

Draft Clinical Standards ~ *August 2004*

Pregnancy and Newborn Screening

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1. Introduction

This document introduces the NHS Quality Improvement Scotland (NHS QIS) *Draft Clinical Standards for Pregnancy and Newborn Screening*. These draft standards apply to specific elements of the service. They include sections on:

- Policy;
- Newborn Screen-Rescreen Arrangements;
- Information and Support;
- Education and Training;
- Audit;
- Laboratory; and
- Child Health Registration and Screening.

When finalised, the standards will be used by NHS QIS to assess performance in these areas in NHS Boards throughout Scotland where pregnancy and newborn screening services are provided.

The initial sections of this document provide background information on NHS QIS and on the process used to develop the draft standards (Sections 2 and 3 respectively).

The development of the *Draft Clinical Standards for Pregnancy and Newborn Screening* is outlined in Section 4, and the membership of the Project Group undertaking this work is given in Section 5. The overarching principles guiding development of the draft standards are provided in Section 6.

Section 7 provides basic information about pregnancy and newborn screening, and the evidence underpinning the draft standards is presented in Section 8.

Section 9 contains the *Draft Clinical Standards for Pregnancy and Newborn Screening*.

Finally, Section 10 contains an appendix detailing the most appropriate timescales for pregnancy screening tests and Section 11 provides a glossary of terms used in the draft standards.

2. Background on NHS Quality Improvement Scotland

NHS Quality Improvement Scotland (NHS QIS) was set up by the Scottish Parliament in 2003 to take the lead in improving the quality of care and treatment delivered by NHSScotland. NHS QIS does this by setting standards and monitoring performance, and by providing NHSScotland with advice, guidance and support on effective clinical practice and service improvements.

A part of this remit is to develop and run a national system of quality assurance of clinical services. Working in partnership with healthcare professionals and members of the public, NHS QIS sets standards for clinical services, assesses performance throughout NHSScotland against these standards, and publishes the findings. The standards are based on the patient's journey as he or she moves through different parts of the health service. A wide range of diseases and services are at present being addressed, including stroke services, anaesthesia services and learning disabilities.

Project Groups

For each service in the work programme, NHS QIS appoints a project group comprising appropriate healthcare professionals and members of the public to:

- oversee the development of, and consultation on, the standards; and
- recommend an external peer review process.

Development of Standards

The way in which standards are developed is a key element of the quality assurance process. Groups working on behalf of NHS QIS are expected to:

- adopt an open and inclusive process involving a wide range of both members of the public and professional people through a variety of mechanisms;
- work within NHS QIS policies and procedures; and
- test standards through undertaking pilot reviews to ensure that they meet the principles of NHS QIS.

In addition to standards for specific services or conditions, healthcare governance standards have been set which apply to all clinical services. (These standards have been revised and are currently being consulted on.)

Review

The framework for the NHS QIS review process is as follows:

- once the standards have been finalised, each relevant NHS Board/service is asked to undertake a self-assessment of its service against the standards;
- a review team visits the NHS Board/service on behalf of NHS QIS to follow up this self-assessment exercise with an external peer review of performance in relation to the standards; and
- NHS QIS reports the findings for the NHS Board/service, based on the self-assessment exercise and on the external peer review.

Peer review teams are multidisciplinary, including both healthcare professionals and members of the public. All teams are led by an experienced clinician and are supported by staff from NHS QIS.

All the processes being developed are subject to review and evaluation, and this will help NHS QIS improve its quality assurance system.

Further Information

For further information about NHS QIS, or to obtain additional copies of these draft standards, please contact:

NHS Quality Improvement Scotland
Edinburgh Office
Elliott House
8-10 Hillside Crescent
Edinburgh
EH7 5EA

Tel: 0131 623 4300

Fax: 0131 623 4299

publications@nhshealthquality.org
www.nhshealthquality.org

Copies of all NHS QIS publications can also be downloaded from the website (www.nhshealthquality.org).

3. Background on Clinical Standards – Basic Principles

The standards set by NHS Quality Improvement Scotland (NHS QIS) are:

- focused on clinical issues and include non-clinical factors that impact on the quality of care;
- written in simple language;
- based on evidence (recognising that levels and types of evidence will vary);
- written to take into account other recognised standards and clinical guidelines;
- clear and measurable;
- achievable but stretching;
- developed by healthcare professionals and members of the public;
- consulted on widely;
- published on paper and electronically (on the Internet); and
- regularly reviewed and revised to make sure they remain relevant and up to date.

Some standards are common to all clinical services, others specific to particular conditions.

Format of Standards and Definition of Terminology

All standards set by NHS QIS follow the same format:

- each standard has a **title**, which summarises the area on which that standard focuses;
- this is followed by the **standard statement**, which explains the level of performance to be achieved;
- the **rationale** section provides the reasons why the standard is considered to be important; and
- the standard statement is expanded in the section headed **criteria**, which states exactly what must be achieved for the standard to be reached.

As already mentioned, NHS QIS aims to set standards that are **achievable but stretching**. This is reflected in the criteria. Most criteria are **essential**, in that it is expected that they will be met wherever a service is provided.

Other criteria are **desirable**, in that they are being met in some parts of the service and demonstrate levels of quality which other providers of a similar service should strive to achieve. Each project group is responsible for determining which criteria are essential and which are desirable.

The criteria are numbered for the sole reason of making the document easier to work with, particularly for the assessment process. The numbering of the criteria is not a reflection of priority. The distinction between 'essential' and 'desirable' is the only way in which criteria have been prioritised.

4. Development of the Draft Clinical Standards for Pregnancy and Newborn Screening

Background to the Development of Pregnancy and Newborn Screening Programmes

Antenatal screening for neural tube defects began in Scotland in 1975 and screening for Down's syndrome in 1987. Newborn screening for phenylketonuria was introduced in 1965, congenital hypothyroidism in 1979 and cystic fibrosis in 2003. Although initially there was inequality in the provision of these services and variation in the type and quality of services provided, screening in pregnancy and in the neonatal period has been steadily developed and is now offered to all women and babies in Scotland. Throughout this period however, there has been no provision of standards or performance targets relating to the overall programme or to specific screening tests.

In February 2001 the Scottish Executive Health Department published *A Framework for Maternity Services in Scotland*. The Framework presents a template for best practice in maternity care in Scotland. One of the principles within the Framework states that:

“A comprehensive antenatal diagnostic and screening service should be available and offered to women in order to detect, where possible, any maternal problems or fetal abnormalities at an early stage.”

Health Department Letters (HDLs) also issued in 2001, recommended that standardised, quality assured and co-ordinated pregnancy and newborn screening programmes should be established with arrangements in place from April 2002. The newborn screening programme was also to be modernised and developed to deliver a more standardised quality assured programme. The subsequent HDL (2001) 51 recommended the introduction of a universal newborn hearing screening programme to detect hearing impairment in babies. Lothian and Tayside NHS Boards were identified as pathfinder sites for newborn hearing screening in preparation for the national roll-out of the programme across Scotland. It is expected that the screening programme for newborn hearing will be rolled out from April 2005.

The Role of Pregnancy and Newborn Screening Standards

The *Draft Clinical Standards for Pregnancy and Newborn Screening* seek to establish quality assurance throughout all aspects of the pregnancy and newborn screening process for mothers and babies. NHS Quality Improvement Scotland will develop a review process to assess the pregnancy and newborn screening programmes against the finalised screening standards. In addition to the role of national co-ordination of

the programmes, the National Services Division, NHS National Services Scotland will be actively involved in work relating to the development of these draft standards and the review process, by providing information on both service developments and national policy on a Scottish and UK basis. As the pregnancy and newborn screening programmes evolve, the standards will be reviewed and updated.

Submitting Your Comments

Responses to the *Draft Clinical Standards for Pregnancy and Newborn Screening* should be submitted by **Friday 22 October 2004**. Please send to:

Joanne Storrar
Project Officer
NHS Quality Improvement Scotland
Glasgow Office
Delta House
50 West Nile Street
Glasgow
G1 2NP

Tel: 0141 225 6866
Fax: 0141 248 3778
Email: joanne.storrar@nhshealthquality.org

5. Membership of the Pregnancy and Newborn Screening Standards Project Group

The membership of the Pregnancy and Newborn Screening Standards Project Group, chaired by Canon Bob Fyffe, Minister, Perth, is presented below:

Name	Title	NHS Board Area/Organisation
Dr David Aitken	Head of Biochemical Genetics, Pregnancy & Newborn Screening	Greater Glasgow
Ms Sally Amor	Specialist in Public Health	Pregnancy & Newborn Screening Co-ordinators Group
Dr Ian Auchterlonie	Consultant Paediatrician	Grampian
Dr Richard Brooker	Consultant Paediatrician	Grampian
Dr Alan Cameron	Obstetrician	Greater Glasgow
Dr Jim Chalmers	Consultant in Public Health Medicine	Information Services, NHS National Services Scotland
Dr Heather Cubie	Consultant Clinical Scientist	Lothian
Dr Malcolm Donaldson	Consultant Paediatrician, Endocrinologist	Greater Glasgow
Dr David Fitzpatrick	Consultant Clinical Geneticist	Lothian
Ms Lyn Hutchison	Project Manager	National Services Division, NHS National Services Scotland
Mrs Anyta Lodge	Lay Representative	Tayside
Dr Sheena MacDonald	General Practitioner	Royal College of General Practitioners
Mrs Joan Mackenzie	Laboratory Newborn Screening Co-ordinator	Greater Glasgow
Dr Ann MacKinnon	Senior Clinical Medical Officer (Paediatric Audiology)	Tayside
Mr Alan MacQueen	Family Officer	National Deaf Children's Society
Mrs Mary Nesbitt	Senior Chief Audiologist (Head of Paediatric Audiology)	Lothian
Dr Andrew Powls	Neonatologist	Greater Glasgow
Mrs Audrey Robertson	Co-ordinator of Newborn Hearing Screening	Lothian
Mrs Lucy Robertson	Midwife Sister	Lanarkshire
Mrs Margaret Rule	Lay Representative	Borders

5. Membership of the Pregnancy and Newborn Screening Standards Project Group

Support from NHS Quality Improvement Scotland is being provided by Mrs Fiona Dagge-Bell (Professional Practice Development Officer), Mr Archie Dalrymple (Project Administrator), Ms Hilary Davison (Team Manager), Ms Clare Echlin (Senior Project Officer), Dr Karen Ritchie (Senior Health Services Researcher), Miss Joanne Storrar (Project Officer) and Mrs Tammy Tinto (Project Assistant).

