Fetal anomaly screening – clinical referral pathways and informed consent

This article describes the latest guidance on consent around the offer of screening tests for Down's syndrome and fetal anomaly and gives an update on the development of clinical referral pathways for neural tube defects, congenital diaphragmatic hernia and cleft lip.

Alissa Delbarre
Programme Associate
NHS Fetal Anomaly Screening Programme
UK National Screening Committee
University of Exeter
alissa.delbarre@ansnsc.co.uk

Keywords

screening; fetal anomaly; consent, Fetal Anomaly Screening Programme; congenital diaphragmatic hernia; cleft lip; neural tube defects, clinical referral pathways

Key points

Delbarre A. Fetal anomaly screening – clinical referral pathways and informed consent. *Infant* 2012; 8(1): 22-25.

1. The NHS Fetal Anomaly Screening Programme (FASP) has published national standards and guidance for England pertaining to the 18^{+0} to 20^{+6} week fetal anomaly scan.
2. There are 11 conditions to be screened for; each has a detection rate which should be achieved.
3. The NHS FASP has been given the remit to develop clinical referral pathways for the 11 conditions screened for at the 18^{+0} to 20^{+6} week fetal anomaly ultrasound scan.
4. A pathway for neural tube defects (NTDs) has already been developed and pathways for congenital diaphragmatic hernia (CDH) and cleft lip will be available soon.
5. Comprehensive and up-to-date guidance for the service on consent for fetal anomaly ultrasound screening is now available.

The NHS Fetal Anomaly Screening Programme (NHS FASP) includes two national screening programmes, the Down's syndrome and the Fetal Anomaly Ultrasound Programmes.

The NHS FASP is part of the overall structure of the UK National Screening Programmes Directorate of the NHS. The remit and aim of the programme is to set standards and oversee the implementation of a screening programme which conforms to an agreed level of quality for all pregnant women in England.

The main aims and objectives of the programme are to:

- Ensure access to a uniform screening programme which conforms to an agreed level of quality.
- Provide information for women so they are able to exercise informed choice.
- Offer options to women and their partners about the management of their pregnancy.

The screening offered is for certain chromosomal and structural abnormalities and includes an ultrasound scan during the early stages of pregnancy and the 18^{+0} to 20^{+6} week fetal anomaly scan.

Screening is optional and the NHS FASP has strived to ensure pregnant women are provided with information to exercise informed choice and are able to access a uniform screening programme which conforms to an agreed level of quality.

NHS FASP programme structure

The availability and remit of the NHS FASP across the UK is shown in **FIGURE 1**.

Fetal anomaly screening

The NHS FASP has published national standards and guidance for England pertaining to the 18^{+0} to 20^{+6} week fetal anomaly scan[1]. There are 11 conditions to be screened for (**TABLE 1**); each has a detection rate which should be achieved.

The standards document provides 'a framework and context with which to work, setting out the key tasks for standardising sonographic practice and professional partnership working both within and beyond the ultrasound department'[1].

After the screening tests

The NHS FASP has been given the remit to develop clinical referral pathways for the 11 conditions screened for at the 18^{+0} to 20^{+6} week fetal anomaly ultrasound scan[1]. During 2010/2011 the NHS FASP has been collaboratively working with support groups and professional colleges to produce these pathways. So far a clinical referral pathway for neural tube defects (NTDs) has been developed and clinical referral pathways for congenital diaphragmatic hernia (CDH) and cleft lip are in the process of being developed.

Clearly FASP, although facilitating discussions to enable a pathway to be formulated, does not see itself as the lead in this area. Therefore, it is imperative that all stakeholders have an equal voice in its development and direction.

Clinical referral pathways for neural tube defects

The NHS FASP began working with Shine (formerly ASBAH – Association for Spina Bifida and Hydrocephalus) in July 2009 to discuss and review service provision for women and families whose unborn baby is diagnosed with a neural tube defect. In November 2009, NHS FASP and ASBAH jointly hosted an initial meeting to explore and discuss possible ways in which the management of neural tube defects (NTD) could be streamlined across all maternity and paediatric Trusts in the future. The meeting gathered together professionals from a wide range of specialisms in the UK.

