NHS Sickle Cell & Thalassaemia
Screening Programme

Standards for the linked Antenatal and Newborn Screening Programme

Second edition
October 2011
The focus of the Programme is now on quality-assuring and evaluating the outcome of the screening process.

Dr Allison Streetly
Programme Director

Acknowledgements
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Second edition – October 2011
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<td>Family Origin Questionnaire</td>
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Glossary of haemoglobinopathy terms

Please see Section 4 for generic glossary of screening definitions

Alpha thalassaemia major (haemoglobin Barts hydrops fetalis) A severe anaemia that affects the fetus. No normal fetal haemoglobin is produced and this leads to stillbirth or neonatal death.

Amniocentesis An invasive procedure undertaken at about the 16th week of pregnancy in order to obtain a sample of the amniotic fluid surrounding the fetus for testing.

Beta thalassaemia major A severe anaemia caused by inheritance of two beta thalassaemia genes, resulting in a lack of normal haemoglobin production. Treatment by regular blood transfusions and drugs to remove excess iron leads to long-term survival. Some affected children can be ‘cured’ by bone marrow transplantation.

Carrier (also referred to as trait) An individual who carries a single altered gene where two altered genes are required for an individual to be affected with a condition that may require treatment. The carrier can pass on the gene to their offspring. The most common haemoglobin carrier states in the UK are Hb S, C, D, E and beta thalassaemia.

Chorionic villus sampling (CVS) An invasive procedure performed under ultrasound guidance through either the cervix or the abdomen after 10 completed weeks of pregnancy, to obtain a sample of placental tissue for testing.

Family origins A term used to describe a person’s ancestry.

Haemoglobin disease Mild or serious diseases that can occur in people who have inherited two haemoglobin gene variants. The most common haemoglobin diseases are sickle cell diseases and thalassaemia disorders, also called haemoglobinopathies.

Prevalence The proportion of people in a population who have an attribute or a given disease.

Normal haemoglobin This is called Haemoglobin A – for adult haemoglobin. This is composed of two α chains and two β chains.

Screening programme The whole system of activities needed to deliver high quality screening. It ranges from identifying and informing those to be offered screening through to the treatment and follow-up of those found to have an abnormality, as well as support for those who develop disease despite screening.

Sickle cell anaemia (Hb-SS) A sickle cell disease caused by inheritance of two genes for haemoglobin S, which often results in significant health problems and requires treatment. Some affected children can be ‘cured’ by bone marrow transplantation.

Sickle cell disease A group of inherited diseases that are characterised by sickling of red blood cells when there is a shortage of oxygen. The most common sickle cell diseases are sickle cell anaemia (SS), haemoglobin SC disease, and haemoglobin S/beta thalassaemia. Sickle cell diseases can cause episodes of acute pain (crisis), anaemia, increased risk of infections, and chest problems. They can be life-threatening, particularly for young children.

Thalassaemia major A group of inherited conditions caused by a reduction in the amount of haemoglobin produced. People with a thalassaemia condition have various degrees of severe anaemia.

Variant A change from the usual, for example, in a gene or protein. A variant haemoglobin gene may result in sickle or another type of haemoglobin in the body.
Our Mission

Support people to make informed choices during pregnancy and before conception.

Provide high quality and accessible care throughout England.

Improve infant health through prompt identification of affected babies.

Promote greater understanding and awareness of the disorders and the value of screening.
Foreword

This document brings together:

• aims and objectives of the linked newborn and antenatal NHS Sickle Cell and Thalassaemia Screening Programme (SCT Programme)

• standards for the SCT Programme

• guidance from the UK National Screening Committee (UK NSC) on quality assurance for non-cancer screening programmes

• supporting guidance on organisational arrangements and laboratory requirements to achieve these standards in England.

The SCT Programme standards were first published in 2006 [1]. During 2010/11 the Programme conducted a consultation on revisions to the standards. This consultation showed good support for:

• continuing to use the Donabedian principles of setting aims, objectives, criteria and standards to assess the quality of the Programme

• a ‘light touch’ revision, mainly to clarify the standards or to revise upwards the minimum and achievable levels

• introducing the wider cross-programme generic objectives for the eight non-cancer screening programmes (Please see Section 3 which lists the generic objectives, cross-referencing to the SCT Programme standards)

• inclusion of the evidence base for the standard (Please see Section 2)

Since 2006 the UK NSC has developed a catalogue of Key Performance Indicators (KPIs), chaired by the Director of the SCT Programme [2]. Three antenatal KPIs were chosen for antenatal sickle cell and thalassaemia screening (coverage, timeliness of test by 10 weeks’ gestation, and completion of Family Origin Questionnaire – FOQ) and one joint newborn KPI was chosen with the UK Newborn Screening Programme Centre (timeliness of result availability). The KPIs are linked to the relevant revised standards in Sections 6 and 7.

The revised standards underwent an Equality Impact Assessment to address possible inequalities and reduce the potential for discrimination. This involves assessing the likely or actual effects of the standards on people in respect of disability, gender and racial equality. The outcome was that we needed to do more work on offering screening and counselling to baby’s fathers, which has been clarified in AP2 (Please see Section 6).

These revised programme standards will become fully operational from 1st April 2012. The organisational context in which screening programmes are commissioned and managed is set to change, although the details are yet to be confirmed. Therefore some of the structures identified in this document may undergo alterations in the future.

Although the standards are specifically aimed at England, they may be useful in the development and implementation of screening programmes in Scotland, Wales and Northern Ireland.

This is the second edition of this document and it will be reviewed at the end of 2014, or earlier if needed.

Please send any comments you have to the Programme Office at the address given below.

Dr Allison Streetly, OBE, FFPH
Programme Director

Please return comments via email or letter to:

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Website: http://sct.screening.nhs.uk
**Summary of changes**

**Revisions to the 1st edition of the document include:**

- Inclusion of evidence to support screening policy and standards (Please see Section 2)
- Incorporation of cross programme quality assurance developments including KPIs, generic screening objectives, generic screening definitions and the generic screening QA pathway. In addition, we have specified clear links with the UK NSC data definitions and key objectives for screening by mapping our SCT Programme objectives to the generic screening objectives and KPIs (Please see Section 3)
- Separation of organisational guidance for national and local screening programmes (Please see Sections 9 and 10), including the addition of guidance on specialist commissioning for preimplantation genetic diagnosis (PGD).
- Clarification of the minimum laboratory criteria requirements, which were revised to include quality improvement across the whole screening pathway (Please see Section 11). The additions to the minimum criteria for laboratories include:
  - Antenatal laboratories: criteria 11 (sensitivity), 12 (failsafe), and 13 (responsibility for regular audit and review)
  - Newborn laboratories: criteria 11 (sensitivity), 12 (failsafe), 13 (links with clinical networks) and 14 (responsibility for regular audit and review)
  - DNA referral laboratories: criteria 11 (sensitivity) and 12 (failsafe)

**NB:** These changes in minimum laboratory criteria will be incorporated into the 3rd Edition of the Handbook for Laboratories, which is due to be published in 2012
- Addition of Appendices on linkage between the antenatal and newborn screening programmes (Appendix One) and guidance concerning issues of non-paternity (Appendix Six).

**Revisions to the programme standards include:**

- Re-naming of ‘minimum (core)’ and ‘achievable (developmental)’ standards to ‘acceptable’ and ‘achievable’ in line with cross programme arrangements
- Minor changes in wording of standards for clarity
- Equality Impact Assessment has been conducted on the standards in order to address inequality and minimise the potential for discrimination.

**Revisions to the Antenatal Standards include:**

- Change in the outcome to be achieved in the antenatal screening programme so that for those women accepting prenatal diagnosis, 50% of prenatal diagnoses are to be performed **before 12 weeks, 6 days**, so that any subsequent action, if requested, can be performed by **13 weeks, 6 days**.
- The timeline for antenatal screening is as follows:
  - 50/70% of all pregnant women offered screening by **10 weeks, 0 days** (AP1);
  - 50/75% of prenatal diagnoses performed before **12 weeks, 6 days** (AO1b);
  - Any subsequent action by **13 weeks, 6 days**
- Increase in the thresholds for acceptable and achievable levels across the antenatal screening programme, in line with the requirements for the KPIs. This includes increasing AO1a (to facilitate informed decisions/choice about screening for all women) to 95/99% for offer and coverage of screening tests
  - Inclusion of clear links with KPIs in AO1aii and AO1aiii
  - Inclusion of a new standard for antenatal laboratories to meet minimum laboratory criteria and share data with the screening programme (AO2a)
  - Minor changes in wording for clarity
  - A table showing all the changes to the Standards for the Antenatal Screening Programme is available on the website at [http://sct.screening.nhs.uk/](http://sct.screening.nhs.uk/)

“The outcome of the Equality Impact Assessment was that we needed to do more work on offering screening and counselling to baby’s fathers”
Revisions to the Newborn Standards include:

- Increase in the thresholds for acceptable and achievable levels across the newborn screening programme, in line with the requirements for the KPIs
- Clearer links with the UK Newborn Screening Programme standards and KPIs
- Inclusion of new standards, including:
  - coverage of newborn screening, linked to KPI NB1 (NO2ii)
  - reporting newborn carrier results using agreed protocol (NP2ii)
- Removal of the requirement to for 95% of infants to have completed the primary Prevenar (conjugate pneumococcal vaccination) course by 15 months. This was withdrawn because PCV13 is now given universally to all babies
- A table showing all the changes to the Standards for the Newborn Screening Programme is available on the website at http://sct.screening.nhs.uk/

Revisions to the Linked Antenatal and Newborn Standards include:

- Increase in the thresholds for acceptable and achievable levels across the linked antenatal and newborn screening programme
- Inclusion of a standard to link antenatal and newborn screening results through an alert system to inform newborn laboratories of screen positive women (LO2)

NB: The direction of travel is to include antenatal screening results in a systematic manner on the Bloodspot card. It is anticipated that the 2011/12 consultation will provide space to record antenatal screening results on the Bloodspot card

- A table showing all the changes to the Standards for the Linked Antenatal and Newborn Screening Programme is available on the website at http://sct.screening.nhs.uk/
Section 1
Background

A About the Programme
B Haemoglobinopathies
C Policy
D Quality assurance across screening programmes
E Setting standards
F Quality Improvement
Background

A About the Programme

The NHS Sickle Cell & Thalassaemia Screening Programme (SCT Programme) is part of a family of non-cancer National Screening Programmes under the management of the UK National Screening Committee:
http://www.screening.nhs.uk/england

The Antenatal and Newborn Screening Programmes in England include:

- Linked NHS Sickle Cell & Thalassaemia Screening Programme
- Fetal Anomaly Screening Programme
- Infectious Diseases in Pregnancy Screening Programme
- The Newborn Hearing Screening Programme
- Newborn and Infant Physical Examination
- Newborn Bloodspot Screening. This includes screening for
  - Phenylketonuria (PKU)
  - Congenital hypothyroidism (CHT)
  - Sickle cell and thalassaemia
  - Cystic fibrosis
  - MCCAD (medium chain acyl-CoA dehydrogenase deficiency).

The SCT Programme mission statement is to:

- support people to make informed choices during pregnancy and before conception
- improve infant health through prompt identification of affected babies
- provide high quality and accessible care throughout England
- promote greater understanding and awareness of the diseases and the value of screening.

The SCT Programme is funded to develop, implement and maintain high quality, uniform antenatal and newborn screening for sickle cell and thalassaemia. To achieve this requires standards to be set for services throughout England to facilitate the aims of the programme. This includes training for health and allied professionals involved in the screening pathway; development of communication materials; programme awareness for the general public and health care professionals; and close working with NHS commissioners.
B Haemoglobinopathies

Haemoglobinopathies are common in people whose family origins are in malarial parts of the world. In the UK, haemoglobinopathies are seen particularly among minority ethnic groups from Africa, the Caribbean, the Mediterranean, South East Asia, the Middle East, and the Far East [3], but can be found (less frequently) in all ethnic groups. Approximately 1000 haemoglobin gene variants have been identified worldwide.

Sickle cell diseases are estimated to affect 1 in every 2,000 births in England [4] [5]. 240,000 people are estimated to be carriers and more than 12,500 people have the disease. The highest prevalence is among Black Africans and Black Caribbeans [4] or [5].

Recent laboratory data submitted to the SCT Programme show that almost 670,000 newborn samples are screened annually for sickle cell disease in England, with approximately 360 babies identified with screen positive results for significant condition, and over 9,600 babies identified as a carrier of a haemoglobin variant [6].