6. Overarching Principles

As mentioned in Section 2, NHS Quality Improvement Scotland (NHS QIS) has developed healthcare governance standards that underpin all clinical services provided by NHSScotland. Generic standards provide a broad context for all NHS QIS condition-specific standards and are also applicable to the mother's journey through the pregnancy screening programme, a process which involves several groups of healthcare professionals.

The following key points underpin the *Draft Clinical Standards for Pregnancy and Newborn Screening*:

Overarching Principles – Screening

- Screening is a public health service offered to groups of the population to identify risk of a particular condition. Screening tests are not compulsory but are offered to help individuals make informed choices about their health.
- Screening is a two-stage process. Usually, the first-line test indicates only a risk or probability that a particular condition is present. A second, diagnostic test is required for confirmation.

Overarching Principles – Pregnancy Screening

- The aim of screening services in pregnancy is to enable women and their partners to make an informed choice about continuing the pregnancy, or to accept treatment at an early stage when it is likely to be more effective. The emphasis should be on detecting the unborn baby at high risk as early as possible to ensure that the woman and her partner have adequate time and opportunity for discussion with healthcare professionals, who can support them through difficult decisions.
- Relevant information, which outlines the benefits that screening bestows, should be provided in a user-friendly manner so that women and their partners can make an informed choice. Information should be available at appropriate stages in pregnancy.
- The pregnancy screening programme must reach all eligible women irrespective of their status, race or any special needs requirements.
- With regard to confidentiality in pregnancy, the woman is the only individual who has legal rights to consent to the screening test and receive the results.

- Healthcare professionals and support staff at all stages and levels in the screening programme should be trained not only within their own care setting, but should also have an understanding of the wider aspects of the programme.
- A woman's satisfaction with the screening process is enhanced if multidisciplinary team working across all the components of the programme is co-ordinated and monitored by the NHS Boards.

Overarching Principles – Newborn Screening

- Newborn screening involves blood spot and hearing tests offered to all apparently healthy term infants, or pre-term infants, with the possibility of detecting a serious disease before any symptoms are evident. However, no screening test is 100% sensitive and specific.
- The aim is to offer treatment at an early stage when it is likely to be more effective. The emphasis should be on performing high quality blood spot and hearing tests, with rapid reporting, as key components of the programme so that repeat tests are minimised and parents have confidence in the value of the process.
- Relevant information, which outlines the benefits that screening bestows, should be provided in a user-friendly manner so that women and their partners can make an informed choice. Information should be available before the screening test.
- The offer of newborn screening tests must reach all eligible infants irrespective of their parental status, race or any special needs requirements.
- With regard to confidentiality in newborn screening, the woman has legal rights to consent to the screening tests and receive the results. The biological father has legal rights to consent to the screening test and receive the results only if he is married to the mother or has sought legal parental responsibility.
- Staff at all stages and levels in the screening programme should be trained not only within their own setting, but should also have an understanding of the wider aspects of the programme.
- Parental satisfaction is enhanced when there is multidisciplinary team working across all the components of the programme, co-ordinated and monitored by the NHS Boards.

The *Draft Clinical Standards for Pregnancy and Newborn Screening* are evidence based and have been developed and finalised in conjunction with many professional and lay people across Scotland. They should represent what are considered to be the key elements of care an infant and parent(s) receives on the journey through the screening process.

7. An Introduction to Pregnancy and Newborn Screening

Screening is a public health service offered to groups of the population to identify risk of a particular condition. Screening tests are not compulsory but are offered to help individuals make informed choices about their health. There is an ethical obligation on agencies to ensure that the timely provision of services meets the needs identified through the screening process. For example, paediatric audiology services, social work and early education services work with children who have a profound hearing loss identified through the newborn screening service. This ensures that the advantages and health gain from identifying this condition at an early point in time are maximised.

Prior to accepting the offer of a screening test, it is important that individuals receive information about the screening in which they are about to participate. While some screening tests have the potential to save lives, or improve quality of life by making possible the early diagnosis of a serious condition, they are not 100% sensitive and specific.

Pregnancy and newborn screening are considered to be important components of good healthcare that should both underpin and inform child and family health and wellbeing and the provision and design of maternity care and child health services.

Screening is a two-stage process. Usually, the first-line test indicates only a risk or probability that a particular condition is present. A second, diagnostic test is required for confirmation. In some cases, the first test provides information on infection in the mother herself and infection would be confirmed by further tests before action is taken.

Pregnancy Screening

In Scotland all pregnant women are eligible for screening. Information on pregnancy screening is provided to women at an early stage in their pregnancy (ideally before the initial booking visit). A woman's decision to accept or decline the screening tests is recorded in her maternity record, but irrespective of her decision she will continue to receive the full range of normal antenatal care.

Down's syndrome is a congenital condition which is associated with moderate to severe learning disability and a spectrum of other health problems. In the absence of screening and pre-natal diagnosis the current birth incidence in Scotland would be around 15 per 1,000 babies born in Scotland. Older mothers are more likely to conceive a child with Down's syndrome than younger mothers. The screening tests which may detect this condition are a blood test carried out at 15-20 weeks of pregnancy or a combined blood test and ultrasound screening test at 11-14 weeks of

pregnancy. The ultrasound scan measures the thickness of the fluid-filled area at the back of the fetal neck (called 'nuchal translucency' or NT). The bigger the measurement, the higher the risk of Down's syndrome being present. About 1 in 20 women will have a screening result that indicates a higher chance of an affected pregnancy. A diagnostic test, chorionic villus sampling (CVS) or amniocentesis is then offered to provide a definitive diagnosis. The 15-20 week blood test can identify 6 out of 10 pregnancies with Down's syndrome and the combined test 8 out of 10 affected pregnancies.

Neural tube defects, such as spina bifida and anencephaly, affect approximately 1 in every 500-1,000 pregnancies in Scotland. Babies with spina bifida have an opening in the bones of the spine, and the nerves to the lower part of the body are damaged. This can result in difficulties with walking and bowel and bladder control. Sometimes there is also a learning disability due to hydrocephalus (an accumulation of fluid in the brain) which often accompanies spina bifida. There is a wide variation in the level of disability caused by this condition and many people with spina bifida are able to lead fulfilling lives. In babies with anencephaly the skull and brain are not properly formed. These babies generally die before or very soon after they are born.

The screening test involves taking a small blood sample from the woman at approximately 15-16 weeks of pregnancy (usually the same sample that is used for the Down's syndrome test described previously) and the concentration of a substance called alphafetoprotein (AFP) is measured. If the AFP result is normal, there is only a small chance that the baby will have a neural tube defect and no further testing is required. An elevated result, which would be expected in around 1 in 25 women, indicates that there is an increased risk of the baby being born with a neural tube defect and an ultrasound scan will be arranged to show whether or not an abnormality is present. The blood test can detect around 8 out of 10 pregnancies affected with a neural tube defect.

Communicable Diseases

A Health Department Letter (HDL 52) issued in June 2002 recommended that HIV testing should be offered routinely to all pregnant women as part of their antenatal care. This test was introduced in Scotland throughout 2002, with a target for full implementation by April 2003. This test will form part of the integrated programme of antenatal screening to limit risk for a number of communicable diseases (hepatitis B, syphilis and rubella as well as HIV).

Tests for infection with HIV, hepatitis B and syphilis, and immunity to rubella are carried out on serum obtained from a single blood sample taken at the first antenatal visit. Occasionally a second blood sample may be required for technical reasons to confirm a test result. Tests for HIV, syphilis and rubella are based on the detection of a specific antibody to each. The test for hepatitis B virus detects viral antigen in the same sample.

HIV can be transmitted from a mother to her baby. The risk of HIV transmission from a mother to her baby can be up to 25% when no treatment is given. Detecting HIV antibody in the mother's blood at any stage of pregnancy, but preferably early on, can help to bring transmission to under 2% through antiviral treatment and appropriate delivery.