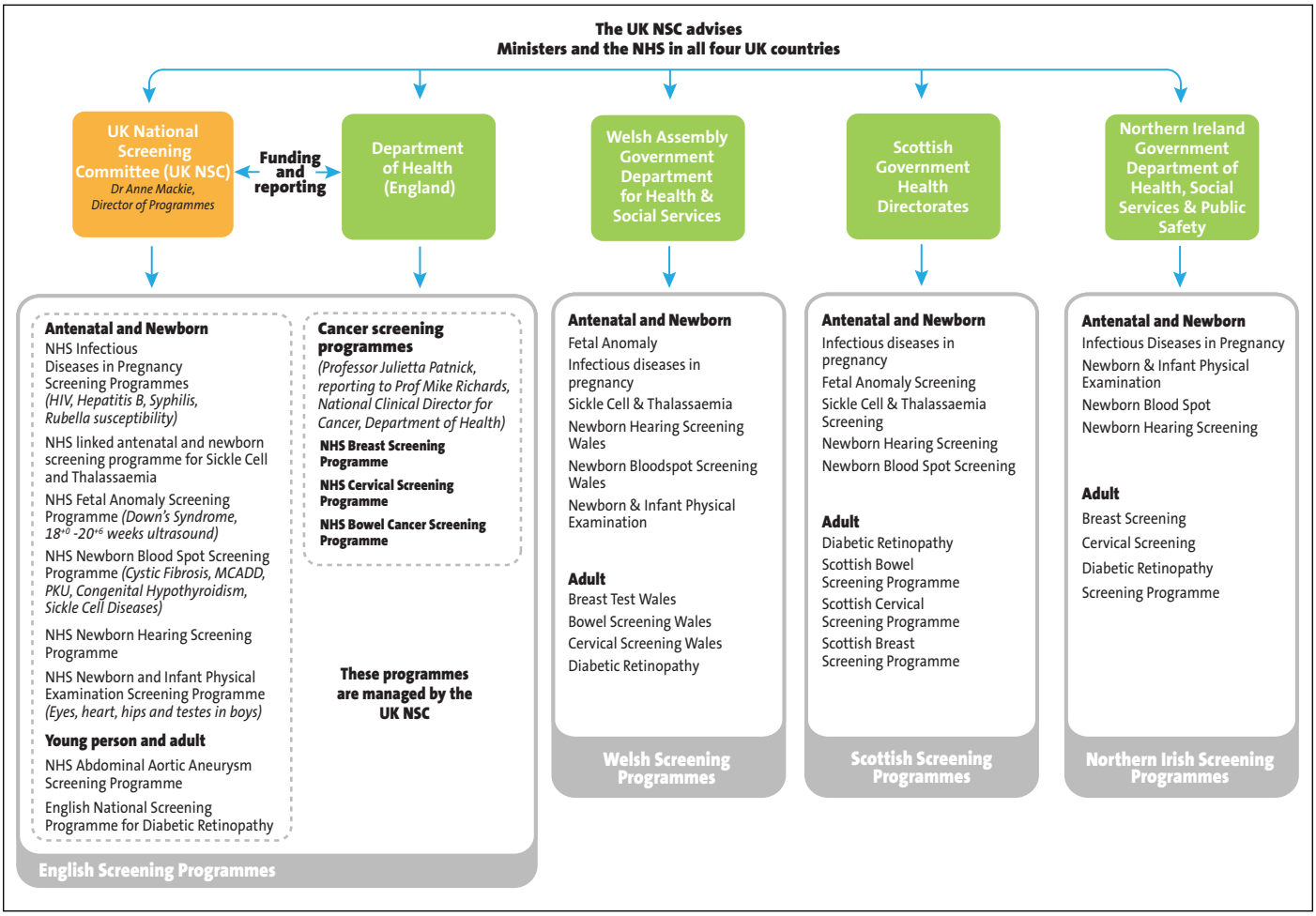

FIGURE 1 Availability and remit of the NHS FASP across the UK.

The discussion and feedback provided was drafted into a meeting report and subsequently the sequence of events for an NTD-affected pregnancy was mapped from screening to paediatric care and these have been illustrated in two separate pathways. The pathways were published in 2011 and distributed to all ultrasound departments in England[2,3].

Clinical referral pathway for CDH

The NHS FASP aims to produce a national clinical care pathway for health professionals, to guide the care of women and families whose unborn baby is diagnosed with CDH. The pathway will be developed in close collaboration with specialised health professionals in this area of care and will commence from detection through to the first year of life.

The first of the meetings to develop the clinical care pathway was held in Birmingham in October 2011. The meeting saw a broad range of health professionals involved all along the clinical referral pathway of care for parents of babies diagnosed with CDH. Health professionals invited to this meeting included fetal medicine specialists, paediatrics surgeons, nurses, midwives and screening coordinators as well as representatives from various support organisations.

