table 4. Abnormalities of these structures can indicate a number of specific conditions. Other conditions may be detected using this ultrasound screening test, but there are insufficient data to confidently predict the standard which should be achieved.

11 conditions are specified that indicate:

- the baby may die shortly after birth
- conditions that may benefit from treatment before birth
- to plan delivery in an appropriate hospital/centre and/or to optimise treatment after the baby is born

(Fetal Anomaly Ultrasound Screening Programme Study: Literature Survey June 2007)

Table 4: The conditions screened for as a minimum in England

Conditions	Detection rate (%)
Anencephaly	98
Open spina bifida	90
Cleft lip	75
Diaphragmatic hernia	60
Gastroschisis	98
Exomphalos	80
Serious cardiac anomalies includes the following: <ul style="list-style-type: none"> • Transposition of the Great Arteries (TGA) • Atrioventricular Septal Defect (AVSD) • Tetralogy of Fallot (TOF) • Hypoplastic Left Heart Syndrome (HLHS) 	50
Bilateral renal agenesis	84
Lethal skeletal dysplasia	60
Edwards' syndrome (Trisomy 18)	95**
Patau's syndrome (Trisomy 13)	95**

**Detections rates will be reviewed once sufficient data is received following implementation of screening as part of the combined screening strategy.

It is accepted that an ultrasound scan at this time can also constitute part of general clinical practice and management as well as screening. The 2 are closely linked.

Although, it is not the remit of the screening programme to set out standards or guidance on the management of these areas, it is acknowledged that by not incorporating a reference to them in the 18⁺⁰ to 20⁺⁶ NHS FASP ultrasound scan base menu, it may give the impression that they should not be noted during the ultrasound scan. The examination of placental position and amniotic fluid, whilst not part of the screening protocol, is good clinical practice.

There is no requirement to determine fetal gender within the NHS FASP in England; it is not part of the 18⁺⁰ to 20⁺⁶ NHS FASP ultrasound scan base menu. There is no programme requirement to recall the woman if the fetal sex is not identified due to poor visualisation or difficult fetal position.

18⁺⁰ to 20⁺⁶ NHS FASP ultrasound scan base menu

The 18⁺⁰ to 20⁺⁶ NHS FASP ultrasound scan base menu (Appendix 1) specifies measuring techniques and defines the anatomical structures to be assessed. This promotes consistency in the examination.

The fetal anatomy to be examined and measured is:

1. Head circumference demonstrating HC measurement and measurement of the atrium of the lateral ventricle.
2. Suboccipitobregmatic view demonstrating measurement of the transcerebellar diameter.
3. Coronal view of lips with nasal tip.
4. Abdominal circumference demonstrating AC measurement.
5. Femur length demonstrating FL measurement.
6. Sagittal view of spine including sacrum and skin covering.

These 6 specific fetal anatomical sections should be captured and archived at examination. A hard copy image of each section (see Appendix 1) and report should be recorded and appropriately stored in any combination of the following formats:

- ultrasound clinical information storage system
- auditable electronic hospital information system
- ultrasound request/report form
- in the woman's hand-held notes

The head circumference (HC), abdominal circumference (AC) and femur length (FL) measurements should be taken to assess growth velocity in a pregnancy where the expected date of delivery (EDD) was previously assigned in line with nationally approved charts and tables.

If the EDD was not previously assigned, the pregnancy should be dated by HC or FL. [Loughna P, et al \(2008\)](#).

Fetal cardiac protocol

The views required are:

1. Situs/Laterality.
2. Four-Chamber: Transverse section of the thorax including a complete rib and crux of the heart.
3. Aorta/Left Ventricular Outflow Tract: This view shows the outflow tract of the left ventricle.

4. Pulmonary/Right Ventricular Outflow Tract: This view shows the outflow tract of the right ventricle only or the Three-Vessel View (3VV): This view shows the outflow tract of the right ventricle including the pulmonary artery.
5. The 3 vessel and trachea view (3VT): a transverse view of the fetal upper mediastinum; it depicts the main pulmonary artery in direct communication with the ductus arteriosus, the transverse aortic arch and the superior vena cava.