Sickle cell disease affects the normal oxygen-carrying capacity of red blood cells due to a change in the biochemistry of haemoglobin. When deoxygenated red blood cells are unable to pass freely through capillaries, they form clusters instead which block blood vessels. This blockage prevents oxygenation of the tissues in the affected areas, resulting in tissue hypoxia and consequent intense pain (known as sickle cell crisis). The other symptoms of sickle cell disease include severe anaemia, damage to major organs and infections [7].

Early detection in newborns and appropriate management can improve quality of life, especially when parents/guardians and users learn to recognise and avoid the risk factors that can trigger a painful ‘crisis’ (attack), and take antibiotics to prevent infections [8].

Beta thalassaemia is thought to affect more than 700 people, with approximately 214,000 carriers in the UK. The highest carrier prevalence is among Cypriot, Italian, Greek, Indian, Pakistani, Bangladeshi, Chinese, other South East Asian and Middle Eastern populations [4] or [5].

Thalassaemia major (Beta thalassaemia) is the most severe form of thalassaemia. The body is unable to produce haemoglobin, leading to life-threatening anaemia. Individuals with the condition require regular blood transfusions and treatment to prevent complications from iron overload, such as diabetes and other endocrine disorders. Bone marrow transplantation may be a treatment option for some individuals [9].

Both conditions (sickle cell disease and beta thalassaemia major) can restrict a child’s or adult’s ability to conduct normal daily activities, and can also have profound psychosocial effects on individuals and their families.

Antenatal screening identifies about 22,000 carriers of sickle cell disease and thalassaemia every year [6].

“Every year, approximately 360 newborn babies are identified with screen positive results for significant condition, and antenatal screening identifies approximately 22,000 sickle cell and thalassaemia carriers”
C Policy
The aim is to deliver a linked newborn and antenatal programme, to ensure that users experience screening as one service – not as separate, disjointed services delivered by different professionals and different organisations.

More information about what we mean by a linked programme, the benefits for patients and the NHS, and the particular challenges for the NHS with this genetic screening programme can be found in Appendix One and at http://sct.screening.nhs.uk/linkage.

### Antenatal screening - conditions to be screened for:

(i) Significant maternal haemoglobinopathies
these should be detected by antenatal screening and are important for maternal care

- Hb-SS and other genotypes of sickle cell disease (Hb SC, Hbs/β thalassaemia, etc)
- β thalassaemia intermedia Hb H disease (/--/α) (β thalassaemia major will be clinically apparent)

(ii) Maternal conditions requiring testing of the baby’s father

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<tr>
<th>a) Conditions in (i)</th>
<th>Potential significant diseases in the fetus</th>
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<td>Hb-SS</td>
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<td>HB-AS</td>
<td>Hb-SC</td>
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<td>HB-AC</td>
<td>Hb-SD/Punjab</td>
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<td>HB-AD/Punjab</td>
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<td>HB-A Lepore</td>
<td>Hb-S/β thalassaemia</td>
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<td>Hb-S/ββ thalassaemia</td>
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<td>α e thalassaemia trait (/--/α α)</td>
<td>β thalassaemia major (except cases with silent or near silent maternal phenotype)</td>
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<td>HPFH</td>
<td>Hb E/β thalassaemia;</td>
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<th>c) Any compound heterozygote state including one or more of the above conditions</th>
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<td>d) Any homozygous state of the above conditions</td>
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1. Antenatal screening policy
The policy for antenatal screening in England is to offer sickle cell and thalassaemia screening to all women as an integral part of early antenatal care [10]. All areas need to collect information on family origin in their maternity populations which, as well as being a screening tool in low prevalence areas, is needed by laboratories to help interpret screening results.

2. Antenatal prevalence
High prevalence areas (Appendix Two)
- All pregnant women offered screening for sickle cell, thalassaemia and other haemoglobin variants using routine blood cell indices and a family origin questionnaire to assist with risk assessment of positive results
- For women identified as carriers, their baby’s father (irrespective of family origin) is offered testing for sickle cell, other haemoglobin variants and thalassaemia.

Low prevalence areas (Appendix Three)
- All pregnant women offered screening for thalassaemia using routine red blood cell indices
- Family origin questionnaire used to screen for the risk of either the woman or the baby’s father being a carrier for sickle cell and other haemoglobin variants. Women in high risk groups or women whose baby’s father is in a high risk group are offered laboratory testing for haemoglobin variants
- All fathers of babies of identified carrier mothers (irrespective of family origin) offered testing for sickle cell, other haemoglobin variants and thalassaemia.

Lists of high and low prevalence areas are available on our website at http://sct.screening.nhs.uk/professional-resources

3. Newborn screening policy
The policy for newborn screening in England is to offer sickle cell screening to all infants as an integral part of the UK Newborn Screening Programme Centre (UK NSPC) [10].

Newborn screening- conditions to be screened for:
- HbSS
- HbS/βthalassaemia (β+, β0, δβ, Lepore)
- HbS/HPFH
- HbSC
- HbSDpunjab
- HbS/E
- HbS Qkush

‘Other clinically significant haemoglobinopathies likely to be detected as by-products by newborn screening’:*  
- β thalassaemia major
- Hb E/β thalassaemia;
- β thalassaemia intermedia
- HbH disease

*The SCT programme obtained the following clarification from the UK NSC in November 2008: “Such findings which are not part of the screening programme but are detected by current screening methods should be reported to the relevant clinician and parents to facilitate management of the consequences of such findings” [10].

Please note: Policy on screening for “other clinically significant haemoglobinopathies is currently undergoing review by the UK NSC. Any changes to screening policy will be found at http://sct.screening.nhs.uk/policy.
“A consistent approach to quality assurance across the eight non-cancer screening programmes will lead to an improved screening journey for service users”

D Setting standards
The approach taken by the SCT Programme builds on the work of Donabedian [11] to:

1. Clarify aims and objectives of the screening programme
2. Use aims and objectives to set criteria for standards
3. Set standards as a subjective judgement of the level of performance that could be achieved
4. Set standards to include an acceptable threshold that all programmes must meet and an achievable threshold that all programmes should aim to meet (see box on page 14).

This approach was originally developed in 2006 by the SCT Programme, in line with some existing NHS screening programmes – for example, the breast and cervical screening programmes. Since 2009 guidance by the UK NSC is for all non-cancer screening programmes to use this approach.

The process of setting standards is iterative and continuous. The cycle is as follows:

• Set standards
• Identify improvements required to meet the standards
• Monitor service against standards
• Aim for continual improvement for good services.

Although the standards are informed by the capacity of existing systems, they should drive the development of information systems and technology to meet the needs of the programme, rather than technology driving what is reported.

A key aspect of the SCT Programme is that the newborn and antenatal programmes are linked. Information from the antenatal screening programme can inform newborn screening in relation to both interpreting results and communicating with parents. With this in mind, standards have been set for the following aspects of the service:

• Antenatal screening
• Newborn screening
• Linked programme

In addition, laboratory ‘standards’ have been agreed and are included in Section 11 of this document. While these are not standards in the true sense of the word (mostly they require 100% compliance), they are commonly referred to as such. To avoid such confusion we have used the term ‘minimum laboratory criteria’. The minimum laboratory criteria in this edition of the standards has been revised to include quality improvement across the whole screening pathway. These changes will be incorporated into the 3rd Edition of the Handbook for Laboratories, which is due to be published in 2012.

E Quality assurance across screening programmes
A consistent approach to quality assurance across the eight non-cancer screening programmes is being developed by the UK NSC (http://www.screening.nhs.uk/quality-assurance). This is strongly supported by the SCT Programme as this will lead to an improved screening journey for service users. This approach will enable local providers – including commissioners and screening leads – to understand the structures and quality assurance issues of each programme by ensuring QA arrangements are made once across all programmes and not duplicated. The result should be increased efficiency and greater leverage than a system based on individual programmes. This will be achieved by:

• Agreed common objectives across all programmes. Programme-specific items will sit beneath a top ‘tier’ of objectives and data measures. Links between the programme standards and these top level objectives are described in more detail in Section 3.
• ‘Mainstreamed’ developments that reflect NHS policy, in patient safety and quality metrics and KPIs.
• Serious Incident (SI) guidance for screening that was developed in conjunction with the National Patient Safety Agency.
• Service specifications which give clarity to commissioners and ensure a joined up service across organisations.
• Use of common terminology across screening programmes. The cross-programme approach uses the terms ‘achievable’ and ‘acceptable’ to describe standard levels (see box on page 14). We have therefore used these terms in place of the terms ‘Minimum (core)’ and ‘Achievable (developmental)’ that were used in the first edition of the standards. A generic glossary of screening definitions is listed in Section 4.
Figure 1: Generic Screening Pathway

1. Coverage
2. Completeness of offer
3. Uptake
4. Acceptance of offer

**EXPECTED**
Population estimated to meet programme criteria

**TOTAL**
Population meeting programme criteria

**ELIGIBLE**
Population that should be offered screening

**INELIGIBLE**
Population that should not be offered screening

**OFFERED**
Population that have been offered screening

**NOT OFFERED**
Population that have not been offered screening

**ACCEPTED**
Population that wish to accept screening

**DECLINED**
Population that do not wish to accept screening

**TESTED**
Population receive conclusive screening

**NOT TESTED**
Population not receiving conclusive screening

**ACTIONED**
Population for whom test is followed up

**NOT ACTIONED**
Population for whom test is not followed up
• Development of generic screening objectives and an outline screening pathway. The generic screening pathway is given in Figure 1, illustrating coverage and uptake.

Further detail on the cross-programme approach can be found at http://www.screening.nhs.uk/quality-assurance.

**Standard levels: ‘acceptable’ and ‘achievable’**

**Acceptable:** The acceptable threshold is the lowest level of performance considered safe. All programmes are expected to exceed the acceptable threshold, and to agree service improvement plans that develop performance towards an achievable level. Programmes not meeting the acceptable threshold are expected to implement recovery plans to ensure rapid and sustained improvement.

**Achievable:** The achievable threshold represents safe and robust performance; screening programmes should budget for and aspire towards performance at this level. Local constraints may sometimes result in programmes failing to meet this threshold. Service improvement plans should focus on the delivery of a balanced service with as many standards as possible meeting the achievable threshold.

The linked NHS Sickle Cell and Thalassaemia Screening Programme is working with the UK NSC Quality Assurance team to ensure high quality screening is delivered across professional and organisational boundaries. In order to achieve accessibility, equity and access to services within an acceptable cost – clear pathways and robust partnerships are required. Communication is a key aspect of maintaining quality in this environment, and a coordinated plan is required to ensure that individual processes do not appear disjointed [12]. User involvement in the process is also essential, and is a key commitment of the coalition government [13].

These standards have been developed to support the SCT Programme and allow the quality aspects of its development to be achievable and measurable. The focus is on care pathways, especially following a positive result (carriers in the antenatal and major disease in the newborn). Coherent, consistently applied systems should involve the whole organisation and all the components of the service – from antenatal screening through to newborn screening and the clinical care of affected children.

A well developed quality management system should indicate when and how any task is to be undertaken and by whom [14], with a named person at each step of the pathway. The whole organisation should be involved in planning, delivering and evaluating the quality of services [12]. This should also be guided by Clinical Governance processes. The screening pathway, along with failsafe points, is outlined on a Map of Medicine pathway, which can be found at:

http://sct.screening.nhs.uk/carepathways and

http://www.screening.nhs.uk/mapofmedicine

Audit and evaluation is required as part of overall quality assurance, including annual and data reports on process and outcome from individual programmes.

"Quality assurance is important for service users and is also used by staff to deliver services efficiently"
Section 2
Evidence to support screening

A Evidence to support the SCT Programme
B Evidence to support antenatal screening
C Evidence to support newborn screening process
D Evidence to support benefits of newborn screening
E Linked antenatal and newborn screening
Evidence to support screening

**A Evidence to support the SCT Programme**

**Evidence supporting screening policy:** Antenatal and newborn SCT screening policy is informed by two health technology assessment (HTA) reviews [15] [16]. In addition, two recent HTA reports have been published – one on how best to achieve screening early in pregnancy [17] and one on reporting newborn carrier results [18]. Since the Programme’s introduction there has been a WHO resolution [19], which urged member states to implement comprehensive programmes for the prevention and management of sickle cell anaemia. Many more countries across the world have now started to introduce screening programmes.

**Evidence supporting the sensitivity of the screening programme:** This data has been obtained from elsewhere in the world and is mostly focussed on newborn screening and prenatal diagnosis [20] [21] [22] [23] [24]. Information on sensitivity and specificity of antenatal screening has been obtained following discussions with experts in the field.