Screening for HIV involves taking a blood sample which is tested for antibodies to HIV. All positive test results are confirmed by retesting, using a different test method. The combination of two tests produces high accuracy with minimal risk of false positive results. HIV antibody tests exceed the performance of most other infectious disease tests in both sensitivity and specificity.

Although these tests are very sensitive, there is an interval known as a 'window period'. This is the period between the onset of infection with HIV and the appearance of detectable antibodies to the virus. This period is usually less than a month, however, it may be longer for some individuals. It is important that women are aware that despite being given a negative test result, it could be that antibodies are not yet present and therefore the result given could be false. If a woman feels she has been/continues to be at risk of exposure to the virus, further 3-monthly testing should be offered throughout pregnancy.

Hepatitis B infection can be transmitted from mother to infant, leading to generally asymptomatic infection but lifelong carriage of the virus and greater risk of cirrhosis of the liver and of hepatocellular carcinoma later in life. Perinatal transmission of the hepatitis B virus occurs if the mother has an acute infection in pregnancy or if she is a chronic carrier of hepatitis B. This can be prevented by identifying a baby at risk before birth and offering immunisation starting in the first 2 days of life.

Syphilis infection can have consequences for both mother and infant, but detection of infection early in pregnancy allows appropriate antibiotic treatment to prevent transmission. About 50% of infected mothers will be asymptomatic and the causal agent, *treponema pallidum* can cross the placenta at any stage of pregnancy. However, maternal antibiotic therapy can prevent nearly all congenital syphilis.

Rubella infection used to be a common mild infection of childhood, and is characterised by a short period of illness and a typical rash. If rubella is contracted during the first trimester of pregnancy it can lead to severe abnormalities in the baby, including deafness and eye and heart disease. The incidence has been largely reduced by immunisation of children and young women before conception, but there is a continuing need to look for evidence of immunity in pregnant women to ensure congenital rubella syndrome is very unlikely in Scotland.

Newborn Screening

A Health Department Letter (HDL 34) issued in April 2001 outlined the National Screening Committee's recommendation for the introduction of a standardised, quality assured co-ordinated programme for newborn screening services for phenylketonuria (PKU) and congenital hypothyroidism (CHT). A subsequent Health Department Letter (HDL 51), issued in June 2001, outlined the National Screening Committee's recommendation for the introduction of a phased neonatal hearing screening programme. A further Health Department Letter (HDL 73), issued in September 2001, endorsed proposals for the introduction of a standardised neonatal cystic fibrosis (CF) screening programme.

Although most babies are perfectly healthy when born, a small number have abnormalities of body chemistry (metabolism) which can lead to problems with growth and development. Some of these problems can be detected through a blood test. This involves taking a small amount of blood from a baby's heel. Midwives usually take blood samples from babies within the first week of life. The blood spot specimen is used to test for three main conditions:

- phenylketonuria
- congenital hypothyroidism
- cystic fibrosis.

Hearing impairment also affects a small number of newborn babies each year and hearing tests have been developed which can help identify affected infants at an early stage.

NHS Boards are responsible for ensuring that all babies are screened and all results are reported to the NHS Board for input onto the department of community child health records system. In addition positive results are reported to the consultant paediatrician/audiologist, GP and the NHS Board who will initiate treatment and/or further testing.

Phenylketonuria affects around 1 in every 8,000 babies born in Scotland. It is a condition that is inherited when both parents are asymptomatic carriers of one active and one inactive copy of the gene. Inheriting two inactive copies of the gene causes deficiency of a liver enzyme known as phenylalanine hydroxylase which, in turn produces high levels of phenylalanine in all body fluids. Babies with PKU cannot metabolise phenylalanine, which is a component of all natural protein in every day foods, eg milk, fish, eggs and cheese. If phenylalanine levels remain high this results in severe damage to the baby's brain. Babies diagnosed with this condition must be seen by a paediatrician as soon as possible and started on a special diet. Therapy for PKU relies on the partial elimination of phenylalanine from the diet. Since phenylalanine is present in all proteins this requires the use of chemically defined protein substitutes with a limited amount of natural protein. The effectiveness of therapy is monitored by regular blood samples which accurately measure the phenylalanine level. With prompt treatment the baby is very likely to develop normally. The treatment is life-long and effective management requires a multidisciplinary team comprising doctors, dieticians, clinical biochemists, psychologists and pharmacists.

Congenital hypothyroidism affects approximately 1 in every 3,500 babies born in Scotland. Babies affected by this condition are unable to produce enough of the hormone thyroxine either because their thyroid gland is missing or it is not working effectively. This condition is corrected by giving thyroxine by mouth, which will help the baby to grow normally. However, if left untreated it will result in slower than normal growth and severe learning difficulties.

Cystic fibrosis is an inherited condition and affects 1 in every 2,500 babies born. The organs most likely to be affected are the pancreas and the lungs, causing poor digestion and chest infections. Screening for CF involves the blood spot specimen being tested in two stages. The first stage tests for a substance called immuno reactive trypsinogen (IRT), which is usually present in increased amounts in the blood of babies with CF during their first few weeks of life. When the IRT is high, deoxyribonucleic acid (DNA) analysis is performed on the same blood spot sample, to look for the commonest genetic mutations associated with CF in the Scottish population. Early treatment may help affected children to maintain good nutrition and minimise chest infections, leading to improved quality of life.

Newborn Hearing Screening

Prevalence of permanent congenital hearing impairment (PCHI) is approximately 1-2 per 1,000 babies born in the UK. This means that approximately 60-65 babies born in Scotland each year are affected. The new Universal Newborn Hearing Screening (UNHS) programme will be more effective in detecting hearing impairment than the current distraction test, which is carried out when the infant is 7-8 months old. The sensitivity of the distraction test is very low and hearing difficulties are often not detected until children are at least 18 months old, and sometimes not until they are 3^{1/2} years old. UNHS will be offered shortly after birth, thus reducing the age at which deafness is confirmed and therefore improving the wellbeing of the child in terms of education and social needs. Positive results are reported to the audiologist.

There are two types of screening tests. The Otoacoustic Emissions (OAE) test involves transmitting sounds to a baby through an ear piece. Response to the sound in each ear is recorded. The Automated Auditory Brainstem Response (AABR) test involves three sensors being placed on a baby's head. Headphones are placed over a baby's ears which transmit clicking sounds to the ear. Again the baby's response to the sound is recorded. Both screening tests are performed while the baby is resting/sleeping. Screening tests do not cause any pain or discomfort to the baby.

8. Evidence Base for the Draft Clinical Standards for Pregnancy and Newborn Screening

Down's Syndrome and Neural Tube Defects

1. Wald N, Kennard A, Hackshaw A, et al. 1998. Antenatal Screening for Down's Syndrome. *Health Technology Assessment*, 2(1): 1-112.
2. Drife J and Donnai D, eds. 1991. *Antenatal Diagnosis of Fetal Abnormalities (Proceedings of RCOG 23rd Study Group, London, 1990)*. London: Springer Verlag.
3. Working Party of the Royal College of Gynaecologists (RCOG). 1993. *Report of RCOG Working Party on Biochemical Markers and the Detection of Down's Syndrome*. London: Royal College of Gynaecologists.
4. Burnett D, Blair C, Haeney M, et al. 2002. Clinical Pathology Accreditation: Standards for the Medical Laboratory. *Journal of Clinical Pathology*, 55(10): 729-733.
5. Aitken D, Crossley J and Spencer K. 2002. Prenatal Screening for Neural Tube Defects and Aneuploidy. in Rimoin D, Connor J, Pyeritz R and Korf B, eds. *Emery and Rimoin's Principles and Practice of Medical Genetics*. 4th ed. London: Churchill Livingstone: 763-801.
6. Balmer S, Bowens A, Bruce E, et al. 2000. *Quality Management for Screening: Report to the National Screening Committee*. Leeds: Nuffield Institute for Health.
www.nuffield.leeds.ac.uk/downloads/screening.pdf [full document] URL accessed 28/06/04.
7. Blackwell Scientific [publisher]. 2001. Special Issue: Informed Choice in Screening. *Health Expectations*, 4(2).
8. Ritchie K, Boynton J, Bradbury I, et al. 2004. *Routine Ultrasound Scanning before 24 Weeks of Pregnancy: Health Technology Assessment Report 5*. Glasgow: NHS Quality Improvement Scotland.
9. National Down's Syndrome Screening Programme for England. 2004. *Antenatal Screening - Working Standards*. Northants: National Down's Syndrome Screening Programme for England.
www.nelh.nhs.uk/screening/dssp/working_standards.pdf [full document] URL accessed 28/06/04.

Universal Newborn Hearing Screening

10. Davis A, Bamford J, Wilson I, et al. 1997. A Critical Review of the Role of Neonatal Hearing Screening in the Detection of Congenital Hearing Impairment. *Health Technology Assessment*, 1(10): i-iv, 1-176.
11. Yoshinaga-Itano C, Sedey A, Coutter D, et al. 1998. Language of Early and Later Deafened Children with Hearing Loss. *Paediatrics*, 102: 1161-1171.