A single repeat scan must be offered and completed by 23⁺⁰ weeks gestation in cases where the image quality of the first examination is compromised by one of the following:

- increased maternal body mass index (BMI)
- uterine fibroids
- abdominal scarring
- sub-optimal fetal position

The woman should be rescanned on the same day or offered a new appointment according to local clinical assessment. If first examination is sub-optimal and the sonographer is suspicious of a possible fetal anomaly, a second opinion should be sought and a referral made for further investigation of the anomaly suspected. This should be documented. There is no requirement to wait for a scan to refer in these circumstances.

Where an adequate assessment of the fetal anatomy remains compromised after the repeat scan, there is no requirement to offer further ultrasound examination for completion of screening. The woman should be informed that the screening is incomplete and this should be recorded.

Normal variant

The introduction of a national Down's syndrome, Edwards' syndrome and Patau's syndrome screening programme in early pregnancy has changed the way in which the 18⁺⁰ to 20⁺⁶ fetal anomaly scan findings are interpreted. NHS FASP recommends that an established screening test result should not be recalculated at this time.

The screening programme is increasingly delivering higher detection rates for lower screen positive rates. Therefore, women who are found to be 'lower chance' through testing in either first or second trimesters, or who have declined screening for Down's syndrome, Edwards' syndromes and Patau's syndrome should not be referred for further assessment of chromosomal anomaly even if normal variants such as the examples below (whether one or more are identified) are seen at the 18⁺⁰ to 20⁺⁶ week fetal anomaly screening scan. The term ultrasound "soft marker" should no longer be used:

1. Choroid plexus cyst(s).
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 and 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 (AP measurement greater than 7 mm).
5. Small measurements compared to dating scan (significantly less than 5th centile on national charts).

Image capture, storage and archiving

The required images are detailed on the 18⁺⁰ to 20⁺⁶ NHS FASP ultrasound scan base menu (see Appendix 1). Ultrasound images should be captured, stored and archived on an electronic reporting system. There should be a permanent electronic record of all imaging studies. All imaging studies should be accompanied by an electronic report available with the images. Every provider should be able to upload ultrasound scan reports and images on an auditable electronic reporting system in order to provide minimum audit data. All required images should be captured, stored and archived for the purposes of a complete maternal record and to fulfil medico-legal requirements.

Training and professional competence

All ultrasound practitioners must hold minimum certification as specified by NHS FASP in [Service Specification No 17](#).

All providers should have multidisciplinary education and training programmes for health professionals involved in obstetric ultrasound and antenatal screening. All diagnostic ultrasound procedures must be undertaken by health professionals who are fully trained in the use of the specialised equipment and in the safe use of ultrasound.

All practitioners undertaking ultrasound screening should be funded by the provider to attend relevant continuous professional development (CPD) training.

Safety of ultrasound

All health professionals working with ultrasound equipment should be aware of the Royal College of Radiologists (RCR) and Society and College of Radiographer's (SCoR) [standards for the provision of an ultrasound service](#).

All health professionals should adhere to the British Medical Ultrasound Society (BMUS) recommended scanning time limits for obstetric scanning. British Medical Ultrasound Society [Guidelines for the safe use of diagnostic ultrasound equipment 2009](#).

Ultrasound machinery used for the 18⁺⁰ to 20⁺⁶ weeks fetal anomaly scan should be capable of producing images of diagnostic quality and include the following features (as a minimum):

- adequate display/screen size for sufficient clear visualisation
- magnification facility
- cineloop function
- callipers that have a precision to one decimal point (ie 0.1 mm)
- adjustable signal processing facilities
- tissue-specific pre-sets for individual clinical applications
- appropriate probe relevant to gestational age
- doppler and harmonic function

Diagnostic testing

Pregnant women should not be offered a diagnostic test for Down's syndrome, Edwards' syndrome and Patau's syndrome based on their age-related chance alone. Diagnostic testing or invasive prenatal diagnosis (IPD) can include Chorionic Villus Sampling or amniocentesis. The procedure should be performed by specially trained health professionals and women may be required to attend a tertiary centre for the procedure.