**Acceptable and Achievable levels:** The evidence to support ‘acceptable’ and ‘achievable’ levels for these standards follows a review of data already collected [6]; data from Key Performance Indicators (KPIs) (http://www.screening.nhs.uk/kpi); and discussions with providers at local and national level. Review of the initial and available KPI data show that the acceptable and achievable levels have been set realistically, although there are variations in performance across the country.

**B Evidence to support antenatal screening**

**Evidence supporting the need for early screening and prenatal diagnosis:** Antenatal screening policy is to offer screening to all pregnant women. If a woman is identified as a carrier, the baby’s father will also be offered screening. Practically this means offering screening early enough to allow time for screening the baby’s father, prenatal diagnosis, making a decision and taking action if wished – including vacuum suction termination – before the pregnancy is public knowledge.

There is a known association between gestation at screening offer and uptake of prenatal diagnosis (PND), with the early offer of screening being associated with greater uptake of PND [25] [26] [27]. One of the main objectives of antenatal screening for sickle cell and thalassaemia is to ensure that all eligible women are offered screening by 10+0 weeks, to allow time for fathers to be tested and for early prenatal diagnosis if wanted, reporting of results, and any subsequent action by the end of 13 weeks of pregnancy. This is consistent with NICE Guidelines [28] and preferred by women [29]. The majority of prenatal diagnostic testing currently takes place after 12+6 weeks’ gestation [30] [6], which is too late to allow parents to make informed and timely reproductive choices, especially for some groups in the population and contrary to users wishes.

**Attitudes towards prenatal diagnosis and termination of affected pregnancies:** Having identified carriers, and subsequently at-risk pregnancies, the effectiveness of any screening programme in reducing the number of babies born with a condition will depend on attitudes towards termination of affected pregnancies. There are many factors which influence views on antenatal screening and termination of pregnancy, including perceptions of the condition (previously reported in the UK [29] and more recently in China [31]). Evidence from Modell et al [25] and Greengross et al [27] show that attitudes in the UK towards prenatal diagnosis are associated with (i) gestation at time of offer and (ii) ethnicity.

Karimi found that in Iran, 93% of beta-thalassaemia major patients and their parents were in favour of prenatal diagnosis, with 87% also favouring early termination of an affected fetus [32]. In contrast, Kolnagou reported that in Cyprus (where prenatal and antenatal diagnosis have almost eliminated births with thalassaemia major), improved supportive care of thalassaemia patients – for instance, effective iron chelation therapy – means some parents are now choosing not to abort an affected fetus [33].

**Evidence supporting use of Family Origin Questionnaire (FOQ):** The SCT Programme commissioned research to develop, pilot and evaluate an evidence-based family origin questionnaire and to establish its effectiveness as a screening tool for identifying individuals at higher risk from specific haemoglobinopathies [34]. This followed on from earlier research to determine the best form of question to be piloted for use as a screening tool. The information obtained was used to develop the format of the final FOQ [35]. NICE guidelines also support the use of the FOQ to

“The early offer of screening is associated with greater uptake of prenatal diagnosis (PND)”
identify those at highest risk of being a carrier for both antenatal screening and pre-conception testing in low prevalence areas [28].

C Evidence to support newborn screening process
Newborn screening for sickle cell is delivered as part of the UK Newborn Bloodspot Screening Programme.

Evidence to support acceptability of newborn screening test: Initial analysis of screen positive results has been published and the uptake of screening is high – with only approximately 300 declines per year across the whole of England (for all newborn testing, not just SCT screening [4]).

Evidence to support upper age limit: Newborn screening programmes in any country will not diagnose all children with significant haemoglobinopathies because of new arrivals. As recently highlighted in Holland, recently arrived immigrant children may often miss out on early diagnosis and appropriate treatment, but can make up a significant number of the cases (27% of cases diagnosed in Holland 2003–7) [36]. In the UK it is recommended that any child up to the age of 1 year arriving in the UK should undergo newborn screening. Above this age, the awareness and vigilance of health professionals is relied on to ensure that appropriate testing is carried out in children from at-risk ethnic groups, if clinically indicated.

Evidence supporting testing babies who have been transfused: An area of concern in newborn screening for sickle cell relates to the possibility of sickle cell disease babies being missed because of a blood transfusion prior to a bloodspot sample being taken [37]. These babies do not have a valid sickle cell screen result. Currently, the Guidelines for Newborn Bloodspot Testing recommend taking one bloodspot prior to transfusion [38]. As a failsafe, the SCT Programme is funding a pilot for DNA testing of transfused babies without a pre-transfusion sample (http://sct.screening.nhs.uk/cms.php?folder=2512 ). In the absence of this failsafe and a pre-transfusion sample the recommendation (specifically for sickle cell screening) is for repeat testing at 4 months post-transfusion using a liquid sample of blood. However, this is challenging, as it is a difficult, costly and very time-consuming process for primary care and specialist counselling staff to administer. For babies that have multiple transfusions, the repeat testing policy could result in a considerable delay in being tested for sickle cell disease with the potential risk of missing a baby with the condition. The difficulties with the repeat testing process are well highlighted by audit data from Sheffield Children’s Hospital which showed that only 3 babies out of 24 were re-tested in a timely manner following transfusion [39]. At King’s College Hospital over an 18 month period, only 28% requests for follow-up of transfused babies were received [40].

D Evidence to support benefits of newborn screening
Evidence supporting effective follow-up of screen positive babies: Newborn screening policy aims to reduce the mortality and morbidity of babies with sickle cell disease by ensuring they enter care quickly. Accurate diagnosis of sickle cell disease in infants allows for identification and recognition of early clinical manifestations, and for early prophylaxis and treatment against infection [41] [42] [43] [44] [45] [46]. The policy for newborn screening is informed by evidence showing the benefits of timely treatment for babies with sickle cell disease [47] [48] [49] and education for their parents [18] [43] [41] [51] [52] [53] [54] [55] [56].

Evidence of outcomes from newborn screening programmes: As yet, there is local but not national evidence available on morbidity or mortality outcomes from the Newborn Screening Programme in the UK [57]. However, there is evidence from some of the more longstanding programmes. For example, Quinn et al showed that American children are clearly benefiting from more timely first visits and early interventions in childhood [58]. They documented reduced mortality rates and changing patterns of mortality in a cohort of children identified by the newborn screening programme in Dallas; sepsis was no longer the leading cause of death and 94–98% of children with sickle cell disease now reach adulthood. Similarly, in Jamaica, Hardie showed high levels for uptake of pneumococcal vaccine and reduced incidence of invasive pneumococcal disease following immunisation in children with sickle cell disease who had been identified via the newborn screening programme [59].
Evidence of mortality and morbidity: Despite improvements in life expectancy, sickle cell disease remains a cause of significant morbidity – even in developed countries. Boulet et al found that compared with other American black children, those with sickle cell disease were more likely to show intellectual disability, probably as a result of cerebral cardiovascular accident (CVA); have higher incidence of severe headaches; more frequent use of prescription medication and have a health status classed as ‘fair’ or ‘poor’ [60]. Vichinsky et al showed that even in neurologically asymptomatic adults with sickle cell disease, there was evidence of significant neurocognitive dysfunction – which worsened with age and anaemia [61]. Two UK studies have looked at the outcome of admissions to critical care. For sickle cell admissions to the paediatric intensive care unit the mortality rate was 6%; two of the three recorded deaths were children who had presented to the hospital in cardiorespiratory arrest [62]. The most common reason for admission was acute chest syndrome, and almost three-quarters of the children admitted underwent an exchange blood transfusion. Gardner studied adult admissions to the critical care facility of a single centre in London [63]. He identified 46 admissions over an eight year period: 30% were for acute chest syndrome, 17% for multiorgan failure and 15% post elective surgery. He found that the mortality rate for sickle cell patients was 19.6% compared with an overall mortality rate for the unit of 17.6%. However, the readmission rate over the period of the study was relatively high at 16%. Clarke et al found that despite the availability of benefits and care from the NHS, quality of life for children with thalassaemia major in the UK was still poor compared with population norms [64].

Evidence of reducing strokes: There is an increased risk of stroke in children with sickle cell disease. In one multicentre study in the US about 11% of people with sickle cell disease had a clinically evident stroke by the age of 20 [65]. Recent evidence has shown the benefits or assessing the risk of stroke in children with sickle cell disease as it is possible to prevent first stroke by regular blood transfusions [66]. There are now clinical guidelines recommending that children with sickle cell disease should be offered annual Trans Cranial Dopplar scans from age 2 years to assess the risk of stroke [67].

Effect of new vaccinations: The introduction of the new pneumococcal vaccine into the general immunisation schedule in England has been a welcome development. There is now evidence from the USA of a marked reduction and possibly elimination of fatal pneumococcal infections in sickle cell babies, following the introduction of the pneumococcal conjugate vaccine [68]. However, babies and children with sickle cell disease have a reduced immunity and require extra immunisations as well as penicillin on a continuous basis until age 5, as not all infections are prevented by these measures.

Evidence supporting carrier identification in newborn screening: Current screening technology identifies not only babies affected by sickle cell or thalassaemia major, but also those who are healthy genetic carriers of sickle and other variant haemoglobin types. The issues for carriers are different than for affected babies as there is no need for treatment, and therefore no immediate health benefit from the screening programme. However, there are a variety of potential psychosocial consequences of disclosing carrier status – including increasing anxiety for families; the possibility of highlighting non-paternity; and an impact on parental behaviour towards the child, parental relationships, and reproductive planning. The Human Genetic Commission (HGC) has provided guidance and opinions [69]. It supports sharing information about genetic status from screening, endorsing the general approach taken by the UK NSC– which is to inform parents of any results identified as a by-product of newborn screening.

Evidence supporting communicating carrier results to parents: In the context of reporting carrier results from screening, research funded by the NHS Health Technology Assessment (HTA) explored current practice, methods and experience of communicating carrier status information following newborn screening. Kai et al [18] showed that while parents may not have been aware of the likelihood of carrier identification, most welcomed this information. They also showed that lack of information, particularly in the earliest stages of the process, contributed to anxiety levels. Linking antenatal and newborn screening results has the potential to reduce anxiety for parents. Kai et al reported considerable variation in who imparted the information and how, with face-to-face contact being the preferred option whenever possible – this is particularly for those without prior knowledge of sickle cell or those for whom English was not their first language [70]. Information needed to be given by someone knowledgeable, but their background/role did not matter. The need for better information across the whole pathway was highlighted, especially for translated literature.
on sickle cell carrier results. Acharya found that in America, with well-developed newborn screening programmes, there was still significant misunderstanding of carrier status and its health implications; formal professional counselling was rare unless a family also had a child with the full-blown disease [71]. Similarly, Lang et al found that amongst a group of largely African-American new mothers in Illinois, there was poor understanding of newborn screening for sickle cell – with only 35% able to recall any mention of it being made antenatally [72]. In parts of Canada, where newborn screening included sickle cell disease from 2006 (but has since become more complex with increased test availability), Miller et al assessed provider views and found that while few providers supported obtaining explicit consent condition by condition, 40% favoured obtaining specific consent to disclose carrier states that might be found as a by-product of the screening process [73]. Most healthcare providers agreed that they had a duty of disclosure to parents. However, it was genetics professionals who were more likely to disagree with disclosure as they felt a child should be able to decide for him or herself whether or not to receive the result in the future [74].

Parents need consistent communication about results and action required for positive screening/carrier results. A robust pathway is essential, with results reported to parents in a timely manner by a trained professional [75] [76] [77] [78] [79] [80]. Data from the screening programme can be used to refine the information provided to parents [4].

The UK NSC has produced a booklet entitled Screening tests for your baby which has been translated into 18 different languages. The evidence shows that screening and carrier testing does not appear to have an adverse emotional impact one month after screening. There are too few studies to comment on the emotional impact in the first four weeks after screening [81].

The screening programme has produced two leaflets to support reporting of newborn carrier information [82] [83], and we recommend use of a standard letter in any communication with parents.

Evidence to support linked antenatal and newborn screening

Evidence supporting linked standards: The standards for the linked programme are informed by user experience, best practice, and patient safety concerns. The need to provide a linked programme is particularly important given the lack of failsafe mechanisms in place across the whole pathway at present.

Evidence supporting the importance of user experience and patient safety: User experience and patient safety are well recognised as important aspects of quality care [84] [13], and are therefore essential components of the screening pathway. The standards are set to facilitate a high quality user experience within the context of a safe screening programme. It is particularly important to have a measure of user experience where Patient Reported Outcome Measures (PROMs) are not considered appropriate [85]. User involvement is a key element in policy development and service implementation for all the screening programmes – including the development of materials for the SCT Programme.