12. Stokes J. 1999. Learning to Listen. *in* Stokes J, ed. *Hearing Impaired Infants: Support in the First 18 Months*. London: Whurr Publishers. p197-230.
13. Clinical Standards Board for Scotland. 2002. *Clinical Standards: Generic*. Edinburgh: Clinical Standards Board for Scotland. www.clinicalstandards.org/pdf/finalstand/generic.pdf [access to full document] URL accessed 28/06/04.
14. Scottish Executive. 2001. *Neonatal Hearing Screening Report*. Edinburgh: Scottish Executive. www.show.scot.nhs.uk/sehd/publications/neonatal/neonatal.htm [access to full document] URL accessed 28/06/04.
15. National Deaf Children's Society. 2000. *Guidelines for the Early Identification and Audiological Management of Children with Hearing Loss*. Quality Standards in Paediatric Audiology, Vol. IV. London: NDCS. www.ndcs.org.uk/information/ndcs_publications/early_ident.html [access to full document] URL accessed 28/06/04.
16. Tyler S and Evans M. 2002. *Quality Assurance Review of the Information Materials Developed for the NHS Newborn Hearing Screening Programme. Report to the Child Subgroup of the National Screening Committee*. www.nhsp.info/protos.shtml [access to full document] URL accessed 28/06/04.
17. Fortnum H, Summerfield A, Marshal D, et al. 2001. Prevalence of Permanent Childhood Hearing Impairment in the United Kingdom and Implications for Universal Neonatal Screening: Questionnaire Based Ascertainment Study. *British Medical Journal*, 323: 536-539.
18. NHS Newborn Hearing Screening Programme. *NHSP Quality Assurance and Management - National Standards: Checklist for Review of Local Performance against National Standards*. www.nhsp.info/documents/qamanagement.shtml [electronic source] URL accessed 28/06/04.
19. Watkin P, Baldwin M, Dixon R, et al. 1998. Maternal Anxiety and Attitudes to Universal Neonatal Hearing Screening. *British Journal of Audiology*, 32(1): 27-37.
20. Weichbold V, Welzl-Mueller K and Mussbacher E. 2001. The Impact of Information on Maternal Attitudes Towards Universal Neonatal Hearing Screening. *British Journal of Audiology*, 35(1): 59-66.
21. Hergilis L and Hergilis A. 2000. Universal Neonatal Hearing Screening - Parental Attitudes and Concerns. *British Journal of Audiology*, 34(6): 321-327.

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22. Cunningham M and Cox E. 2003. Hearing Assessment in Infants and Children: Recommendations Beyond Neonatal Screening. *Paediatrics*, 111(2): 436-440.

Cystic Fibrosis

23. Farrell P, Kosorok M, Rock M, et al. 2001. Early Diagnosis of Cystic Fibrosis through Neonatal Screening Prevents Severe Malnutrition and Improves Long-Term Growth. *Paediatrics*, 107(1): 1-13.
24. Wang S, O'Leary L, FitzSimmons S, et al. 2002. The Impact of Early Cystic Fibrosis Diagnosis on Pulmonary Function in Children. *Journal of Pediatrics*, 141(6): 804-810.

Phenylketonuria

25. Beasley M, Costello P and Smith I. 1994. Outcome of Treatment in Young Adults with Phenylketonuria Detected by Routine Neonatal Screening between 1964 and 1971. *Quarterly Journal of Medicine*, 87(3): 155-160.
26. Smith I, Beasley M and Ades A. 1990. Intelligence and Quality of Dietary Treatment in Phenylketonuria. *Archives of Disease in Childhood*, 65(5): 472-478.
27. Ades A, Walker J, Jones R, et al. 2001. Coverage of Neonatal Screening: Failure of Coverage or Failure of Information Systems. *Archives of Disease in Childhood*, 84(6): 476-479.

Communicable Diseases

28. Scottish Executive Health Department (SEHD). *Offering HIV Testing to Women Receiving Antenatal Care*. NHS HDL(2002)52. Edinburgh: Scottish Executive. www.show.scot.nhs.uk/sehd/mels/HDL2002_52.pdf [full document] URL accessed 28/06/04.
29. Cradock-Watson J. 1991. Laboratory Diagnosis of Rubella: Past, Present and Future. *Epidemiology and Infection*, 107(1): 1-15.
30. Salisbury D and Begg N, eds. 1996. *Immunisation against Infectious Disease*. London: Department of Health; HMSO.
31. Morgan-Capner P and Crowcroft N on behalf of the PHCS Joint Working Party of the Advisory Committees of Virology and Vaccines and Immunisation. 2000. Guidance on the Management of, and Exposure to, Rash Illness in Pregnancy (Including Consideration of Relevant Antibody Screening Programmes in Pregnancy). *Communicable Disease and Public Health*, 5(1): 59-71.

32. Public Health Laboratory Service and Communicable Disease Surveillance Centre with the PHLS Working Group. 1998. *Antenatal Syphilis Screening in the UK: A Systematic Review and National Options Appraisal with Recommendations (Report to the National Screening Committee)*. London: Public Health Laboratory Service.
33. Connor N, Roberts J and Nicholl A. 2000. Strategic Options for Antenatal Screening for Syphilis in the United Kingdom: A Cost Effective Analysis. *Journal of Medical Screening* 7(1): 7-13.
34. Scottish Office Department of Health. 1998. *Screening of Pregnant Women for Hepatitis B, and Immunisation of Babies at Risk*. HSC 1998/127. Edinburgh: Scottish Office Department of Health.

Other References

35. Hall D and Elliman D. 2003. *Health for All Children*. 4th ed. Joint Working Party on Child Health Surveillance. Oxford: Oxford University Press.

9. Draft Clinical Standards for Pregnancy and Newborn Screening

STANDARD 1 – Policy

STANDARD 2 – Newborn Screen-Rescreen Arrangements

STANDARD 3 – Information and Support

STANDARD 4 – Education and Training

STANDARD 5 – Audit

STANDARD 6 – Laboratory

STANDARD 7 – Child Health Registration and Screening

STANDARD 1 ~ Policy

Standard Statement	Rationale
<p>1(a). Effective pregnancy and newborn screening programmes are available and offered in all NHS Board areas.</p>	<p>There is evidence that an effective pregnancy screening programme can identify a high proportion of pregnancies at risk, which facilitates early detection and allows informed choice regarding management.</p> <p>References: (1), (5), (8), (28), (29), (30), (31), (32), (33), (34)</p> <p>There is evidence that an effective newborn screening programme can identify risk of specific disorders, which promotes early detection and treatment management.</p> <p>References: (23), (24), (25), (26)</p> <p>There is evidence that early identification of hearing loss and appropriate intervention by 6 months of age is the most effective strategy for the maximisation of development of language in deaf and hard of hearing infants and toddlers.</p> <p>References: (10), (11), (12)</p>



Criteria
<p>Essential</p> <p>1(a) 1. There is an accredited consultant in public health medicine or accredited specialist in public health at NHS Board level responsible for overseeing and monitoring the provision of effective pregnancy and newborn screening in their area. (Responsibility may be delegated to key staff as appropriate.)</p> <p>1(a) 2. There is a named healthcare professional at an operational level co-ordinating the provision of effective pregnancy and newborn screening.</p> <p>1(a) 3. Every NHS Board has a named multidisciplinary screening co-ordinating group with lay representation that meets regularly and reports annually to the NHS Board.</p> <p>1(a) 4. This group is responsible for ensuring that effective pregnancy and newborn screening is offered and available for all eligible women and babies, in accordance with current national guidance.</p>

STANDARD 1 ~ Policy (continued)

Standard Statement	Rationale
<p>1(b). All NHS Boards have written specifications and protocols available for pregnancy and newborn screening which take account of the national recommendation and local circumstances.</p>	<p>All women and babies should receive the same standard of screening regardless of where they live.</p> <p>References: (2), (3), (6), (7), (8)</p>
<p>1(c). Pregnancy and newborn screening, conforming to the guidelines for the Scottish National Programme, is offered to all women and for their babies.</p>	<p>National guidelines for pregnancy and newborn screening promote equity of provision and care.</p> <p>Conditions screened for are those where testing at an appropriate stage maximises the chance of detection and allows treatment which can improve the long-term outcome and development of affected children.</p> <p>References: (23), (24), (25), (26), (27)</p>

Criteria

Essential

1(b) 1. Written screening specifications and protocols, which take account of national screening recommendations and the local programme, are in place for:

- Down's syndrome;
- neural tube defects;
- HIV;
- rubella;
- syphilis;
- hepatitis B;
- phenylketonuria (PKU);
- congenital hypothyroidism (CHT);
- cystic fibrosis (CF); and
- newborn hearing screening.

1(b) 2. NHS Boards have arrangements in place to ensure that the specification is met and monitored on a regular basis.

Essential

1(c) 1. There is a policy in place to offer all women the following screening tests at the most appropriate stage in their pregnancy:

- Down's syndrome;
- neural tube defects;
- HIV;
- rubella;
- syphilis; and
- hepatitis B.

The policy also includes procedures for follow-up diagnostic testing. (see Appendix 1.)

1(c) 2. Evidence that women have been offered all pregnancy screening tests is documented in the maternity record.

1(c) 3. The following tests are offered to all babies at the most appropriate time after birth:

- PKU between 96 and 168 hours of life;
- CHT between 96 and 168 hours of life;
- CF between 96 and 168 hours of life; and
- newborn hearing screening within the first month of life, unless born prematurely or ill.