Chorionic Villus Sampling (CVS) is an abdominal or sometimes cervical invasive procedure performed under continuous ultrasound guidance. The CVS can be performed from 10 weeks but is usually only performed from 11 weeks of pregnancy to obtain a sample of placental tissue for chromosomal or genetic analysis. Up to 1 out of every 100 women who have a CVS will miscarry.

Amniocentesis is an invasive procedure undertaken from about 15 completed weeks (15⁺⁰) onwards to obtain a sample of amniotic fluid surrounding the fetus. Using an aseptic technique whilst under continuous ultrasound guidance, a sterile needle is passed through the mother's abdomen, uterus and amniotic sac. A sample of amniotic fluid is aspirated and sent for chromosomal or genetic analysis. Up to 1 out of every 100 women who have an amniocentesis will miscarry.

The reason for offering the woman the test should be explained, for example:

- a history of an inherited disorder
- a previous pregnancy or a child with a chromosome disorder
- a raised chance of Down's syndrome and Edwards' syndrome or Patau's syndrome following screening
- suspected anomaly following an ultrasound scan

In twin pregnancies, invasive prenatal diagnosis should be conducted at a tertiary fetal medicine unit due to the specialised nature of the procedures and the increased risk of miscarriage and in line with [Royal College of Obstetrics and Gynaecology](#) and [National Institute for Health and Care Excellence \(NICE\) guidelines](#).

Where the indication for undertaking IPD is a higher chance screening result, the sample is sent to the cytogenetic laboratory for quantitative fluorescence polymerase chain reaction (QF-PCR) testing.

Where the QF-PCR result from a CVS sample indicates that the baby may be affected by Down's syndrome, Edwards' syndrome or Patau's syndrome, the direct culture result should be used to confirm the QF-PCR result prior to any decisions being made

regarding ongoing care or termination of the pregnancy. This is particularly important in cases where the ultrasound shows no fetal anomalies or where the QF-PCR trace is indicative of either an M2 or postzygotic error.

If karyotyping is offered following an anomaly suspected on ultrasound scan, the woman should be informed that subtle chromosomal changes and single gene defects will not normally be detected. The implications of this should be explained, ie not all inherited conditions will be identified. The woman should be informed of the usual reporting times for karyotyping and/or QF-PCR before the procedure.

Results of diagnostic testing

All providers should have a written pathway for communication of results. The process for communicating results should be discussed and agreed with the woman before the procedure. All women must be informed of the CVS or amniocentesis result by an appropriately trained person. When a CVS or amniocentesis is performed at a tertiary centre, that centre should provide written results to the referring clinician. The woman should be informed of the results of diagnostic testing as per local policy.

Audit

Each department performing CVS and amniocentesis procedures should maintain a register of procedures performed and outcome of pregnancy. To facilitate audit, pregnancy outcome forms should be completed and returned to the screening laboratory, or other locally agreed collating centre, at the end of the pregnancy. The provider should develop a written pathway for the completion and return of pregnancy outcome forms to the centre collecting the data.

Patient evaluation of service provision is an integral aspect of overall service audit and should be included as part of the audit and performance management framework. Information should be shared with the [National Congenital Anomaly and Rare Disease Registration Service](#) (NCARDRS) for quality and monitoring of screening programme outcomes.

Non-invasive prenatal testing (NIPT)

During pregnancy, the placenta sheds DNA into the mother`s bloodstream which results in the mother`s blood containing both her own and placental DNA. This is known as total cell free DNA (cfDNA) and in most cases, the placental DNA will be the same as fetal DNA. The total cfDNA is extracted from a maternal blood sample, and sequenced and counted. This sequence is then compared to a reference range to see if any DNA is over represented for chromosomes 21, 18 or 13. An overrepresentation of these

chromosomes means that there is a higher chance of the fetus being affected by Down's syndrome, Edwards' syndrome or Patau's syndrome.