Evidence supporting information for parents: There is a lot of information about haemoglobinopathy screening available, which varies in quality and accuracy. Miller assessed the views and knowledge of providers, parents and members of the sickle cell community, finding that there were still inconsistent messages and difficulties communicating with parents [86]. This was a view supported by Downing et al, who highlighted the challenges of providing information in a timely and easily accessible way to a variety of users [87]. They also highlighted the need to improve the infrastructure of electronic information to facilitate information exchange and ensure consistent messages throughout all stages of the process – from antenatal care through to early childhood.

The SCT Programme continues to develop materials for users along the whole screening journey, from pre-conception to antenatal and newborn screening. This includes a range of material on the Programme website, information developed with other organisations (such as the Sickle Cell Society and the UK Thalassaemia Society), and a booklet produced by the UK NSC – Screening tests for you and your baby – which has been translated into 18 different languages [88].
Section 3
Generic screening objectives
These generic objectives have been developed and agreed as part of the UK NSC cross-programme initiative across the eight non-cancer screening programmes (fetal anomaly; antenatal infectious disease, newborn blood spot, newborn hearing, newborn infant and physical examination, diabetic retinopathy and abdominal aortic aneurysm). The table below maps the generic objectives against the SCT programme objectives. In our standards table we have kept the original order to maintain the consistency of antenatal and newborn pathways, but have included the link to the generic objective.

<table>
<thead>
<tr>
<th>Generic Objective</th>
<th>SCT Programme Objective</th>
<th>Programme Standard</th>
</tr>
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<tbody>
<tr>
<td><strong>1 Identify Population:</strong> To accurately identify population to whom screening is offered</td>
<td>To facilitate informed decisions/choice in the screening programme for: - All women in England - All women and couples identified as ‘at high risk’ of an affected pregnancy.</td>
<td>AO1ai, AO1aiii</td>
</tr>
<tr>
<td></td>
<td>Completeness of Coverage</td>
<td>NO2i, Newborn Bloodspot Standard 9</td>
</tr>
<tr>
<td><strong>2 Inform:</strong> To maximise the appropriate (timely and informed) offer of screening and testing to eligible population</td>
<td>To facilitate informed decisions/choice in the screening programme for: - All women in England - All women and couples identified as ‘at high risk’ of an affected pregnancy.</td>
<td>AO1aii</td>
</tr>
<tr>
<td></td>
<td>Timely offer of screening by 10+0 weeks of pregnancy.</td>
<td>AP1</td>
</tr>
<tr>
<td></td>
<td>Completeness of offer</td>
<td>Newborn Bloodspot Standard 1</td>
</tr>
<tr>
<td><strong>3 Uptake:</strong> To maximise uptake in the eligible population who wish to participate and have been fully informed about the screening programme.</td>
<td>Timely offer of screening by 10+0 weeks of pregnancy.</td>
<td>AP1</td>
</tr>
<tr>
<td></td>
<td>To ensure that education and community awareness of the value of screening is provided to the wider population in an acceptable and accessible manner, is appropriate to the level and needs of the relevant community groups and aims to reduce the stigma sometimes associated with the conditions screened for.</td>
<td>LO3</td>
</tr>
<tr>
<td><strong>4 Test:</strong> To maximise accuracy of screening test</td>
<td>To accurately identify women and couples with genotypes specified as requiring further investigation.</td>
<td>AO2a</td>
</tr>
<tr>
<td></td>
<td>To identify with specified sensitivity babies born with conditions where early intervention is likely to be beneficial.</td>
<td>NO2i</td>
</tr>
<tr>
<td></td>
<td>To identify and arrange timely follow-up of infants identified as needing further investigation.</td>
<td>NP1, NP5</td>
</tr>
<tr>
<td></td>
<td>Quality laboratory screening services</td>
<td>AO2a, NS2</td>
</tr>
<tr>
<td>Generic Objective</td>
<td>SCT Programme Objective</td>
<td>Programme Standard</td>
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| **5 Minimising harm:**  
To minimise potential harms in those screened and in the population | To ensure high quality pathway for women/couples ‘at high risk’ of an affected pregnancy. | AP2 |
|  | To provide timely expert counselling for women/couples with ‘high risk’ pregnancies (includes effective and acceptable follow-up for those undergoing prenatal diagnosis and those not choosing prenatal diagnosis). | AP4 |
|  | To minimise the adverse effects of screening including anxiety, misunderstanding, failure to communicate results, inaccurate information, unnecessary investigation & follow-up and inappropriate disclosure of information. | LO2  
NP7i |
|  | Reliable screening sample on all transfused babies | NP1 |
|  | To report results of screening in a timely manner. | NP2 |
|  | Timely communication of screening result. | NP3  
NP5 |
|  | To ensure treatment is offered and parental education started in a timely manner for screen positive children. | NP6iii |
|  | To minimise the adverse effects of screening – including failure to follow-up screen positive infants, inaccurate or inadequate information, unnecessary investigation and follow-up, inappropriate disclosure of information and failure to communicate results to parents (normal and carrier). | NP7i |
|  | To ensure that education and community awareness of the value of screening is provided to the wider population in an acceptable and accessible manner, is appropriate to the level and needs of the relevant community groups and aims to reduce the stigma sometimes associated with the conditions screened for. | LO3 |
| **6 Diagnose:**  
To maximise accuracy of diagnostic test | To identify, with specific sensitivity, specified conditions where prenatal diagnosis is undertaken. | AO2b |
|  | To identify, with specified sensitivity babies born with conditions where early intervention is likely to be beneficial. | NO2i |
|  | Timely confirmation of diagnosis for infants with a positive screening result for specified conditions | NP5 |
|  | Quality newborn laboratory screening service | NS2 |
| **7 Intervention / Treatment:**  
To ensure high quality and timely intervention in those who wish to participate and have been fully informed about the screening programme. | To ensure high quality and timely intervention for women/couples ‘at high risk’ of an affected pregnancy. | AP3 |
|  | To ensure effective and acceptable follow-up, care and support for affected infants and their carers. | NP4 |
|  | To ensure treatment is offered and parental education started in a timely manner. | NP6 i, ii and iii |
### Generic screening objectives

<table>
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<tr>
<th>Generic Objective</th>
<th>SCT Programme Objective</th>
<th>Programme Standard</th>
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</table>
| **8 Outcome:** To optimise population and individual health outcomes in target population | For all at-risk women and couples:  
-To ensure that women accepting the offer of prenatal diagnosis are tested within the specified timeframe | AO1b |
| | To ensure screening tests are offered by 10+0 weeks of pregnancy. | AP1 |
| | Mortality rate in children under 5 is less than 4 per 1000 person years of life (2 deaths per100 affected infants) | NO1 |
| **9 Staff:** To ensure that the whole screening programme is provided by a trained and competent workforce | To provide timely expert counselling for women/couples with “high risk” pregnancies (includes effective and acceptable follow-up for those undergoing prenatal diagnosis and those not choosing prenatal diagnosis). | AP4 |
| | To ensure an appropriate level of understanding about screening and these conditions among professionals involved with the programme to allow the effective and sensitive delivery of the screening programme to the population in general. | LO1 |
| | To minimise the adverse effects of screening – including anxiety, & inappropriate follow-up. | LO2 |
| **10 Commissioning/Governance:** To ensure effective (i) commissioning and (ii) governance of the screening programme | To ensure that responsibility, accountability and performance management for all aspects of the programme are clear, and that these link together from local to national level and between the newborn and antenatal programme. | AS1  
NP7ii  
NS1 |
| | To provide regular written feedback of the effectiveness and quality of the service to:  
- the population screened  
- the commissioners and providers of services  
- the range of bodies concerned with quality of health services as part of openness and transparency about service quality. | LO4 |
| | To ensure that responsibility and accountability for all aspects of the programme are clear, with direct links from National Programme accountability through regional teams to local level accountability. | LS1 |
| | To evaluate the service on an ongoing basis to assist continuous quality improvement. | LS2 |
| | To make effective and efficient use of resources to the benefit of the population. | LS3 |

*User experience and equality are included in each theme*
Section 4
Generic Glossary of Screening Definitions
**Generic Glossary of Screening Definitions**

The Glossary defines terms that are consistent across the Quality Assurance standards / service objectives of all screening programmes. A broken underline indicates that a term is used according to its definition in this glossary.

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
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<tbody>
<tr>
<td>accept</td>
<td>A response to an offer which indicates that a screening subject is willing to proceed with a screening encounter. Acceptance may be inferred from conduct provided that an offer has been made. In the case of newborn screening programmes, a responsible parent / guardian can accept screening on behalf of the subject baby.</td>
</tr>
<tr>
<td>acceptance of offer</td>
<td>The proportion of those offered screening who accept the offer. Low acceptance of offer might indicate that: i) the offer is not being communicated or delivered effectively (no response); and/or ii) screening is not deemed necessary or desirable by an entitled population (declined).</td>
</tr>
<tr>
<td>affected case</td>
<td>An individual in whom the condition being screened for is present.</td>
</tr>
<tr>
<td>booking</td>
<td>The point at which a pregnant woman first sees a midwife for an antenatal booking history, when details of the current pregnancy are documented in maternity records (which should be an auditable information system but may be a paper-based record where appropriate information systems have not been implemented).</td>
</tr>
<tr>
<td>communication</td>
<td>An interchange that the subject is capable of understanding and acting upon.</td>
</tr>
<tr>
<td>completeness of offer</td>
<td>The proportion of those eligible for screening who are offered screening. Completeness of offer is a measure of how effectively a programme offers screening to the eligible population.</td>
</tr>
<tr>
<td>coverage</td>
<td>The proportion of those eligible for screening who are tested. Coverage is a measure of the delivery of timely screening to an eligible population. Low coverage might indicate that: i) not all eligible people have been offered screening; ii) those offered screening are not accepting the test; and/or iii) those accepting the test are not being tested.</td>
</tr>
<tr>
<td>day of report</td>
<td>The day on which data to support an audit or performance return are collated. Usually there will be a time lag between the end of the reporting period and the day of report to allow for the completion of processes being measured and the collation of report data.</td>
</tr>
<tr>
<td>decline</td>
<td>A response to an offer which indicates that a screening subject does not wish to proceed with a screening encounter.</td>
</tr>
<tr>
<td>diagnosis</td>
<td>A diagnostic process following a screen positive result to determine whether the subject is an affected case.</td>
</tr>
<tr>
<td>effective timeframe</td>
<td>The period of time within which a screening test can be delivered such that a reliable result is most likely to be obtained. The effective timeframe for a test is usually specified in the policy / guidance for the relevant screening programme.</td>
</tr>
<tr>
<td>eligible</td>
<td>The population that is entitled to an offer of screening. The criteria for eligibility may be administrative, demographic, clinical, or any combination of these, and may take into account individual circumstances such as time of presentation to the screening service.</td>
</tr>
<tr>
<td>failed offer</td>
<td>Any indication that an attempted offer failed, such as a Post Office return. An offer will be deemed as a failed offer if: i) it did not reach the subject; ii) the subject was not capable of understanding or acting upon it; iii) the screening service lacked the capacity to realise it; and/or iv) it did not offer an opportunity of testing within an effective timeframe.</td>
</tr>
<tr>
<td>false negative</td>
<td>A screen negative result in an affected case.</td>
</tr>
<tr>
<td>false positive</td>
<td>A screen positive result for a subject in whom the condition being screened for is absent.</td>
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<tr>
<td>Term</td>
<td>Definition</td>
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<tr>
<td>first registered</td>
<td>First notification to a GP practice and/or Child Health Records Department of responsibility for the care of a baby. Following first registration, the baby is deemed as being registered with the GP practice and/or Child Health Records Department to which the notification was made. In most cases this will be an automatic notification to a Child Health Records Department following the birth of a baby.</td>
</tr>
<tr>
<td>general population</td>
<td>The overall population of the geographic area for which a screening service is responsible. The boundaries of a screening service, and thus the general population for which it is responsible, are usually defined by a list of General Practices. The total population will be identified from patients registered with any of the General Practices within the general population.</td>
</tr>
<tr>
<td>maternity service</td>
<td>A co-ordinated network of healthcare professionals contracted to or working under the policies and procedures agreed with a single acute Trust, with collective responsibility for the provision of antenatal, intrapartum and postpartum care. A single maternity service may include: • obstetric-led maternity units; • midwifery-led maternity units; • units responsible for the management of home births; • Newborn Intensive Care Units (NICU); • Special Care Baby Units (SCBU); and/or • Paediatric Intensive Care Units (PICU).</td>
</tr>
<tr>
<td>NN4B Number</td>
<td>An NHS number issued by the NN4B (NHS Numbers for Babies) system.</td>
</tr>
<tr>
<td>offer</td>
<td>A formal communication made by the screening service, giving a specific subject a realisable opportunity to be tested within an effective timeframe. An offer or invitation will only count as an offer if: i) it reaches the subject; ii) the subject is capable of understanding and acting upon it; iii) the screening service has the capacity to realise it; and iv) it offers an opportunity of testing within an effective timeframe. In the case of newborn screening programmes, the offer of screening is made to a responsible parent / guardian rather than the subject baby.</td>
</tr>
<tr>
<td>pregnancy</td>
<td>Confirmed pregnancy, recorded in a woman’s health notes.</td>
</tr>
<tr>
<td>presentation</td>
<td>The first attendance of a screening subject for a screening encounter.</td>
</tr>
<tr>
<td>realisable</td>
<td>Capable of being acted upon, concluded or delivered.</td>
</tr>
<tr>
<td>refer</td>
<td>The process of securing further diagnosis / specialist assessment following a screen positive test. The date of referral is the first realisable assessment date offered by an appropriate specialist unit to a screening subject following a screen positive result. Allocation to a pending list or a referral subsequently cancelled by the specialist unit is not a referral.</td>
</tr>
<tr>
<td>registered</td>
<td>Formally recognised as being the primary provider of ongoing care to an individual and holding sufficient details to uniquely identify and contact that individual.</td>
</tr>
<tr>
<td>reporting period</td>
<td>The defined time period over which activities should be included in an aggregate audit or performance return. A reporting period can relate to any specified period but for routine reports is usually quarterly or annual. Most screening processes occur over a period of days or weeks, to allow a scan or sample to be assessed. In such cases, a single point in the process (such as the screening encounter) should be used to determine whether the process falls within a particular reporting period.</td>
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<tr>
<td>Term</td>
<td>Definition</td>
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<tr>
<td>result</td>
<td>A formal and completed assessment of the risk of a condition being screened for in a subject, following a screening encounter. Usually a result will be screen positive or screen negative. Insufficient and unassessable indicate a failure to obtain a result, and are not themselves results.</td>
</tr>
<tr>
<td>screen negative</td>
<td>An indication following a test that the condition being screened for is low-risk / not suspected in a subject.</td>
</tr>
<tr>
<td>screen positive</td>
<td>An indication following a test that the condition being screened is high-risk / suspected in a subject.</td>
</tr>
<tr>
<td>screening</td>
<td>Testing people who do not have or have not recognised the signs or symptoms of the condition being tested for, either with the aim of reducing risk of an adverse outcome, or with the aim of giving information about risk.</td>
</tr>
<tr>
<td>screening encounter</td>
<td>The provision of screening to a screening subject, usually through a process such as a scan or the collection of a sample. A screening encounter is usually characterised by contact between the screening subject and a healthcare professional, but some screening may be self-administered.</td>
</tr>
<tr>
<td>screener</td>
<td>A healthcare professional responsible for administering screening tests.</td>
</tr>
<tr>
<td>screening episode</td>
<td>The end-to-end screening process from the perspective of a subject who has accepted an offer of screening. A complete screening episode starts with an offer and ends with the communication of a conclusive result. Some screening episodes may end prematurely, for example if the subject fails to attend a booked screening encounter.</td>
</tr>
<tr>
<td>subject</td>
<td>An eligible individual.</td>
</tr>
<tr>
<td>subject record</td>
<td>The personal information stored on the programme database about a subject.</td>
</tr>
<tr>
<td>test</td>
<td>A screening encounter leading to the determination of a conclusive result.</td>
</tr>
<tr>
<td>total population</td>
<td>The population that meets the general criteria for inclusion within a screening programme. The criteria for inclusion within a screening programme may be administrative, demographic, clinical, or any combination of these. Not everyone in the total population is likely to be eligible for screening (for example, those who present later than it would be possible to test).</td>
</tr>
<tr>
<td>true positive</td>
<td>A screen positive result in an affected case.</td>
</tr>
<tr>
<td>uptake</td>
<td>The proportion of those offered screening who are tested. Uptake is a measure of the delivery of screening in the population to which it is offered. Low uptake might indicate that: i) those offered screening are not accepting the test; and/or ii) those accepting the test are not being tested.</td>
</tr>
</tbody>
</table>