1(c) 4. A protocol is in place for screening babies who are ill or born prematurely.

STANDARD 2 ~ Newborn Screen-Rescreen Arrangements

Standard Statement	Rationale
<p>2(a). Effective newborn screen-rescreen arrangements are in place in all NHS Boards.</p>	<p>There is evidence that effective newborn screen-rescreen arrangements, which allow tracking of babies within an NHS Board area and beyond, improves uptake and coverage.</p> <p>Reference: (35)</p>



Criteria
Newborn Hearing Screening Essential 2(a) 1. 95% of infants commence the hearing screening process before 4 weeks of age. 2(a) 2. 95% of infants complete the hearing screening process by 3 months corrected age. 2(a) 3. A local protocol is in place for the management of non-attenders. 2(a) 4. A follow-up protocol is in place, appropriate to the outcome of the screen and any risk factors the baby may have for acquired and progressive hearing loss. Newborn Hearing Screening Desirable 2(a) 5. 95% of infants complete the hearing screening process by 2 months corrected age.

STANDARD 3 ~ Information and Support

Standard Statement	Rationale
<p>3(a). All women/parents/carers receive clear information (written or in other media) to help them to make an informed choice about pregnancy and newborn screening.</p>	<p>Good communication and information throughout pregnancy and after birth reduces anxieties and concerns about a baby's health and wellbeing.</p> <p>References: (3), (7), (8)</p> <p>There is good evidence that providing information about tests and investigations reduces anxiety and encourages participation.</p> <p>References: (19), (20), (21)</p> <p>Good communication before and after screening reduces unnecessary delays, anxieties and concerns.</p> <p>Reference: (13)</p> <p>Parents should be given written information about all newborn screening tests antenatally and given time to reflect on this before making a decision.</p> <p>References: (15), (16)</p>

Criteria

Essential

3(a) 1. All women/parents/carers are provided with information which takes account of their physical, cultural, ethical, educational and mental health needs.

Pregnancy Screening

Essential

3(a) 2. Information provided to women/carers during appropriate antenatal visits conforms to national guidelines and includes:

- information about the screening test and the test method;
- an explanation of the limitations of screening tests;
- the meaning of screening results;
- the options available following results;
- how further information and support can be obtained; and
- information regarding other conditions which the screening process may detect.

3(a) 3. Information is provided to all women at least 24 hours in advance of the screening tests.

3(a) 4. An opportunity to discuss this information is provided.

Newborn Screening

Essential

3(a) 5. Information is provided to all parents/carers at least 24 hours in advance of their baby's screening tests.

3(a) 6. An opportunity to discuss this information is provided.

3(a) 7. The decision to accept or decline testing is recorded on a signed consent form.

3(a) 8. In all cases where blood spot testing has been declined, a card containing the baby's details is marked 'declined' and sent to the laboratory.

3(a) 9. Systems are in place to allow parents/carers, who opted out of screening, to have their baby referred for a screen or appropriate assessment at a later date, if wished.

STANDARD 3 ~ Information and Support (continued)

Standard Statement	Rationale
<p>3(b). There is a clearly defined regional or local policy for informing women/parents/carers of the results of screening tests, which includes the most appropriate methods of communication for the local population.</p>	<p>Women have a right to know how and when they will receive the results of the screening tests.</p> <p>Reference: (2)</p> <p>There is evidence that delayed reporting increases anxiety. To further reduce anxiety it is important that women/parents/carers receive information regarding their result in a format that they can easily understand.</p> <p>Reference: (13)</p> <p>It is essential that the parents/carers and involved professionals are informed about the outcome of the screen as soon as possible in order to put in place any supports that may be required.</p> <p>There is evidence that delays in follow-up testing can increase anxiety. It is important that women/parents/carers receive information regarding follow-up testing.</p>

Criteria
<p>Essential</p> <p>3(b) 1. All women/parents/carers are informed of the timescale within which the results will be made available, and the format in which they will be communicated, and by whom.</p>
<p>Pregnancy Screening</p> <p>Essential</p> <p>3(b) 2. All results are confirmed to the requesting clinician in writing within 15 working days of the screen being performed.</p> <p>3(b) 3. All non-positive results are communicated to the woman by 20 weeks of pregnancy.</p> <p>3(b) 4. All positive results are communicated to the woman within 15 working days of the screen test.</p>
<p>Pregnancy Screening</p> <p>Desirable</p> <p>3(b) 5. All results are communicated in writing to the woman within 15 working days of the screen test.</p>
<p>Newborn Bloodspot Screening</p> <p>Essential</p> <p>3(b) 6. 99.5% of positive results are received by parents/carers 10–14 days from specimen collection.</p> <p>3(b) 7. All non-positive results are available to parents/carers.</p>
<p>Newborn Bloodspot Screening</p> <p>Desirable</p> <p>3(b) 8. All results are communicated in writing to parents/carers.</p>
<p>Newborn Hearing Screening</p> <p>Essential</p> <p>3(b) 9. Parents/carers are informed of the outcome of the screen immediately the screening episode is completed (or in special circumstances, as soon as possible thereafter). For those requiring follow-up, appropriate written and verbal information is provided.</p> <p>3(b) 10. Hearing screen results are forwarded to the GP, health visitor and the relevant child health surveillance system within 7 working days of the screen being undertaken.</p>

STANDARD 3 ~ Information and Support (continued)

Standard Statement	Rationale
<p>3(c). Support is available for all women/parents/carers who have received a 'higher chance' result or positive screening result.</p>	<p>The opportunity to discuss concerns with healthcare professionals can support women/parents/carers through a difficult decision-making process. Positive screening results generate anxiety among carers which can be reduced by appropriate counselling and support.</p> <p>References: (2), (3), (6), (8)</p>



Criteria
Essential
3(c) 1. All women/parents/carers have access to appropriately trained healthcare professionals within the timescales outlined in Appendix 1, to discuss results, treatment options and/or further tests.
3(c) 2. Information is provided to women/parents/carers on local and national support groups for the conditions being screened.

STANDARD 4 ~ Education and Training

Standard Statement	Rationale
<p>4(a). There is training and ongoing education for healthcare professionals involved in pregnancy and newborn screening.</p>	<p>A healthcare organisation requires staff to be qualified and trained to an appropriate standard to meet the needs of the population it serves. Review of individual competencies and continuing professional development (CPD) are essential.</p> <p>Minimum standards can be reproduced more consistently through the use of a standardised training tool. There is evidence that staff with good communication skills who are sensitive to the needs of the parents/carers and have good baby handling skills achieve better screening results.</p> <p>Reference: (35)</p>



Criteria
Essential
4(a) 1. Relevant education and training on pregnancy and newborn screening is available for all new healthcare professionals during induction and annually for existing staff who provide the screening service.
4(a) 2. A record of all training/education sessions is maintained by a named person.
4(a) 3. Local training/education courses are monitored and reviewed by a named person.
4(a) 4. Healthcare professionals contribute to the screening service only after successful completion of training.

STANDARD 5 ~ Audit

Standard Statement	Rationale
<p>5(a). The pregnancy and newborn screening programmes are audited at a local and national (Scottish) level.</p>	<p>Audit is essential to ensure standardised and quality assured pregnancy and newborn screening programmes and to establish the sensitivity and specificity of screening. It also helps to identify variations in practice, encourages examination of the reasons for these and helps to identify the changes that are required to affect improvements.</p> <p>References: (3), (6), (8)</p>



Criteria
Essential
5(a) 1. The pregnancy and newborn screening programmes are audited annually.
5(a) 2. Each maternity unit, laboratory, maternity ultrasound department, department of child health and audiology department contributes to and participates in the audit process.
5(a) 3. Audit and monitoring of the pregnancy and newborn screening programmes are overseen by a multiprofessional advisory group which meets regularly and reports annually to the NHS Board.
5(a) 4. Carer satisfaction with the screening process is audited annually.
Desirable
5(a) 5. Information technology arrangements are in place to support the national (Scottish) and local audit process.

STANDARD 6 ~ Laboratory

Standard Statement	Rationale
<p>6(a). Laboratories providing pregnancy and newborn screening services meet recognised professional standards, and the screening tests undertaken are subjected to rigorous quality control.</p>	<p>There is evidence that laboratories accredited and working to agreed standards achieve a high level of reporting accuracy. Quality control, both external and internal, is essential to provide an independent assessment of the performance of laboratory screening tests.</p> <p>Reference: (4)</p>
<p>6(b). The quality of the newborn screening service is continually assessed and monitored.</p>	<p>Regular review of the laboratory screening service is essential to assess the effectiveness of screening and compliance with the standards.</p>
<p>6(c). Each NHS Board has clear links between the local clinical service and the screening laboratories.</p>	<p>Explicit communication channels are essential as pregnancy and newborn screening programmes are multiprofessional and are provided on a regional or national level.</p> <p>References: (2), (3), (6)</p>
<p>6(d). There is a senior member of the laboratory staff at consultant level responsible for the pregnancy and newborn screening service who is supported by a designated laboratory co-ordinator and suitably qualified staff.</p>	<p>The responsibilities and accountabilities associated with testing and reporting and for the management and development of the laboratory service are consistent with the duties of a consultant grade.</p> <p>Reference: (4)</p>

Criteria

Essential

- 6(a) 1. The laboratory is accredited by an appropriate body, eg Clinical Pathology Accreditation (CPA) UK.
- 6(a) 2. There is a designated quality manager for the laboratory.
- 6(a) 3. The laboratory participates in accredited external quality assurance schemes, eg National External Quality Assessment Service (NEQAS), appropriate to the analyses carried out and shows satisfactory performance.
- 6(a) 4. Internal quality control procedures are undertaken and documented.