Current availability of NIPT

The UK NSC has recommended introducing NIPT into the NHS FASP screening pathway for with Down's syndrome, Edwards' syndrome and Patau's syndrome as an evaluative roll out. This means that any necessary changes can be made in a timely fashion. NIPT will be an additional option for those women who have a higher chance (1 in 2 to 1 in 150) of having a baby with Down's syndrome, Edwards' syndrome and Patau's syndrome following first trimester combined or second trimester quadruple screening (singleton pregnancies only). Planning is underway with a view to implement the offer of NIPT as an additional option in the current screening pathway during 2018 to 2019.

Quality assurance

Each NHS screening programme has a defined set of **standards** that providers have to meet to ensure that local programmes are safe and effective. Quality assurance (QA) is the process of checking that these standards are met and encouraging continuous improvement and includes:

- advising on the development of national quality standards
- monitoring of how services meet (or fail to meet) standards
- providing expert screening advice for incident management
- facilitating quality review of services, including peer advice
- supporting on a day-to-day basis, those involved in commissioning or providing screening services

QA covers the entire screening pathway; from identifying who is eligible to be invited to screening, through to referral and treatment where required/appropriate.

The aim of QA is to maintain minimum standards and drive continuous improvement in the performance of all aspects of screening to ensure that all women and their babies have access to high quality screening wherever they live. QA is essential in order to minimise harm and maximise benefits of screening.

Formal QA visits to local screening programmes provide the forum for a peer review of the whole multidisciplinary screening pathway, and an assessment of the effectiveness of team working within the local screening programme and associated referral sites:

- regional teams advise providers and commissioners about reducing risks in local screening programmes
- they assess the robustness of local arrangements through audit, as part of peer review and in the investigation of any incidents as they occur
- they act as a conduit for information and dialogue at national, regional and local levels, additionally sharing good practice
- participation in a formal process of QA is the responsibility of each local screening programme
- the performance of the local programmes is monitored in a variety of ways such as review of statistics, regional meetings or informal visits, all of which offer a valuable insight into the activity of a local programme

Key performance indicators

Key performance indicators (KPIs) for the NHS screening programmes were introduced to provide a way of measuring how well the screening programmes are doing in important areas. They contribute to the quality assurance of screening programmes but are not, in themselves, sufficient to quality assure or performance manage screening services. They help local screening services to identify potential problems. So they can be put right and have led to changes in practice and implementation of measures to prevent errors occurring in the screening pathway.

There are currently 3 KPIs for the fetal anomaly screening programme. **KPI FA1** relates to the completion of the request form to ensure all required information is available at the point of calculation of the screening risk to prevent delays in screening and reduce inaccuracies in screening results.

KPI FA2 is a measure of coverage of the 18⁺⁰ to 20⁺⁶ week scan. This is a measure to make sure that all women who have been offered and accepted screening have completed screening by 23⁺⁰ weeks of pregnancy.

KPI FA3 is a measure of coverage of Down's syndrome, Edwards' syndrome and Patau's syndrome screening. This is a measure to make sure screening is offered to everyone who is eligible and each individual who chooses to accept screening has a conclusive screening result.

Screening safety incidents

A screening safety incident is any unintended or unexpected incident(s) that could have or did lead to harm to one or more persons who are eligible for NHS screening; or to staff working in the screening programme.

A screening safety incident can affect populations as well as individuals. It is an actual or possible failure in the screening pathway and at the interface between screening and the next stage of care. Although the level of risk to an individual in an incident may be low, because of the large numbers of people offered screening, this may equate to a high corporate risk. It is important to ensure that there is a proportionate response based on an accurate investigation and assessment of the risk of harm. Due to the public interest in screening, the likelihood of adverse media coverage with resulting public concern is high even if no harm occurs.

Find more information and guidance [about managing screening safety incidents](#) here.

Glossary

Amniocentesis

An invasive procedure undertaken from about 15 completed weeks (15⁺⁰) onwards to obtain a sample of amniotic fluid (liquor) surrounding the fetus. Using an aseptic technique whilst under continuous ultrasound guidance, a sterile needle is passed through the mother's abdomen, uterus and amniotic sac. A sample of amniotic fluid is aspirated with a syringe and sent for analysis to test for a range of chromosomal and inherited disorders. Up to 1 out of every 100 women who have an amniocentesis will miscarry.