The minimum laboratory criteria requirements have been revised to include quality improvement across the whole screening pathway. These changes will be incorporated into the 3rd Edition of the Handbook for Laboratories, which is due to be published in 2012.
Section 5
Aims & objectives

A. Antenatal Sickle Cell & Thalassaemia Screening Programme

B. Newborn Sickle Cell Screening Programme (as part of the Bloodspot Screening Programme)

C. Linked Antenatal and Newborn Sickle Cell & Thalassaemia Screening Programme
Offer timely antenatal sickle cell and thalassaemia screening to all women (and couples) to facilitate informed decision-making, with any subsequent action (if required) taken by the end of 13 weeks of pregnancy.

Ensure screening tests are offered by 10+0 weeks of pregnancy.

For those women accepting prenatal diagnosis, 50% of prenatal diagnoses to be performed before 12 weeks, 6 days.
Antenatal Sickle Cell & Thalassaemia Screening Programme

Aims & objectives

Overall aim of the Programme:
To offer* timely antenatal sickle cell and thalassaemia screening to all women (and couples) to facilitate informed decision-making.

*The offer includes: the offer of, uptake of, and reporting of results of prenatal diagnosis and any subsequent action by the end of 13 weeks of pregnancy

Outcomes to be achieved:
For those women accepting prenatal diagnosis, 50% of prenatal diagnoses to be performed before 12 weeks 6 days.

Programme objectives:

AO1 To facilitate informed decisions/choice in the screening programme for:
• All women in England
• All women and couples identified as ‘at high risk’ of an affected pregnancy.

AO2a To accurately identify women and couples with genotypes specified (please see Section 1) as requiring further investigation.

AO2b To identify with specific sensitivity the conditions where prenatal diagnosis is undertaken (please see Section 1).

AP1 To ensure screening tests are offered by 10+0 weeks of pregnancy

AP2 To ensure high quality pathway for women/couples ‘at high risk’ of an affected pregnancy

AP3 To ensure timely intervention for women/couples ‘at high risk’ of an affected pregnancy.

AP4 To provide timely, expert counselling for women/couples with ‘high risk’ pregnancies (includes effective and acceptable follow-up for those undergoing prenatal diagnosis and those not choosing prenatal diagnosis).

AS1 To ensure that responsibility, accountability and performance management for all aspects of the programme are clear, and that these link together from local to national level and between the newborn and antenatal programmes.

“For those women accepting prenatal diagnosis, 50% should be performed before 12 weeks, 6 days”
To achieve the lowest possible childhood death rate and to minimise childhood morbidity from sickle cell diseases

To offer screening for sickle cell diseases to all infants in a timely manner with appropriate follow-up

Best possible survival as assessed by mortality rate in children under 5 of less than 4 per 1,000 person years of life (2 deaths per 100 affected infants)
Newborn Sickle Cell Screening Programme
(as part of the Bloodspot Screening Programme)

Aims & objectives

**Programme aims:**
To achieve the lowest possible childhood death rate and to minimise childhood morbidity from sickle cell diseases.

**Overall outcome to be achieved at national level:**

**NO1**
Best possible survival for infants detected with a sickle cell disease by the screening programme as assessed by a mortality rate in children under 5 is less than 4 per 1000 person years of life (2 deaths per 100 affected infants)

**NO2i**
To identify with specific sensitivity babies born with conditions where early intervention is likely to be beneficial (as listed in the Handbook for Laboratories [10]).

**Programme objectives:**

To offer screening for sickle cell diseases to all infants*

**NP2/NP3**
To process tests and report results in a timely manner.*

**NP1/NP5**
To identify and arrange timely follow-up of infants identified as needing further investigation.

**NP4/NP6i**
To ensure effective and acceptable follow-up, care and support for affected infants and their carers.^

**NP6i/NP6iii**
To ensure treatment is offered and parental education started in a timely manner.

**NP2/NP7i**
To minimise the adverse effects of screening – including failure to follow up screen positive infants, inaccurate or inadequate information, unnecessary investigation and follow-up, inappropriate disclosure of information and failure to communicate results to parents (normal and carrier).

**NP7ii**
To ensure that responsibility, accountability and performance management for all aspects of the programme are clear and that these link together from local to national level and between the newborn and antenatal programmes.

**NS1i**
Failsafe to ensure ongoing care.

**NS2**
Quality newborn laboratory screening service.

**NOTES:**

* These standards should be read in conjunction with the UK Newborn Screening Programme Centre Standards [38] see http://sct.screening.nhs.uk/cms.php?folder=2493

^ Please refer to the ‘Sickle Cell Disease in Childhood. Standards & guidelines for Clinical Care’ [67]. The standards can be found at http://sct.screening.nhs.uk/cms.php?folder=249

“Our aim is to achieve the lowest possible childhood death rate and to minimise childhood morbidity from sickle cell diseases”
Linked Antenatal and Newborn Sickle Cell & Thalassaemia Screening Programme:

Aims & objectives

LO1  To ensure an appropriate level of understanding about screening and these conditions among professionals involved with the programme to allow the effective and sensitive delivery of the screening programme to the population in general.

LO2/ NB 7i  To minimise the adverse effects of screening – anxiety, misunderstanding, inaccurate information, unnecessary investigation and inappropriate follow-up and that links are made across the antenatal and newborn screening pathway.

NP7ii  To ensure that responsibility, accountability and performance management for all aspects of the programme are clear and that these link together between the newborn and antenatal programmes.

LO3  To ensure that education and community awareness of the value of screening is provided to the wider population in an acceptable and accessible manner, is appropriate to the level and needs of the relevant community groups, and aims to reduce the stigma sometimes associated with the conditions screened for.

LO4  To provide regular written feedback of the effectiveness and quality of the service to:
- the population screened
- the commissioners and providers of services

LS1  To ensure that responsibility and accountability for all aspects of the programme are clear, with direct links from National Programme accountability through regional teams to local level accountability.

To evaluate the service on an ongoing basis to assist continuous quality improvement.

LS3  To make effective and efficient use of resources to the benefit of the population.
To support research to inform the development of the programme.

To make the NHS Sickle Cell and Thalassaemia programme a model programme worldwide and specifically in Europe.

“It is our aim to minimise the adverse effects of screening and to ensure that links are made across the antenatal and newborn screening pathway”
Section 6
Standards for the Antenatal Screening Programme
These standards should be read in conjunction with the Standards for the Linked Antenatal and Newborn Screening Programme. A broken underline indicates that a term is used according to its definition in the glossary in Section 4.

<table>
<thead>
<tr>
<th>Number</th>
<th>Objective</th>
<th>Criteria</th>
<th>Acceptable standard</th>
<th>Achievable standard</th>
<th>Level for monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td>AO1</td>
<td>To facilitate informed decisions/ choice about screening</td>
<td>Offer of screening tests</td>
<td>95% of pregnant women with a documented report of offer of screening</td>
<td>99% of pregnant women with a documented report of offer of screening</td>
<td>Local</td>
</tr>
<tr>
<td>AO1ai</td>
<td>For all women Generic theme 1</td>
<td>Coverage of screening test (tested/eligible)</td>
<td>95% of eligible pregnant women receive a conclusive screening test</td>
<td>99% of eligible pregnant women receive a conclusive screening test</td>
<td>KPI SCT1- coverage</td>
</tr>
<tr>
<td>AO1a</td>
<td>Generic theme 2</td>
<td>Completed screening test i) To identify high risk groups for screening (LP) ii) To aid test interpretation (HP &amp; LP)</td>
<td>90% of antenatal sickle cell and thalassaemia samples submitted to the laboratory supported by a completed Family Origin Questionnaire (FOQ)</td>
<td>95% of antenatal sickle cell and thalassaemia samples submitted to the laboratory supported by a completed Family Origin Questionnaire (FOQ)</td>
<td>KPI SCT3- completion of FOQ</td>
</tr>
<tr>
<td>AO1b</td>
<td>For all at-risk women and couples Generic theme 8</td>
<td>Women accepting the offer of prenatal diagnosis and being tested</td>
<td>50% of prenatal diagnoses performed before 12 weeks 6 days of pregnancy</td>
<td>75% of prenatal diagnoses performed before 12 weeks 6 days of pregnancy</td>
<td>Local</td>
</tr>
<tr>
<td>AO2</td>
<td>Accurate detection of affected pregnancies</td>
<td>Sensitivity of laboratory methods at detecting affected and carrier individuals</td>
<td>In high prevalence areas: i) 95% of affected/ carrier individuals of thalassaemia and significant haemoglobin variants to be detected by laboratory tests In low prevalence areas: i) 95% of affected/ carrier individuals of thalassaemia to be detected by laboratory tests ii) 95% of affected/ carrier individuals of significant haemoglobin variants to be identified as requiring laboratory tests using the family origin questionnaire</td>
<td>In high prevalence areas: ii) 98% of affected/ carrier individuals of thalassaemia and significant haemoglobin variants to be detected by laboratory tests In low prevalence areas: iii) 98% of affected/ carrier individuals of thalassaemia to be detected by laboratory tests iv) 98% of affected/ carrier individuals of significant haemoglobin variants to be identified as requiring laboratory tests using the family origin questionnaire</td>
<td>National via reporting of incidents to the SCT Programme and detection of incidents by the newborn screening programme and national evaluation (newborn outcomes project)</td>
</tr>
</tbody>
</table>
Standards for the Antenatal Screening Programme