Newborn Bloodspot Screening

Essential

- 6(b) 1. There is a failsafe policy for action on positive cases. Follow-up action is agreed and in place.
- 6(b) 2. 95% of positive CHT, PKU and CF cases have started treatment by 14 days of age, unless deliberately delayed for further testing.
- 6(b) 3. 100% of positive cases have started treatment for CHT and PKU within 21 days of age, and for CF within 42 days of age, unless deliberately delayed for further testing.

Essential

- 6(c) 1. There are clearly defined systems and arrangements for documenting information between the local clinical service and the screening laboratory.
- 6(c) 2. 99.5% of infants who have undergone screening tests for CHT, PKU and CF have a screening result or are recalled for repeat testing by 20 days of age.

Essential

- 6(d) 1. There is a designated director of the laboratory screening service.
- 6(d) 2. There is a designated laboratory screening co-ordinator.
- 6(d) 3. All screening, testing and reporting is carried out by qualified staff.

STANDARD 6 ~ Laboratory (continued)

Standard Statement	Rationale
<p>6(e). Laboratory reports are issued for all screened pregnancies and newborn babies with minimum delay and the content conforms to the nationally agreed minimum data set.</p>	<p>Rapid reporting minimises patient anxiety and allows follow-up testing (where indicated) to be carried out as early as possible in pregnancy.</p> <p>Reference: (2)</p> <p>Meaningful interpretation of screening results must take into consideration a number of maternal and pregnancy variables.</p> <p>References: (1), (5), (8)</p> <p>The minimum data set ensures that the report is accurate, meaningful and easily interpreted and contains positive patient identification.</p>

Criteria

Pregnancy Screening: Down's Syndrome

Essential

- 6(e) 1. 95% of pregnancy screening reports are issued within 3 working days of receipt of the specimen at the laboratory.
- 6(e) 2. Interpretation of results takes account of:
- maternal variables including date of birth, maternal weight and maternal smoking status; and
 - pregnancy variables including gestation, previous affected pregnancies and assisted conception.

HIV

Essential

- 6(e) 3. 95% of screen non-positive results are issued within 5 working days to the requesting clinician.
- 6(e) 4. All screen positive results are confirmed and issued to the obstetric team within 10 working days.

Communicable Diseases:

Rubella, Syphilis and Hepatitis B

Essential

- 6(e) 5. 95% of all hepatitis B and rubella results are issued within 5 working days.
- 6(e) 6. 95% of syphilis results are issued within 10 working days.

Newborn Bloodspot Screening

Essential

- 6(e) 7. 95% of results are available within 2 working days of receipt of sample by laboratory.
- 6(e) 8. Interpretation of results takes account of:
- age at specimen collection;
 - health indicators on card; and
 - feeding status.

STANDARD 6 ~ Laboratory (continued)

Standard Statement	Rationale
<p>6(f). Screening laboratories combine appropriate markers with other identifiable risk factors to derive a risk of:</p> <ul style="list-style-type: none"> • Down’s syndrome; • neural tube defects; • HIV; • rubella; • syphilis; and • hepatitis B. <p>6(g). A risk threshold or agreed cut-off point is used as the criteria for offering follow-up diagnostic testing.</p>	<p>Sensitivity and specificity of screening is influenced by the choice of markers.</p> <p>References: (1), (5), (8)</p> <p>Choice of threshold risk determines the number of women who will be offered follow-up testing and the proportion of Down’s syndrome and neural tube defect pregnancies detected.</p> <p>References: (1), (5), (8)</p>

Criteria

Essential

6(f) 1. The markers used in the laboratory conform to those in Appendix 1.

Pregnancy Screening: Down's Syndrome

Desirable

6(g) 1. The follow-up rate is 5–7% in a typical screened population.

6(g) 2. The detection rate is 85% of Down's syndrome cases by first trimester combined ultrasound and biochemical (CUB) screening using a threshold risk of 1:250 at term.

6(g) 3. The detection rate is 65% of Down's syndrome cases by second trimester screening using a cut-off risk of 1:250 at term.

Neural Tube Defects

Desirable

6(g) 4. The follow-up rate is 2–4% in a typical screened population using a cut-off maternal serum alphafetoprotein (MsAFP) level of 2.0 MoM.

6(g) 5. The detection rate is 80–90% of pregnancies with open neural tube defects using a cut-off MsAFP level of 2.0 MoM.

STANDARD 6 ~ Laboratory (continued)

Standard Statement	Rationale
<p>6(h). The laboratory workload for the pregnancy and newborn screening programmes is adequate to generate stable median and screen positive rates on a monthly basis and large enough to contain sufficient Down's syndrome and neural tube defect pregnancies to allow meaningful annual audit of detection rates.</p>	<p>Small sample numbers affect the calculation of median and false positive results and limit the experience of the laboratory in dealing with affected pregnancies and complications.</p> <p>Reference: (9)</p>

Criteria

Pregnancy Screening: Down's Syndrome and Neural Tube Defects

Essential

- 6(h) 1. The laboratory processes a minimum of 1,000 pregnancy screening samples annually.
- 6(h) 2. Laboratories processing between 1,000 and 5,000 serum screening specimens annually are part of a network of laboratories measuring the same screening markers and using common analytical methods and risk calculation software, so that the combined workload exceeds 5,000 specimens annually.
- 6(h) 3. Laboratories processing more than 5,000 serum screening specimens annually may function independently and participate in multidisciplinary audit at local and Scottish level.

Communicable Diseases

Essential

- 6(h) 4. Laboratories processing screening samples for HIV and communicable diseases are part of a network with an established protocol for prompt referral of reactive samples to a specialist laboratory for HIV, hepatitis B or syphilis.
- 6(h) 5. Laboratories processing more than 1,000 screening samples annually for HIV and communicable diseases and performing confirmation assays may function independently.

Newborn Bloodspot Screening

Essential

- 6(h) 6. The laboratory processes a minimum of 25,000 screening samples annually.

STANDARD 7 ~ Child Health Registration and Screening

Standard Statement	Rationale
<p>7(a). The local child health department has a system in place to ensure that all newborn babies are registered within the NHS Board and registered on the Community Health Index (CHI).</p>	<p>The local registration system ensures that appropriate screening tests are offered to all babies.</p>

Criteria

Essential

7(a) 1. There is a weekly check by the local child health department to ensure that all babies born within the NHS Board have been recorded on the CHI.

Desirable

7(a) 2. There is a twice-weekly check by the local child health department to ensure that all babies born within the NHS Board have been recorded on the CHI.

Essential

7 (a) 3. There is a check of all babies on the record to ensure that a result has been recorded within 20 days.

7(a) 4. There is a local failsafe protocol to ensure that all babies have been offered screening and follow-up action taken where no results have been recorded.

Newborn Bloodspot Screening

Essential

7(a) 5. 100% of infants with no result are identified by 20 days of age.

7(a) 6. 99.5% of eligible infants are offered testing/retesting within 1 week of identification.

10. Appendix 1: Procedures for Follow-up Diagnostic Testing

The following screening tests are offered to all women at the most appropriate stage in their pregnancy:

Screening For:	Gestation	Screening Test
Down's Syndrome	11-14 weeks Or	Combined NT scan and biochemical screening (MsFβhCG, PAPP-A and maternal age)
	15-20 weeks	
Neural Tube Defects	10-14 weeks	1 st antenatal visit ultrasound scan Biochemical screening by MsAFP
	15-20 weeks Or	
	18-20 weeks	
HIV	10-14 weeks/1 st antenatal appointment (still worthwhile up to the point of delivery)	HIV antibody screening test
Communicable Diseases - Rubella, Syphilis and Hepatitis B	10-14 weeks/1 st antenatal appointment	Test for immunity to rubella and evidence of current infection with hepatitis B and syphilis

Diagnostic Follow Up

Down's Syndrome	11-14 weeks Or	Chorionic Villus Sampling (CVS)
	15 weeks +	Amniocentesis
Neural Tube Defects	18-20 weeks	Detailed ultrasound anomaly scanning

Action to be taken once results known

HIV

- Confirmation of a positive result is obtained by further antibody testing on primary sample.
- Confirmation of a screen positive result occurs in the laboratory, using at least two assays based on different components, before any release to the obstetric team.
- Confirmation of identity is obtained from repeat sample. Management of woman is transferred to an obstetrician according to local guidelines.

Rubella

If screen shows lack of immunity advise antenatal care team to ensure:

- serological test for infection (IgM) is repeated on contact with or development of a rash at any stage throughout pregnancy.
- lack of immunity is confirmed by a second assay.
- post-partum immunisation is offered. Confirmation of lack of immunity is performed by a different serological test on the primary sample.

Hepatitis B

- Confirmation of a screen positive result is carried out using a hepatitis B surface antigen neutralisation test.
- Establish the acute or chronic nature of infection by testing for additional serological markers of hepatitis B infection.
- Advise on prophylaxis and immunisation programme for protection of neonate.