Amniotic fluid

Also known as 'liquor', this is the fluid surrounding the fetus during pregnancy. It contains substances and cells from the fetus, which can be removed by amniocentesis and examined.

Anomaly

An aberration or change often used related to a gene or physical structure that may or may not result in a disease or condition.

Biochemical markers

Analytes (commonly referred to as markers) measured by the laboratory that are used to calculate the likelihood of a pregnancy being affected by a condition or syndrome.

Chance

The likelihood that an event will occur.

Chance cut-off

Determines those women who are in the 'higher chance' group and considered 'screen positive'.

Chorionic Villus Sampling (CVS)

An abdominal or cervical procedure performed under continuous ultrasound guidance after 10 completed weeks in pregnancy to obtain a sample of placental tissue for chromosomal or genetic analysis. The range of chromosomal and genetic conditions that can be detected is similar to those for amniocentesis. Up to 1 out of every 100 women who have a CVS will miscarry.

Combined test

Between 11⁺² weeks and 14⁺¹ weeks of pregnancy, a combination of the nuchal scan 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 with Down's syndrome.

Crown rump length (CRL)

Ultrasound measurement between the top of the head (crown) to the bottom of the buttocks (rump). To be eligible for first trimester combined screening as part of the NHS screening programme the CRL should measure between 45.0mm and 84.0mm.

Detection rate

The proportion of affected individuals with a positive screening result.

Diagnostic test

Refers to the process involved in obtaining a definite diagnosis. For example, the diagnostic test on an amniocentesis sample (invasive procedure) is the full karyotype or QF-PCR.

Down's Syndrome (trisomy 21)

Down's syndrome is caused by an extra copy of chromosome 21 in all or some cells of the body.

A person with Down's syndrome will have some level of learning disability. This means they will find it harder than most people to understand and to learn new things. They may have communication challenges and difficulty managing some everyday tasks. People with Down's syndrome have distinctive facial features but they do not all look the same.

Some health problems are more common in people with Down's syndrome. These include heart conditions and problems with hearing and vision. Many health problems can be treated but unfortunately around 5% of babies will not live past their first birthday.

Edwards' Syndrome (trisomy 18)

Babies with Edwards' syndrome have an extra copy of chromosome 18 in all or some cells. A condition caused by the presence of an extra copy (3 instead of 2) of chromosome 18.

Sadly the survival rates are low and of those babies born alive only around 10% live past their first birthday. Some babies may survive to adulthood but this is rare.

All babies born with Edwards' syndrome will have a learning disability and a wide range of physical challenges, which can be extremely serious. They may have problems with

their heart, limbs, kidneys and digestive system. Babies with Edwards' syndrome may have a low birthweight.

Fetal anomaly

Structural abnormalities with how the fetus has developed.

Fetal anomaly ultrasound scan

A detailed ultrasound scan, sometimes called the mid-pregnancy or 20-week scan. It is a screening test offered to all pregnant women and is usually carried out between 18 and 21 weeks of pregnancy. It produces a 2-dimensional black and white image that gives only a side view of the baby and it checks for major physical abnormalities in the baby; although it can't pick up every abnormality.

Gestational age

The duration of an ongoing or completed pregnancy, measured from the first day of the last menstrual period (usually about 2 weeks longer than that measured from conception). Gestational age is usually measured in weeks and days.

Invasive diagnostic procedure

A method used to obtain a sample used to aid diagnosis, for example, amniocentesis or chorionic villus sampling.

Marker

An identifiable physical location on a chromosome whose inheritance can be monitored. Markers can be expressed regions of DNA (genes) or some segment of DNA with no known coding function but whose pattern of inheritance can be determined.

Nuchal scan (Nuchal translucency scan NT)

Between 11⁺² weeks and 14⁺¹ weeks of pregnancy the thickness of fluid in the tissue space within the nape of the fetal neck, the nuchal translucency, can be 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 between 1 in 2 and 1 in 150 a diagnostic test, such as CVS, will be offered.