<table>
<thead>
<tr>
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</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>High quality antenatal laboratory service</td>
<td>i) 95% of laboratories with a turnaround time of 3 days or less for processing samples; ii) 99% provide a data return to the programme centre</td>
<td>i) 98% of laboratories with a turnaround time of 3 days or less for processing samples; ii) 99.5% provide a data return to the programme centre</td>
<td>Collected locally and collated nationally</td>
</tr>
</tbody>
</table>

AO2b To identify with specific sensitivity, specified conditions were prenatal diagnosis is undertaken 2 Generic theme 6

| | | Accuracy of testing sample for prenatal diagnosis | 99% sensitivity for Hb-SS, & Hb-SC and beta thalassaemia major at-risk or affected pregnancies 3 | 99.5% sensitivity for Hb-SS, & Hb-SC and beta thalassaemia major at-risk or affected pregnancies 3 | National via reporting of incidents to the SCT Programme and detection of serious incidents by the newborn screening programme |

Process

| | | The proportion of pregnant women offered screening by 10 weeks’ gestation | 50% of all pregnant women offered screening by 10 weeks, 0 days 4 | 75% of all pregnant women offered screening by 10 weeks, 0 days 4 | Links with KPI ST2- timeliness of test |

AP1 Timely offer of screening by 10+0 weeks of pregnancy Generic themes 2, 3 and 8

| | | Antenatal carrier results reported to women and advice on the result and offer of testing to baby’s father, with appropriate support | All carrier results to be reported to women (see AP4) | 95% of carrier results provided to women verbally and in writing, supported by appropriate materials | Links with KPI ST2- timeliness of test |

AP2i To ensure high quality pathway for women/ couples ‘at high risk’ of an affected pregnancy Generic theme 5

| | | Fathers of carrier women’s babies offered screening and counselled by trained health care professionals | All fathers of carrier women’s babies to be offered information about counselling and testing (see AP4) | 75% of fathers of carrier women’s babies provided with information supported by appropriate materials | Local |

AP2ii Fathers of carrier women’s babies offered screening and counselled by trained health care professionals

| | | All fathers of carrier women’s babies to be offered information about counselling and testing (see AP4) | All fathers of carrier women’s babies to be offered information about counselling and testing (see AP4) | All fathers of carrier women’s babies provided with information supported by appropriate materials | Local |
### Standards for the Antenatal Screening Programme

<table>
<thead>
<tr>
<th>Number</th>
<th>Objective</th>
<th>Criteria</th>
<th>Acceptable standard</th>
<th>Achievable standard</th>
<th>Level for monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td>AP3</td>
<td>To ensure timely intervention for women/couples 'at high risk' of an affected pregnancy</td>
<td>Results and counselling of woman within 5 days of PND procedure</td>
<td>90% informed and counselled regarding PND result within 5 days of diagnostic test (also see AP4)</td>
<td>95% informed and counselled regarding PND result within 5 days of diagnostic test (also see AP4)</td>
<td>Local</td>
</tr>
<tr>
<td>AP4</td>
<td>Appropriate counselling for at-risk women/couples</td>
<td>Women/couples counselled by personnel with appropriate training and skills</td>
<td>80% of at risk couples to be counselled by PEGASUS or equivalent accredited trained personnel for 'at-risk' couple counselling 5</td>
<td>95% of at risk couples to be counselled by PEGASUS or equivalent accredited trained personnel for 'at-risk' couple counselling 5</td>
<td>Local and national</td>
</tr>
</tbody>
</table>

### Structure

<table>
<thead>
<tr>
<th>Number</th>
<th>Objective</th>
<th>Criteria</th>
<th>Acceptable standard</th>
<th>Achievable standard</th>
<th>Level for monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td>AS1</td>
<td>Responsibility for the programme delivery is clear across all professional boundaries, with a named individual responsible for each step of the screening pathway</td>
<td>Joint failsafe policy and arrangements for routinely checking and acting upon positive results established in laboratory and maternity services 6</td>
<td>Local programmes to ensure that a joint review of screen positive results across the linked antenatal and newborn pathway occurs on a regular basis. This could be a virtual clinic 7. 80% to be held at least quarterly</td>
<td>Local programmes to ensure that a joint review of screen positive results across the linked antenatal and newborn pathway occurs on a regular basis. This could be a virtual clinic 7. 90% to be held at least quarterly</td>
<td>Local</td>
</tr>
</tbody>
</table>

### Notes:

1. This standard assumes systems are in place to follow up samples received in the laboratory without a completed FOQ.

2. Specified conditions for antenatal screening: **Significant maternal Haemoglobinopathies:** Hb SS and other genotypes of sickle cell disease (Hb SC, HbS/β thalassaemia, etc) β thalassaemia intermedia Hb H disease (–/α-) (β-thalassaemia major will be clinically apparent); **Carrier states in mother:** Hb AS, Hb AC, Hb AD<sup>αα</sup>, Hb AE, Hb AO<sup>αα</sup>, Hb A Lepore, β thalassaemia trait, β-β thalassaemia trait, α<sup>α</sup> thalassaemia trait (–/αα), HPFH; **Significant disorders to be detected in the fetus:** Hb SS, Hb SC, Hb<sup>SD<sup>αα</sup></sup>, Hb SE, Hb SO<sup>αα</sup>, Hb S,Lepore and Hb β<sup>β</sup>Lepore, Hb S/β thalassaemia, Hb S/β thalassaemia, Hb Bart’s Hydrops Fetalis (–/–), β thalassaemia major (except cases with silent or near silent maternal phenotype), HbE/β thalassaemia; HbS/HPFH; Any compound heterozygote state including one or more of the above conditions; Any homozygous state of the above conditions.

3. For other conditions requiring detection by PND evidence on sensitivity is not available.

4. Not all units are currently able to assess offer of screening by 10,0 weeks. More units can provide data on testing by 10,0 weeks and this is used as a proxy measure for offer of screening by 10,0 weeks in KPI ST2. If the maternity data set is accepted, it will be possible to measure offer by 10,0 weeks.

5. This includes follow-up for those undergoing prenatal diagnosis and those not choosing prenatal diagnosis. An alternative equivalent course for 'at risk' couple counselling is “Genetic risk assessment and counselling module” run by King’s College London [http://sct.screening.nhs.uk/externaltraining](http://sct.screening.nhs.uk/externaltraining)

6. This should link with failsafe information on Map of Medicine, which can be found at [http://sct.screening.nhs.uk/carepathways](http://sct.screening.nhs.uk/carepathways)

7. This group should review timely actions taken on screen positive results such as (i) at least three attempts to contact screen positive women and the baby's father; (ii) for ongoing pregnancies ensuring the antenatal screen results are included on the blood spot card; (iii) review gestation at offer of antenatal screening.
Section 7
Standards for the Newborn Screening Programme
Standards for the Newborn Screening Programme

These standards should be read in conjunction with the Standards for the Linked Antenatal and Newborn Screening Programme and the UK Newborn Screening Programme Centre Standards: http://newbornbloodspot.screening.nhs.uk/getdata.php?id=10940.

A broken underline indicates that a term is used according to its definition in the glossary in Section 4.

<table>
<thead>
<tr>
<th>Number</th>
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<th>Achievable standard</th>
<th>Note</th>
<th>Level for monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Outcome</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>NO1</td>
<td>Best possible survival for infants detected with sickle cell disease by the screening programme ¹</td>
<td>Mortality rates expressed in person years</td>
<td>Mortality rate from sickle cell disease and its complications in children under five of less than 4 per 1000 person years of life (2 deaths per 100 affected children)</td>
<td>Mortality rate from sickle cell disease and its complications in children under five of less than 2 per 1000 person years of life (1 death per 100 affected children)</td>
<td>National evaluation via newborn outcomes project</td>
<td></td>
</tr>
</tbody>
</table>

¹ Generic Theme 8

| NO2i | To identify, with specified sensitivity babies born with conditions where early intervention is likely to be beneficial ¹ | Sensitivity of laboratory methods at detecting affected and carrier individuals | 99% detection for Hb-SS 98% detection for Hb-SC 95% detection for other conditions | 99.5% for Hb-SS 99% detection for Hb-SC 98% detection for other conditions | National evaluation using audit and incident data | |

¹ Generic Themes 4 and 6

| NO2ii | Coverage Generic Theme 2 | Coverage of screening test (tested/eligible) | 95% of babies eligible for newborn sickle cell screening receive a conclusive screening test as part of bloodspot screening | 99% of babies eligible for newborn sickle cell screening receive a conclusive screening test as part of bloodspot screening | In addition, information on refusal rate and monitoring by ethnic group obtained from joint return with newborn bloodspot programme | |

² Generic Themes 4 and 6

| **Process** | | | | | | |
| NP1 | Reliable screening sample on all transfused babies ³ | Result received on babies requiring tests outside normal testing time due to blood transfusion | Of babies needing an early transfusion, 95% should have a pre-transfusion sample taken | Of babies needing an early transfusion, 98% should have a pre-transfusion sample taken | Links with bloodspot standard 3, guideline 7 | Local monitoring of rate of transfusion samples by laboratory and local area |

³ Generic theme 4

| NP2i | To report results of screening including carrier results in a timely manner ³ | All results to be routinely reported to parents, GPs and health visitors by six weeks of age | 95% screen negative results, including haemoglobinopathy carrier, available for communication by 6 weeks of age | 98% screen negative results, including haemoglobinopathy carrier, available for communication by 6 weeks of age | Links with Bloodspot KPI | KPI NB3-timeliness of result availability |

³ Generic theme 5

| NP2ii | Information given to parents of carrier babies | 80% of parents of carrier babies given written information ⁴ ideally during a face-to-face discussion by trained healthcare professionals | 90% of parents of carrier babies given written information ⁴ ideally during a face-to-face discussion by trained healthcare professionals | | Local audits | |
## Standards for the Newborn Screening Programme

<table>
<thead>
<tr>
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</tr>
</thead>
<tbody>
<tr>
<td>NP3</td>
<td>Timely communication of positive screening results (sickle cell disease) – including a review of parental results</td>
<td>Time by which affected baby results are communicated to parents</td>
<td>90% of sickle cell disease results communicated to parents by 4 weeks of age</td>
<td>95% of sickle cell disease results communicated to parents by 4 weeks of age</td>
<td>Links with clinical standards</td>
<td>Local with clinical networks (for example, as recorded by some newborn laboratories)</td>
</tr>
<tr>
<td>NP4</td>
<td>Effective follow-up of infants with positive screening results (sickle cell disease) – all babies to be registered with a local clinic / centre (or clinic working as part of clinical network)</td>
<td>Completeness of infants with a positive screening result followed up and entered into care</td>
<td>90% of babies identified are referred by eight weeks to a designated healthcare professional</td>
<td>95% of babies identified are referred by eight weeks to a designated healthcare professional</td>
<td>Links with clinical standards</td>
<td>Local with clinical networks</td>
</tr>
<tr>
<td>NP5</td>
<td>Timely confirmation of diagnosis for infants with a positive screening result for specified conditions</td>
<td>Diagnostic confirmation of newborn screening results</td>
<td>90% of cases of Hb SS &amp; Hb SC have confirmation of result documented in clinical notes by six months of age</td>
<td>95% of cases of Hb SS &amp; Hb SC have confirmation of result documented in clinical notes by six months of age</td>
<td>Links with clinical standards</td>
<td>Local with clinical networks</td>
</tr>
<tr>
<td>NP6i</td>
<td>To ensure treatment is offered and parental education started in a timely manner for children with conditions as specified in the clinical standards</td>
<td>Offer and prescription of Prophylactic penicillin (or alternative)</td>
<td>90% offered and prescribed Penicillin V (or alternative) by three months</td>
<td>99% offered and prescribed Penicillin V (or alternative) by six months</td>
<td>Links with clinical standards</td>
<td>Local with clinical networks</td>
</tr>
<tr>
<td>Number</td>
<td>Objective</td>
<td>Criteria</td>
<td>Acceptable standard</td>
<td>Achievable standard</td>
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</tbody>
</table>
| NP6ii  | Communication to professionals by clinical network or named health professional  
Generic theme 7 | Communication about infant with a positive screening result | 85% of GP and HVs informed of infants with a positive screening result by four weeks of age | 95% of GP and HVs informed of infants with a positive screening result by four weeks of age | | Local with clinical networks |
| NP6iii | Communication to parents  
Generic themes 5 and 7 | Education of parents about screen positive results and provision of information in an accessible format/language | 97% of families offered verbal and written information on condition, follow-up, and treatment at first visit with named professional | 99% of families offered verbal and written information on condition, follow-up, and treatment at first visit with named professional | | Local |
| NP7i   | Minimise the adverse effects of screening – eg. anxiety, misunderstanding, dissatisfaction  
Generic theme 5 | Consistent communication about results and action required for positive screening results and carrier results | 80% of laboratories and Child Health Teams to use standard reporting including letters for results (when available). This will include status codes (v.2) once they are available | 90% of laboratories and Child Health Teams to use standard reporting letters for results (when available). This will include status codes (v.2) once they are available | | Local |
| NP7ii  | To ensure that responsibility, governance and performance management for all aspects of the programme are clear and these link together between the newborn and antenatal programme  
Generic theme 10 | Each local programme has explicit governance arrangements at all levels of the programme and there are established links between the antenatal and newborn programmes (http://sct.screening.nhs.uk/linkage) | 95% of local programmes to conduct a regular joint review to ensure that screen positive results across the pathway have been appropriately acted upon | 97% of local programmes to conduct a regular joint review to ensure that screen positive results across the pathway have been appropriately acted upon | | Local |
### Standards for the Newborn Screening Programme