Syphilis

- Confirm with a range of tests to establish infectivity. Advise on maternal treatment.

11. Glossary

abnormality	A finding requiring further investigation/treatment.
accreditation	A process, based on a system of external peer review using written standards, designed to assess the quality of an activity, service or organisation.
affected pregnancy	A pregnancy where the baby has a particular condition, such as Down's syndrome or neural tube defects.
alphafetoprotein (AFP)	A hormone that occurs naturally during pregnancy. The level of AFP can be detected by a maternal blood test, and can aid in assessing the risk of Down's syndrome or neural tube defects.
amniocentesis	A test carried out during or after 15 weeks of pregnancy for fetal abnormality. The test involves the removal of a small amount of fluid from the amniotic sac using a needle through the abdominal wall, for diagnostic purposes.
anencephaly	One of the two main types of condition which together are known as neural tube defects. The other condition is spina bifida. In babies with anencephaly, the skull and the brain are not properly formed.
antenatal	Relating to the period between conception and birth.
antibody	A protein produced in the blood, in response to the presence of harmful substances (called antigens) which it then destroys.
antigen	A substance that the body identifies as 'foreign', eg bacteria, cancer cells, transplanted tissue cells and pollen. The body generates antibodies to destroy antigens.
antigen neutralisation test	A test using a substance which is capable, under appropriate conditions, of inducing a specific immune response and of reacting with the products of that response.
assay	A laboratory test.
assisted conception	Medical help to conceive a baby, eg <i>in vitro</i> fertilisation.
asymptomatic infection	An infection without obvious signs or symptoms.
audiologist	A healthcare professional specialising in the measurement of hearing and the management of hearing impairments or hearing loss.
audiology	The science dealing with hearing impairments, their detection and management.
audit	The measuring and evaluation of care against agreed standards with a view to improving practice and care delivery.
Automated Auditory Brainstem Response (AABR)	A hearing assessment carried out on newborn babies, which records brain activity in response to sounds.

biochemical	Relating to the chemical processes and substances occurring in living things.
biochemical screening	Screening tests to assess for disorders of the body's chemistry, such as phenylketonuria (PKU) and congenital hypothyroidism (CHT).
child health surveillance system	A system used by NHS Boards to monitor certain aspects of child health and development.
chorionic villus sampling (CVS)	A test carried out usually up to 14 weeks of pregnancy. The test involves the removal of a small amount of tissue from the placenta.
chronic	Describes a disease or infection lasting a long time.
cirrhosis	Liver disease which is marked by the degeneration of cells and thickening of the tissue, as scar tissue replaces healthy cells.
clinical biochemist	A specialist who manages and develops methods of analysis and interpretation of patient samples, to assist with the investigation, diagnosis and treatment of diseases.
Clinical Pathology Accreditation (CPA) UK	An accreditation process which requires laboratories to meet pre-determined standards. Website: www.cpa-uk.co.uk
communicable disease	Any disease that is transmissible either directly or indirectly.
Community Health Index (CHI)	A unique patient identifier that is allocated to every patient registered with a GP in Scotland. It is entered onto a database that underpins a wide range of patient care processes in Scotland. There are strict controls on access to patient identifiable details.
conception	The start of pregnancy.
congenital	A disease or abnormality which is present at birth.
congenital hypothyroidism (CHT)	A rare condition in which a baby is born with a defect of the thyroid gland. This results in an underproduction of thyroxine.
congenital rubella syndrome	Fetal infection with the rubella virus during the first trimester of pregnancy, resulting in a series of congenital abnormalities including heart disease, deafness and blindness.
continuing professional development (CPD)	An ongoing commitment to learning in various forms, which maintains and enhances professional standards of work, and develops the ability to recognise good practice.
corrected age	The age adjusted for prematurity.
CUB screened	A combined ultrasound and biochemical test to determine the risk of a fetus having a condition such as Down's syndrome.
cystic fibrosis (CF)	An inherited disorder, characterised principally by chronic lung disease, a deficiency of pancreatic enzymes needed for digestion, and high levels of electrolytes (eg sodium, potassium and chloride) in sweat.

data set	A list of required and specific information relating to a specific disease.
diagnosis	Identification of an illness or health problem by means of its signs and symptoms. This involves ruling out other illnesses and causal factors for the symptoms.
diagnostic test	A test to confirm the presence of a condition.
dietician	An expert in nutrition who helps people with special health needs plan the kinds and amount of foods to eat.
DNA	Deoxyribonucleic acid. Material in the nucleus (brain of the cell) that codes what that cell will become structurally and functionally.
Down's syndrome	A medical condition that is present from birth. It causes learning difficulties and can be associated with other health problems such as heart defects.
enzyme	A protein that acts as a catalyst for biochemical reactions in the body, to speed up a biochemical reaction in the cell.
evaluation	The study of the performance of a service (or element of treatment and care) with the aim of identifying successful and problem areas of activity.
evidence-based	Evidence-based clinical practice is an approach to decision making in which the clinician uses the best evidence available, in consultation with the patient, to decide upon the option which suits that patient best.
External Quality Assurance (EQA) Schemes	A service where participating clinical laboratories are sent samples on a regular basis. They test these samples as if they had come from patients. The results are returned to EQA centres which provide a report that compares the participant's performance with that of all laboratories and/or groups of laboratories using the same test method(s).
failsafe	Reliable back-up.
false negative	A test result which indicates no abnormality, when one exists.
false positive	A test result which indicates an abnormality, when one does not exist.
fetus	Clinical term for an unborn child, more than 8 weeks after conception.
free beta hCG	A hormone which can be measured in a pregnant woman's blood.
gene	A distinct sequence of DNA forming part of a chromosome, which provides the genetic blueprint for all offspring.
genetic mutation	A change in the DNA of a cell, or the change this causes in a characteristic of the individual.
gestational age	The number of weeks which have passed in pregnancy since conception.
GP	General Practitioner. Also known as a family doctor.

guidelines	Systematically developed statements which help in deciding how to treat particular conditions.
Health Department Letter (HDL)	Formal communications from the Scottish Executive Health Department to NHSScotland (formerly known as Management Executive Letter - MEL).
health visitor	A public health nurse who has undertaken extra training in child development and health promotion, and who works in the community, either with a GP practice or according to a specific area.
healthcare professional	A person qualified in a health discipline.
hepatitis B	A virus commonly spread through contact with infected blood products (through transfusion) or blood contaminated needles. It may also be spread sexually or from mother to baby during pregnancy. It is a virus which infects the liver. It can be carried in the blood for many years before causing any signs of illness.
hepatocellular carcinoma	A cancer arising from the major cell of the liver (hepatocytes). Infection with hepatitis B or C increases the risk of developing the cancer. Also known as primary liver cancer.
higher chance	A screening test result which indicates an increased chance of abnormality.
HIV	Human immunodeficiency virus is the virus that causes AIDS (acquired immunodeficiency syndrome). Women can pass HIV to their babies during pregnancy, childbirth, and also through breastfeeding.
human chorionic gonadotrophin (hCG)	A hormone that occurs naturally during pregnancy, which is present in the blood and urine.
hydrocephalus	An accumulation of fluid in the brain. Hydrocephalus often accompanies spina bifida.
hypothyroidism	An underactive thyroid, which produces an insufficient amount of thyroid hormones.
immunisation	An artificial way of creating protection against certain infections, by using relatively harmless antigens that come from, or are similar to the micro-organisms that cause the diseases.
immuno reactive trypsinogen (IRT)	A protein in the bloodstream used to measure the risk of cystic fibrosis in a newborn baby.
Management Executive Letter (MEL)	Formal communications from the Scottish Executive Health Department to NHSScotland, now known as Health Department Letters (HDLs).
marker	A sign or substance which is used to determine the risk of having a particular medical condition.

median	The middle observation of a series arranged in ascending order. This can also be stated as: the number in the middle of a set of increasing numbers (eg the median of the following numbers is 5: 1,2,5,8,9).
metabolism	All chemical reactions that occur in the body using absorbed nutrients to provide energy and make new or replacement body substances.
MoM	Multiple of the median. A measurement used to record the results of some antenatal screening tests.
monitoring	The systematic process of collecting information on the performance of clinical or non-clinical activities, actions or systems. Monitoring may be intermittent or continuous. It may also be undertaken in relation to specific incidents of concern or to check key performance areas. Monitoring is used to appraise strengths, weaknesses, opportunities and threats.
MsAFP	Maternal serum alphafetoprotein.
MsFBhCG	Maternal serum free beta-human chorionic gonadotrophin.
MshCG	Maternal serum human chorionic gonadotrophin.
multidisciplinary	An approach combining the knowledge, skills and expertise of a range of organisations and professionals.
National Deaf Children's Society (NDCS)	A UK charity which is solely dedicated to the support of all deaf children, young deaf people, carers, families and professionals working on their behalf. Website: www.ndcs.org.uk
National External Quality Assessment Service (NEQAS)	In the UK, the National External Quality Assessment Service (NEQAS) is a confederation of external quality assurance schemes, whose members comply with a code of practice.
National Services Division (NSD)	The division of the Scottish Common Services Agency (known since May 2004 as NHS National Services Scotland) with responsibility for ensuring the provision of national screening programmes and specialist services on behalf of NHSScotland. Website: www.show.scot.nhs.uk/nsd/
neonatal	A term used to describe the first 28 days of a baby's life.
neural tube defect	Birth defects that involve the development of the brain or incomplete spinal cord and/or protective coverings for these organs. The two main types of neural tube defects are anencephaly and spina bifida.
NHS	National Health Service.