Patau's Syndrome (trisomy 13)

Babies with Patau's syndrome have an extra copy of chromosome 13 in all or some cells.

Sadly the survival rates are low and of those babies born alive only around 10% live past their first birthday. Some babies may survive to adulthood but this is rare.

All babies born with Patau's syndrome will have a learning disability and a wide range of physical challenges, which can be extremely serious. They may have problems with their heart, limbs, kidneys and digestive system. Around half of babies with Patau's syndrome will have a cleft lip and palate. Babies with Patau's syndrome may have a low birthweight.

Prenatal

Relating to the period before birth.

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 (QA)

A system for monitoring and maintaining high standards in every aspect of a screening programme.

Screening

Testing people who do not have or have not recognised the signs or symptoms of the condition being tested for. Either with the aim of reducing risk of an adverse outcome, or with the aim of giving information about risk.

Screening pathway

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.

Screen positive rate (SPR)

The number of women who receive a higher chance result.

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 safety incident

An unintended or unexpected incident(s) that could have or did lead to harm to one or more persons who are eligible for NHS screening; or to staff working in the screening programme.

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 chance groups.

Syndrome

Combination of symptoms and signs grouped together to form a disorder.

Throughput

Number of samples undertaken per cycle.

Trisomy

3 copies of a particular chromosome rather than 2.

Ultrasound scan

An ultrasound scan is a safe and painless test that uses sound waves to make images. It is like radar.

Appendix 1

18⁺⁰ to 20⁺⁶ FASP ultrasound scan base menu

Structure/Area	Detail	Fetal Measurements*	Images/measurements to capture/archive
Head and neck <ul style="list-style-type: none"> • Skull • Brain • Neck 	Head shape	*Head circumference (HC)	Yes, to include HC measurement, CSP, posterior horn and measurement of the ventricular atrium at the level of the glomus of the choroid plexus
	Cavum septum pellucidum (CSP)	Measurement not required	
	Ventricular Atrium (VA)	*Atrium of the lateral Ventricle	
	Cerebellum	*Transcerebellar diameter (TCD)	Yes, to include measurement of the TCD in the suboccipitobregmatic view
	Nuchal Fold (NF) Measure if appears large	Distance between the outer border of the occipital bone and the outer skin edge	Yes, if measurement ≥ 6mm
<ul style="list-style-type: none"> • Facial Features 	Coronal view of lips and nasal tip	Measurement not required	Yes
<ul style="list-style-type: none"> • Lungs • Heart 	Visceral situs/laterality of heart	Measurement not required	No
	a) Four chamber view (FCV)		
	b) Aorta (Ao) arising from left ventricle		No
	c) Pulmonary artery (PA) arising from right ventricle, or the 3 vessel view (3VV)		No
	d) 3 vessel and trachea view (3VT)		No

Structure/Area	Detail	Fetal Measurements*	Images/measurements to capture/archive
Abdominal content	Stomach and position	Measurement not required *Abdominal circumference (AC)	Yes
	Short intra-hepatic section of the umbilical vein (UV)		
	Abdominal wall and cord insertion		
	Diaphragm	Measurement not required	
	Kidneys Measure AP renal pelvis diameter if it appears large	Measurement not required unless renal pelvis AP diameter >7mm	Yes, if AP renal pelvis diameter measures >7mm
	Bladder	Measurement not required	
Spine • Cervical • Thoracic • Lumbar • Sacral	Vertebrae Skin covering	Measurement not required	Yes, image either sagittal or coronal plane
Limbs • Upper and lower	Femur, tibia and fibula (both legs)	*Femur length	Yes, image and measure a single femur only
	Metatarsals (both feet)	Digit count not required	
	Radius, ulna, humerus (both arms)	Measurement not required	
	Metacarpals (both hands)	Digit count not required	
Uterine cavity • Uterine content	Placenta	According to local policy/protocol	
	Amniotic fluid	According to local policy/protocol	

Appendix 2. Ultrasound images and schematics

Head circumference (HC) and ventricular atrium (VA).