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<tr>
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</tr>
</thead>
<tbody>
<tr>
<td><strong>NS1</strong></td>
<td><strong>Failsafe to ensure ongoing care</strong>&lt;br&gt;Generic theme 10</td>
<td>Failsafe checks in clinical networks to check if all children with a positive screening result (Sickle Cell Disease) attend for follow-up and action is taken where they do not</td>
<td>As specified in the model service specifications, newborn laboratories should report screen positive results locally for action and to the designated clinical network centre for oversight. The designated clinical centre is responsible for checking the baby has entered the care pathway</td>
<td></td>
<td></td>
<td>Local with clinical network</td>
</tr>
</tbody>
</table>

**NS2**<br>**Quality newborn laboratory screening service**<br>Generic themes 4 and 6

<table>
<thead>
<tr>
<th>Objective</th>
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</tr>
</thead>
<tbody>
<tr>
<td>High quality newborn laboratory service&lt;br&gt;Newborn laboratories meet minimum laboratory criteria defined in Laboratory Handbook.</td>
<td>i) 95% of laboratories use standard reporting&lt;br&gt;ii) 98% share data with the screening programme&lt;br&gt;iii) 95% working towards using newborn status codes</td>
<td>i) 99% of laboratories use standard reporting&lt;br&gt;ii) 99% share data with the screening programme&lt;br&gt;iii) 99% working towards using newborn status codes</td>
<td>Local and national</td>
</tr>
</tbody>
</table>

### Notes:

All standards (except those in italics) are the responsibility of the NHS Sickle Cell and Thalassaemia Screening Programme.

*Standards in italics are incorporated as part of UK Newborn Screening Programme Centre standards and monitoring arrangements.*

http://newbornbloodspot.screening.nhs.uk/getdata.php?id=10940

1. Specified conditions to be detected in newborn screening: HbSS, HbS/β thalassaemia (β+, β+, β-, Lepore), HbS/HPFH, HbSC, HbSD*/αα*, HbS/E, HbS O/Are and other clinically significant Haemoglobinopathies likely to be detected as by-products by newborn screening including β thalassaemia major, Hb E/β thalassaemia, β thalassaemia intermedia and HbH disease

2. At present KPI NB1- coverage (PCT responsibility at birth) assesses coverage for PKU as a proxy indicator for sickle cell screening.

3. These standards should be read in conjunction with the UK Newborn Screening Programme Centre Standards (2008) http://newbornbloodspot.screening.nhs.uk/getdata.php?id=10940


7. Planned for 2012
Section 8
Standards for the linked Antenatal and Newborn Screening Programme
## Standards for the linked Antenatal and Newborn Screening Programmes

<table>
<thead>
<tr>
<th>Number</th>
<th>Objective</th>
<th>Criteria</th>
<th>Acceptable standard</th>
<th>Achievable standard</th>
<th>Level for monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td>LO1</td>
<td>To facilitate an appropriate level of understanding, knowledge and skills among professionals involved with the screening programmes at all levels Generic theme 9</td>
<td>Assessment of coverage of education programmes</td>
<td>80% of key staff in a unit attend educational programmes appropriate to their involvement in the screening programmes on an ongoing basis</td>
<td>95% of key staff in a unit attend educational programmes appropriate to their involvement in the screening programmes on an ongoing basis</td>
<td>Assessed by: - use of e-learning resource - annual Education and Training audit - attendance at national training programmes (collected nationally)</td>
</tr>
<tr>
<td>LO2</td>
<td>To minimise the adverse effects of screening Generic themes 5 and 9</td>
<td>Use of national information for parents to be provided before and after screening. Availability of accessible and standardised materials in maternity units &amp; key personnel responsible for giving antenatal and newborn screening information in appropriate languages</td>
<td>97% of maternity units able to access written and verbal information to parents and other healthcare professionals in the pathway. This will include direction to programme website for web-based information and translations</td>
<td>100% of maternity units able to access written and verbal information to parents and other healthcare professionals in the pathway. This will include direction to programme website for web-based information and translations</td>
<td>Local, assessed by asking if a sample of users are aware of information about screening. This could be part of overall Trust review of user experience</td>
</tr>
<tr>
<td></td>
<td>Linking antenatal and newborn results</td>
<td>An alert system should be in place to inform newborn laboratories of at risk couples 1</td>
<td>An alert system should be in place to inform newborn laboratories of at risk couples and maternal carrier results 1</td>
<td>Local</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Parental satisfaction with the experience of screening and follow-up</td>
<td>Surveys of parents’ satisfaction and attitudes</td>
<td>70% of maternity units conduct surveys of user experience across the linked screening programme at least once every 3 years</td>
<td>90% of maternity units conduct surveys of user experience across the linked screening programme at least once every 3 years</td>
<td>Local survey</td>
</tr>
</tbody>
</table>
### Standards for the linked Antenatal and Newborn Screening Programmes

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</thead>
<tbody>
<tr>
<td>LO3</td>
<td>Education and awareness of the value of screening to be provided to the wider population in an acceptable and accessible manner, with the aim to reduce the stigma sometimes associated with the conditions screened for Generic themes 3 and 5</td>
<td>Community awareness, understanding and acceptance of Antenatal and Newborn Screening Programmes</td>
<td>The national programme to develop materials to support community awareness and understanding of screening programmes.</td>
<td></td>
<td>National by survey and quantitative assessment</td>
</tr>
<tr>
<td>LO4</td>
<td>To provide regular feedback on the effectiveness and quality of the service to the population screened and the providers of the service Annual reports available and collated Generic theme 10</td>
<td>Acceptable format of feedback provided to professionals and the public</td>
<td>95% of maternity units and associated provider Trusts to produce annual reports of linked Antenatal and Newborn Programme. Report to include coverage, timeliness and entry into care</td>
<td>100% of maternity units and associated provider Trusts to produce annual reports of linked Antenatal and Newborn Programme. Report to include coverage, timeliness and entry into care</td>
<td>Local</td>
</tr>
</tbody>
</table>

### Structure

<p>| LS1    | To ensure that responsibility and accountability for all aspects of the programme are clear, with direct links from National Programme accountability through regional teams to local level accountability Generic theme 10 | Each programme has explicit arrangements for accountability and responsibility across the programme and there is an established linkage between the antenatal and newborn programmes as described in linkage policy (Appendix One) | Trusts have regular meetings to review accountability and responsibility across linked SCT Antenatal and Newborn Programme, including failsafes and report areas for development (see also Antenatal standard AS1 and newborn standard NP7ii) 80% to be held at least quarterly | Trusts have regular meetings to review accountability and responsibility across linked SCT Antenatal and Newborn Programme, including failsafes services and report areas for development (see also Antenatal standard AS1 and newborn standard NP7ii) 90% to be held at least quarterly | Locally as evidenced by minutes of meetings |</p>
<table>
<thead>
<tr>
<th>Number</th>
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<th>Criteria</th>
<th>Acceptable standard</th>
<th>Achievable standard</th>
<th>Level for monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td>LS2i</td>
<td>To evaluate the service on an ongoing basis to assist continuous quality improvement</td>
<td>95% of Trusts to produce annual reports. These can be part of Trust annual reports for antenatal and newborn screening and should be circulated to commissioners, regional teams and National Programme (see also Linked standard LO4)</td>
<td>100% of Trusts to produce annual reports. These can be part of Trust annual reports and should be circulated to commissioners, regional teams and National Programme (see also Linked standard LO4)</td>
<td>Local</td>
<td></td>
</tr>
<tr>
<td>LS2ii</td>
<td>Local programmes to ensure that a joint review of screen positive results across the linked antenatal and newborn pathway occurs on a regular basis. 80% to be held at least quarterly</td>
<td>Local programmes to ensure that a joint review of screen positive results across the linked antenatal and newborn pathway occurs on a regular basis. 90% to be held at least quarterly</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>LS3</td>
<td>To make effective and efficient use of resources to the benefit of the population based on up-to-date research evidence Generic theme 10</td>
<td>Review and report in national annual report on relevant policy and research issues each year</td>
<td>Review of programme against published evidence eg. HTA reports. Select areas for review as criteria, and targets according to progress and evidence base in this area To ensure that evidence on cost-effectiveness (screening technologies &amp; treatment) is incorporated into the design of the National Programme To review new relevant evidence on a systematic basis annually To make the NHS Sickle Cell and Thalassaemia screening programme a model programme worldwide and specifically in Europe</td>
<td></td>
<td>National</td>
</tr>
</tbody>
</table>

1. The direction of travel is to include antenatal screening results in a systematic manner on the Bloodspot card. It is anticipated that the 2011/12 consultation will provide space to record antenatal screening results on the Bloodspot card.
Section 9
Arrangements for the national screening programmes

A Roles and responsibilities of national screening programmes
B Quality aspects for the National Programme
C Support structures for the National Programme
A Roles and responsibilities of effective national screening programmes

A cross-programme approach to quality assurance (QA) for the non-cancer screening programmes has been agreed by the Regional Directors of Public Health (RDsPH). This approach is based on development of regional level structures to coordinate and combine QA for the non-cancer screening programmes. This will allow a consistent approach across the eight non-cancer screening programmes to enable RDsPH, SHA and PCT screening leads and commissioners to understand the structures and QA issues of each programme. To ensure efficiency in this system we need to ensure the role of all stakeholders is clear so that there are no duplications or gaps in oversight of the screening pathway.

A statement has been agreed with the aim of providing clarity on what should be done at a national level [89]. The roles and responsibilities at national level will need to link with roles and responsibilities for regional and local teams to ensure that there are not a myriad of conflicting requests on QA. We also need to be mindful of the changing role of SHAs and associated organisations in the current NHS reorganisation.

National screening programmes are responsible for things that are best done once across the country. For example setting standards or providing information about screening developments. This needs to be followed by good local dissemination to ensure an efficient process is in place that also meets local needs.

So what does this mean in practice? The list below is not exhaustive but includes areas that fall within the remit of national programmes:

1. **Setting standards:** The SCT Programme is responsible for setting programme standards against which performance can be managed, with a transparent process for reviewing and resetting standards. The standards are reviewed on a three-yearly cycle under the remit of the Steering Group and in conjunction with UK NSC work streams.

2. **Provide expertise and advice:** The SCT Programme is seen as the expert on all aspects of the screening programme. This expertise will be informed by published literature, examples of best practice and views of stakeholders. The process for agreeing screening policy is through the UK National Screening Committee.

3. **Policy review:** The SCT Programme is responsible for identifying areas of policy requiring review, using their expertise and other evidence. The SCT Programme is responsible for taking these proposals through the policy review process and providing advice on implementing any resultant changes.

4. **Equality in delivery of a screening programme:** The SCT Programme has a responsibility to consider equality issues arising from local implementation of the national screening programmes.