NHS Board	NHS Boards are responsible for the strategic planning, service delivery, performance management and governance of each of Scotland's 15 local health systems. Since 1 April 2004, most NHS Board areas (excluding Island NHS Boards) have contained NHS operating divisions which are the successors to the NHS Acute and Primary Care Trusts. Divisions are not equivalent to Trusts, since the latter had a separately-identifiable legal status and were overseen by an appointed Board with a Chair, remunerated members, etc. See NHS operating division.
NHS operating division	On 1 April 2004, NHS Trusts in Scotland were replaced by NHS operating divisions. NHS operating divisions are committees of an NHS Board, with schemes of delegated authority setting out operational freedom for the delivery of services. While they are successors to the NHS Acute and Primary Care Trusts, they have no separate legal identity from the NHS Board. See NHS Board and NHS Trust.
NHS Quality Improvement Scotland (NHS QIS)	NHS Quality Improvement Scotland is a statutory body, established as a Special Health Board in January 2003. Its role is to focus on improving the quality of patient care and the health of patients. It will have a particular emphasis on the quality of care and the patient journey for vulnerable groups. Website: www.nhshealthquality.org
NHS Trust	NHS Trusts were organisations responsible for providing a group of healthcare services for the local population. An Acute Trust provided hospital services. A Primary Care Trust provided primary care/community health services. Mental health services (both hospital and community-based) were usually provided by Primary Care Trusts. Since 2001, Trusts operated within an overall framework drawn up by their NHS Board. Trusts were dissolved on 31 March 2004, becoming operating divisions of the NHS Board. See NHS Board and NHS operating division.
NHSScotland	The National Health Service in Scotland.
nuchal	Relating to the region of the back or nape of the neck.
nuchal translucency (NT)	How the collection of fluid at the back of a fetus' neck appears on an ultrasound scan.
obstetric(s)	The branch of medicine and surgery that deals with pregnancy and childbirth.
obstetrician	A doctor specialising in pregnancy and childbirth.
Otoacoustic Emissions (OAE)	A neonatal screening test to detect risk of hearing impairment in babies.
paediatrician	A specialist doctor who treats children and infants.
PAPP-A	Pregnancy-associated plasma protein-A.

patient	A person who is receiving care or medical treatment. A person who is registered with a doctor, dentist, or other healthcare professional, and is treated by him/her when necessary. Sometimes referred to as a user.
patient journey	The pathway through the health services taken by the patient (the person who is receiving treatment), and as viewed by the patient.
peer review	Review of a service by those with expertise and experience in that service, either as a provider, user or carer, but who are not involved in its provision in the area under review. In the NHS Quality Improvement Scotland approach, all members of a review team are equal.
permanent congenital hearing impairment (PCHI)	Hearing loss which is present at birth.
pharmacist	A qualified professional who understands the nature and effect of medicines and how they are produced and used to prevent and treat illness, relieve symptoms or assist in the diagnosis of disease. Pharmacists use their expertise for the wellbeing and safety of users and the public.
phenylalanine hydroxylase	An enzyme which processes phenylalanine, an amino acid. Phenylalanine is necessary for growth in infants.
phenylketonuria (PKU)	A rare inherited metabolic disorder which prevents the normal use of protein. A substance called phenylalanine builds up in abnormally high amounts in the body. Very high amounts can damage the brain. The condition can be successfully treated if diagnosed shortly after birth.
physician	A specialist in medicine.
placenta	An organ within the uterus joining mother and offspring, and which sustains the pregnancy.
post-partum	Relating to the period of a few days immediately after birth.
premature	Born before the expected date of delivery.
pre-natal	The period between conception and birth.
pre-term	Born before the expected date of delivery.
primary care	The conventional first point of contact between a patient and the NHS. This is the component of care delivered to patients outside hospitals and is typically, though by no means exclusively, delivered through general practices. Primary care services are the most frequently used of all services provided by the NHS. Primary care encompasses a range of family health services provided by family doctors, dentists, pharmacists, optometrists and ophthalmic medical practitioners.

prophylaxis	The prevention of disease; preventive treatment.
protein	Proteins are made of amino acids, and are the main component of human cells. There are many different proteins in the human body and they all have different functions. Protein is one of the three main food types, found for example in meat, fish, poultry, and eggs, and is required to allow the body to grow and to repair its cells.
protocol	A policy or strategy which defines appropriate action in specific circumstances. Protocols may be national, or agreed locally to take into account local requirements.
psychologist	A specialist in the scientific study of the mind and trained in assessing emotional and behavioural problems.
quality assurance (QA)	Improving performance and preventing problems through planned and systematic activities including documentation, training and review.
quality control	Steps that are taken to ensure the service is of sufficiently high quality.
reactive	A positive result.
referral	The process whereby a patient is transferred from one professional to another, usually for specialist advice and/or treatment.
rubella	A highly contagious virus also known as German measles.
Scottish Executive	The devolved government for Scotland.
Scottish Executive Health Department (SEHD)	The Scottish Executive Health Department is responsible for health policy and the administration of NHSScotland. Website: www.show.scot.nhs.uk/sehd
screening	A public health service offered to groups of the population to identify risk of a particular disorder or disease. This therefore involves examination of people with no symptoms, to detect unsuspected disease.
screening programme	The systematic and co-ordinated screening process.
self-assessment	Assessment of performance against standards by individual/clinical team/NHS operating division/NHS Board providing the service to which the standards are related.
sensitivity	The probability that when a person has a condition it will be picked up by the test.
serological test	A test performed on serum to determine if specific disease and antibodies are present.
serum	The clear fluid portion of blood that is left when the blood clots.

Special Health Board	The name given to Health Boards with a national remit. These boards are focused on specific areas, eg NHS Education for Scotland, or NHS Quality Improvement Scotland. Special Health Boards match regional NHS Boards in terms of administrative grading.
specificity	The probability that when a person does not have a condition the test will be negative.
specimen	A biological sample.
spina bifida	A neural tube defect which affects the spinal cord.
statutory	Enacted by statute; depending on statute for its authority as a statutory provision. Required by law.
syphilis	A sexually transmitted disease caused by bacteria. Syphilis may also be passed on from mother to baby during pregnancy.
threshold risk	The value at which a diagnostic test is considered to be appropriate.
thyroid (gland)	A gland in the neck which secretes the hormone thyroxine, which is important in the development of a baby's brain and controls the body's metabolic rate.
thyroxine	A hormone which controls the body's metabolic rate.
<i>treponema pallidum</i>	The bacterium causing syphilis.
trimester	Three months, ie one-third of the length of a pregnancy.
Trust	See NHS Trust.
ultrasound	An image created by the use of sound waves above the audible range of the human ear. It is useful in the confirmation of pregnancy, the determination of fetal size and wellbeing.
Universal Newborn Hearing Screening (UNHS) programme	A national hearing screening programme, which aims to implement a hearing screen for all newborn babies. It is expected that the programme will be rolled out from April 2005.
viral antigen	Part of a virus that the body regards as foreign or potentially dangerous and against which it produces an antibody.
virus	A very simple sub-microscopic organism which can cause disease and is only able to reproduce inside the living cells of a host.
window period	The period between the onset of an infection and the ability to make the diagnosis of the condition.

Our Commitment

Our work will be undertaken in line with the following values:

- **patient and public focus**
 - ~ promoting a patient-focused NHS that is responsive to the views of the public
- **independence**
 - ~ reaching our own conclusions and communicating what we find
- **partnership**
 - ~ involving patients, carers and the public in all parts of our work
 - ~ working with and supporting NHS staff in improving quality
 - ~ collaborating with other organisations such as public bodies, voluntary organisations and manufacturers to avoid duplication of effort
- **evidence-based**
 - ~ basing conclusions and recommendations on the best evidence available
- **openness and transparency**
 - ~ promoting understanding of our work
 - ~ explaining the rationale for our recommendations and conclusions
 - ~ communicating in language and formats that are easily accessible
- **quality assurance**
 - ~ aiming to focus our work on areas where significant improvements can be made
 - ~ ensuring that our work is subject to internal and external quality assurance and evaluation
- **professionalism**
 - ~ promoting excellence individually and as teams and ensuring value for money in the use of public resources (human and financial)
- **sensitivity**
 - ~ recognising the needs, opinions and beliefs of individuals and organisations and respecting and encouraging diversity

This document can be viewed on the NHS Quality Improvement Scotland website. It is also available, on request, from NHS Quality Improvement Scotland in the following formats:

- Electronic
- Audio cassette
- Large print

NHS Quality Improvement Scotland

Glasgow Office ~ Delta House 50 West Nile Street Glasgow G1 2NP Tel 0141 225 6999

Edinburgh Office ~ Elliott House 8-10 Hillside Crescent Edinburgh EH7 5EA Tel 0131 623 4300

comments@nhshealthquality.org www.nhshealthquality.org