The stakeholders all agreed that a generic pathway outlining the care and support that parents should be offered prenatally and postnatally was needed. While the group acknowledged that there is a lack of evidence to support best prognostic method prenatally to predict postnatal outcome, and insufficient evidence in a lot of cases to indicate the most effective treatment protocol, the group recommended an evidence-based prenatal and postnatal pathway could still be developed.

A final report has yet to be published outlining the main points discussed. It should be stressed that these are discussion points at the moment and are not an indication of standards. However the main comments from the meeting can be summarised as follows:

- All suspected cases of CDH should be referred to a fetal medicine unit and all pregnant women with a diagnosis of CDH should be referred to a regional centre in their area.
- Women should be seen within three days for specialist referral.
- Better information is needed for women and their partners. In particular, information about alternative therapies such as fetoscopic tracheal occlusion (FETO) should be given to women during counselling as the support group present at the

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

TABLE 1 The 11 conditions screened for at the ultrasound stage.

FIGURE 2 Woman discussing the 18^{+0} to 20^{+6} week fetal anomaly scan with the sonographer.

meeting clearly indicated that women were well aware of these procedures and wanted to be given information about them, even if their unborn baby was not suitable for the procedure.

- Whole multidisciplinary team (MDT) meetings are needed with parents following a diagnosis to ensure the best care for parents and their babies.
- Good communication between all health professionals involved in antenatal and postnatal care of babies diagnosed and born with CDH and parents is essential.
- Parents should have a constant link person in the antenatal and postnatal periods whom they can contact and speak to about the care and treatment of their baby.
- All babies with CDH should be born in a tertiary centre.
- Every tertiary centre should have a standardised protocol in place for delivering babies with CDH.
- There should be a CDH anomaly register to record patient information and outcomes as concerns were raised that there was no system in the UK of registering CDH cases like the system currently in place in the USA (which certain UK hospitals feed into currently).
- All areas should have a regional network that meets annually to compare practice.

The draft pathways should be available for wider consultation on the NHS FASP website in the coming months.

Clinical referral pathway for cleft lip

The first stakeholder meeting for the development of a cleft lip clinical referral pathway took place in December 2011 to develop a pathway following the previous process explained for CDH. An official report from this initial meeting will be published early in 2012 with any pathways being available for consultation in the next 6-9 months.

Consent standards to support clinical pathways for the NHS FASP

Facilitating informed choice and obtaining informed consent are integral to these pathways and essential aspects of the screening process. To do so is a professional obligation and a matter of basic respect between health professionals and pregnant women. Making a choice and giving consent are part of an ongoing process not a one off event and with such an integral role in this process, it is vital that health professionals such as midwives are aware and supported by the newly updated NHS FASP consent standards and guidance.

The NHS FASP has developed new guidance around the offer of screening tests for Down's syndrome and fetal anomaly[4]. These standards and guidance are intended to maintain and improve the quality of the screening and testing process. They are also intended to enable women to exercise informed choice about any decisions they make.

A review of the consent standards was initiated and undertaken in June 2010. The main outcome of this review has been to provide comprehensive and up-to-date guidance for the service on consent for fetal anomaly ultrasound screening. It is underpinned by robust rationale and evidence and highlights the requirements (including documentation and process) from the service around consent.

The four new standards are:

1 All hospital trusts must have a care pathway to provide evidence that the UK National Screening Committee (UK NSC) and NHS FASP information booklet and leaflets are being used.
2 All pregnant women must be offered up-to-date information on fetal anomaly screening based on the current available evidence. The NHS FASP recommends the use of the National Screening Committee leaflet entitled 'Screening Tests for you and your baby', available on

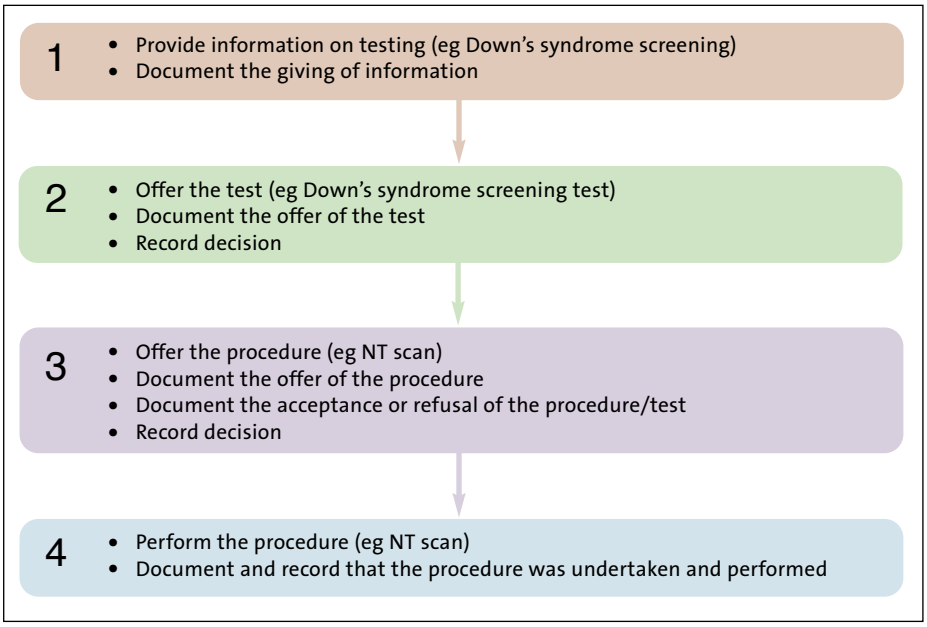

FIGURE 3 Steps to obtaining valid consent[5].

the NHS FASP website: www.fetalanomaly.screening.nhs.uk. Ordering details feature on the back of the leaflet.

3 All eligible pregnant women must be offered 'testing' and this offer must be recorded in the woman's notes and/or hospital information system at the booking appointment.

4 All decisions about the test itself must be recorded in the woman's hand-held notes and/or in the hospital information system.

The main principles outlined in this standards document focus on the fact that there are four main steps to obtaining valid consent. These are outlined in **FIGURE 3**.

This guidance is designed to provide more comprehensive and up-to-date guidance for the health service on consent for Down's syndrome and fetal anomaly ultrasound screening.

Fetal anomaly condition leaflets

Once a diagnosis of one of the 11 previously mentioned conditions has been made, either at the ultrasound scan or after a diagnostic procedure, the NHS FASP has developed leaflets to help inform and support women and professionals.

The NHS FASP initially developed 22 condition leaflets in 2009. The health professional leaflets are produced to support health professionals involved in counselling pregnant women and their partners when a suspected or confirmed fetal anomaly diagnosis has been made following an ultrasound scan.

The parent leaflets are intended for units to give to patients before they see a subspecialist. These are not intended to provide in-depth information on the fetal anomaly but to be used in conjunction with other supporting materials produced by the NHS FASP, the units themselves and information provided by the specialist during the counselling appointment.

A working group was set up in 2011 to review these leaflets and bring them up-to-date with relevant clinical developments. All of the leaflets and additional supporting information will be printed in a spiral bound folder in early 2012 and two copies will be sent to every ultrasound department in England. We recommend that units keep this reference book in their ultrasound department and photocopy the appropriate leaflets for women. Individuals may download and print additional copies from the NHS FASP website. Health professionals may also directly refer parents to the website.

The NHS FASP leaflets have received endorsement from the following colleges and organisations:

- British Maternal & Fetal Medicine Society (BMFMS)
- Royal College of Midwives (RCM)

The NHS FASP is also very grateful to have received the formal support of the following colleges:

- Royal College of Obstetricians and Gynaecologists (RCOG)
- Royal College of General Practitioners (RCGP)

The parent leaflets have received a crystal mark from the Plain English Campaign and will all possess the Plain English logo.

Supporting information for health professionals and parents

A tear-off pad and poster are available to inform women of the purpose of the early scan and 18^{+0} to 20^{+6} weeks fetal anomaly ultrasound scan, from the NHS FASP website www.fetalanomaly.screening.nhs.uk/educationalresources.

For more information on fetal anomaly and the conditions that can be scanned for at the ultrasound stage, parents should consult the following two publications: *"Testing for Down's syndrome during pregnancy"* and *"Having a mid-pregnancy ultrasound scan"*. These leaflets, produced by the NHS FASP, give women and their partners information about the two ultrasound scans which are offered during pregnancy in their local hospital.

If a problem is suspected at the ultrasound scan or as a result of the biochemistry analysis, the woman may be offered an invasive diagnostic test such as chorionic villus sampling (CVS) or amniocentesis. More information for health professionals not usually involved in CVS or amniocentesis and for women being offered CVS or amniocentesis has been produced by the NHS FASP and can be found on the website. The leaflets are:

- *Information for pregnant women about the amniocentesis test.*
- *Information for pregnant women about chorionic villus sampling (CVS).*
- *Chorionic villus sampling (CVS) and amniocentesis: information sheet for health professionals.*

References

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