5. **Serious incidents:** The SCT Programme provides expertise and advice in the management of screening incidents. This includes advice on what is an incident; how to investigate an incident; how to prevent the incident affecting additional people; providing information to healthcare professionals and users about the incident; providing information to local and national communications teams about the incident; and monitoring progress on resolving an incident. The expertise and perspective at a national level can often identify patterns that may not emerge at a local level and national action may also be required (e.g. equipment failures).

6. **Dissemination of lessons learnt:** The SCT Programme takes the lead on disseminating lessons learnt from serious incidents. Information on lessons learnt for the SCT Programme is available at http://sct.screening.nhs.uk/quality-improvement

7. **Failsafe:** The SCT Programme has specified failsafe points and actions across the screening pathway on the Map of Medicine. These are specified at http://sct.screening.nhs.uk/carepathways For failsafe to be effective there needs to be local action.

8. **Implementation:** The SCT Programme monitors progress of the implementation of policy changes.
and quality improvement assurance processes. At the moment this includes:

- Reporting on national trends and comparison of data across SHAs and PCTs as appropriate, to inform local performance management
- Monitoring overall effectiveness and outcomes of the screening programme
- Monitoring test performance in specialist areas such as three prenatal diagnostic laboratories for SCT screening
- Observing the functioning of the regional EQA to provide comment on the quality of the service provided
- Specifying information, management and technology (IM&T) requirements and a minimum dataset. This can include national procurement to ensure consistency of data collection.

9. **Training:** Training and Education sits within an agreed UK NSC Education Framework. The SCT Programme identifies needs in skills and competencies and ensure these are addressed through appropriate channels such as professional bodies.

10. **Peer review:** The SCT Programme ensures that specialist aspects of peer review (specific to each programme) are integrated and compatible with the regional peer review cycle. National data will be used to inform local peer review.

11. **Communications:** The SCT Programme produces a range of materials to support the screening programme for people offered screening, and for healthcare professionals providing screening. The SCT Programme also works with the UK NSC communications team to provide information and communication about generic screening issues.

12. **Equipment:** The SCT Programme produces specifications for equipment, and input into national framework procurement.

13. **Commissioning:** The SCT Programme commissions some nationally-relevant services to support local screening programmes (eg. EQA, national software systems) but does not directly commission services. The SCT Programme acts as a source of expertise for commissioners across the screening pathway.

14. **Audit:** The SCT Programme conducts or commissions specific national audits & reports – for example, the evaluation of the PEGASUS training course.

15. **Research:** The SCT Programme recommends research topics for consultation by appropriate bodies such as the Health Technology Assessment (HTA).

B Quality aspects for the National Programme

The Quality Improvement work streams for the Sickle Cell and Thalassaemia Screening Programme are detailed in the QI plan, which is intended to provide clarity on development and implementation in this area over the next three years. The aim is to improve screening so that pregnant women and their families are offered safer, higher quality screening and a better user experience. This encompasses user engagement and reduction of health inequalities.

Development of the SCT Programme QI Plan is informed by six areas of work, and by the expertise and valuable contributions of public and professionals across the country. We thank them warmly for their help and support.

1. **The SCT programme policy.** The SCT Programme is a linked Antenatal and Newborn Screening Programme (http://sct.screening.nhs.uk/linkage) delivered across professional and organisational boundaries.

2. **The UK National Screening Committee (NSC) plan on Quality Assurance** (http://www.screening.nhs.uk/quality-assurance). The NSC plan aims to provide assurance to Regional Directors of Public Health that the non-cancer screening programmes are operating effectively. This means that across all non-cancer screening programmes, the quality of services remains high in order to maintain their effectiveness and safety.

3. **The Donabedian Principles of Quality Assurance.** Donabedian identifies the need for quality to include...
Structure, Process and Outcomes of the screening pathway [11]. The mechanism to measure and monitor quality improvement is to identify clear aims and objectives for the screening programme. The aims and objectives are supported by measurable criteria, all with associated achievable and acceptable standards (http://sct.screening.nhs.uk/cms.php?folder=2493).

4. The governance structure of the SCT programme.
The SCT Programme is advised by a steering group with a ministerially appointed lay chair. This group has identified that the term ‘quality assurance’ was not clear to the general public. Therefore the SCT Programme use the term Quality Improvement (QI) to describe this area of work.

5. Cross-programme principles. The SCT Programme QI team is actively engaged with cross-programme groups. It works on the principle that quality assurance arrangements should be made once across all programmes and not duplicated – resulting in increased efficiency and greater leverage than a QA system based on individual programmes. The cross-programme approach is based on development of regional level structures to coordinate and combine QA for the non-cancer screening programmes. The SCT Programme Director is chair of the Routine Reporting Task Group, and the SCT Deputy Director is chair of Programme’s Only QA Group and vice-chair of the Cross-Programme QA Group. The SCT QI team is represented at PIPD and also works closely with the UK Newborn Screening Programme Centre through the Joint Working Group, Bloodspot Advisory Group and UK Laboratory Quality Assurance Development Group. More information on the UK NSC Cross-Programme QA Group and cross-programme principles can be found at http://www.screening.nhs.uk/qa-group.

6. Commissioning. The commissioning of screening programmes varies across the country and across the screening pathway. This results primarily from the complexity of screening pathways which involve primary, secondary and tertiary providers who are supported by a range of different clinical and non-clinical data management systems. It is anticipated that future oversight by the NHS Commissioning Board will improve consistency and co-ordination of commissioning of screening programmes.

Currently, until April 2013, Primary Care Trusts are responsible for securing and funding population screening programmes, and are required to collaborate and establish specialised services commissioning as appropriate for some elements of programmes (e.g. laboratory services). SHAs are required to ensure that commissioning arrangements are robust, and to monitor performance against national standards.

However, the organisational context in which screening programmes are commissioned and managed is set to change, following the publication of the Public Health White Paper Healthy Lives Healthy People [90] and Equity and Excellence: Liberating the NHS [13]. Subject to legislation, PCTs and SHAs are to be abolished in 2013. Their commissioning responsibilities will be taken over by new Clinical Commissioning Consortia and the NHS Commissioning Board (NHSCB). In addition, a new public health service – Public Health England – is to be established and its proposed responsibilities will cover screening including mandating the NHSCB to commission screening services from a devolved budget.

The operational details of the new commissioning system have yet to be fully defined and confirmed in statute. However, the SCT programme is well placed to support the ongoing provision of high quality screening during the transition period and to provide expert advice to the next generation of commissioners in the form of generic service specifications, quality assurance frameworks, and maintenance of clearly defined programme standards – as laid out in the Quality Improvement (Assurance) Plan: 2011/12-2013/14.
C Support structures for the National Programme

1. Education and training
The SCT Programme Education and Training Framework is informed by the National Screening Committee (UK NSC) strategy on Education and Training. The UK NSC strategy aims to “ensure individuals offered screening within the NHS are fully informed of their choices and receive the best possible care and experience along the screening pathway”. This aim is underpinned by the UK NSC principles of support, integration, sustainability, inclusivity, shared learning and a sound evidence base.

The SCT Education and Training Framework is also informed by other SCT programme policies including:

• The SCT Programme mission to develop a linked programme of high quality screening and care

• The SCT Programme training and education strategy, which states: “The Programme’s educational reach covers any member of the wider health and social care team who may have any reason to be involved with members of the public and/or patients in the context of a discussion about genetic and more specifically haemoglobinopathy related conditions”. The strategy also identifies that the Training and Education Framework will be informed by evaluation.

Programme specific training initiatives include:

• one-day laboratory training for antenatal laboratories. These are supported by web-based training material and informed by the laboratory support service

• one-day training for reporting newborn carrier results

• web-based training for front line professionals, including GPs and other primary care staff

• web-based training for those counselling couples at risk of an affected pregnancy

• five-day specialist training for those counselling couples at risk of an affected pregnancy

Further details can be found at www.screening.sct.nhs.uk

The SCT Programme has also procured a formal laboratory support service to allow individuals to contact experts in laboratory aspects of haemoglobinopathy screening and testing for individual advice. This service is provided by The National Haemoglobinopathy Reference Service at the Oxford Radcliffe Hospital NHS Trust, which will provide support services for screening laboratories across England. The new service started in October 2010 and can be accessed by email, telephone or fax. Details are at http://sct.screening.nhs.uk/cms.php?folder=2464#fileid11139

UK NSC education and training initiatives:
Training and Education for the SCT Programme sits within the UK NSC Education Strategy. Examples of work being led by the UK NSC include the e-Module on antenatal and newborn screening for midwives, launched in February 2011; the DH E-Learning for Health module on the ‘Healthy Child Programme’ – of which UK NSC has written four screening modules, one relating to SCT; and a recently commissioned BMJ online module which will cover all AN/NB programmes, and will cover the SCT key messages (provided by the Programme).

Further details of the UK NSC work on training and education can be found at http://cpd.screening.nhs.uk/

2. Communications
It is important that service providers are aware of resources to support both professionals and the public and that there is consistency of message to professionals and the public.

An integrated communications strategy has been developed to support a range of information needs:

• A wide range of health professionals, administrative staff and in some cases volunteers deliver the programme. They need to understand the aims and remit of the programme, their role within the overall patient journey and how best to support the public along that journey
• People offered screening need to understand the screening offer, what screening can tell them (and what it cannot) the choices that may arise and where to find further information and support

• Both the public and health professionals need to understand the aims and benefits of offering screening and to find the screening programme acceptable

• To support all these communications needs, we work to create a baseline of understanding both about genetic screening generically and about the conditions for which we are screening.

Materials for the public and for health professionals:
A wide range of information has been developed in a range of formats and media including printed materials, information on the web, and DVDs. Significant effort has been put into making these accessible to a range of audiences by providing translations, large print, Braille and audio versions of key documents to make the service equitable. Materials developed to date can be accessed at http://sct.screening.nhs.uk/publications.htm. The SCT Programme have a series of filmed real life stories from carriers, patients and carers who give their account of what sickle cell or thalassaemia means to them. This is available at http://sct.screening.nhs.uk/stories. The SCT Programme reaches out to professionals through exhibitions, conferences, workshops and a range of newsletters and briefings.

In addition, the healthtalkonline (www.healthtalkonline.org) contains information and experiences of those screened (written, audio and video) aimed at both health professionals and women/couples.

Public outreach & engagement:
We have developed an extensive programme of direct engagement with communities at higher risk of being carriers for these conditions. This is helping to create a baseline understanding about the conditions and the screening offer. It is intended both to ensure the Programme is acceptable to the communities it serves, and also to support people’s ability to make informed choices about screening. This work particularly targets people who normal NHS communications channels may fail to reach, and aims to develop sustainable partnerships in the voluntary sector and other health services.

Media relations:
We work with both the professional and public-facing media to promote baseline understanding and to publicise specific developments in services or materials available. We have particularly targeted media addressing black and minority ethnic audiences.

Special projects:
These are developed where there is a particular need to focus on specific audiences. Currently we are encouraging more men to participate in screening, and working with primary care to support screening processes.

The voluntary sector (specialist and generalist) also plays an important part in communication, and their involvement should be incorporated whenever possible.
Section 10
Guidance on local organisational arrangements for the screening programmes

A. Policy
B. Support structures for the Programme
C. Infrastructure requirements
D. Clinical and specialist support
E. Quality aspects of the Programme
A Policy

1. Policy for the Screening Programme:
Each provider organisation involved in delivering the Programme requires a written policy (integrating all relevant antenatal and newborn screening for which it is responsible), which applies national policy and guidance to the local situation and which provides contact details for key individuals responsible for specific aspects of the screening pathway. This needs to link with the UK NSC commissioning frameworks for antenatal and newborn screening programmes [91].

2. Specific policy for review:
Local written policies must be reviewed annually - and incorporate a joint review of newborn and antenatal policy and services including linkages between the antenatal and newborn screening.

The delivery of the Antenatal Screening Programme must ensure that sickle cell and thalassaemia screening (along with offer of screening) is integrated as part of general antenatal care and is not delivered in isolation. This specifically relates to the offer of screening, review of results and to invasive procedures.

Each maternity trust/unit must have a named individual responsible for the overall coordination of the Antenatal and Newborn Programme who is ultimately accountable to the chief executive.

Robust links and two way data transfer arrangements between Primary Care and maternity services need to be established to ensure that the objectives of the Antenatal Screening Programme are realised – ie. Offer of sickle cell/thalassaemia screening before 10 weeks of pregnancy, therefore enabling the offer of, uptake of and reporting of results of prenatal diagnosis and any subsequent action by the end of 13 weeks of pregnancy.

Each of the Trusts with a newborn screening laboratory must have a named individual responsible for the overall co-ordination of the linked antenatal and newborn programme accountable to the chief executive.

All local screening programmes and maternity services must identify named individuals responsible for each aspect of the antenatal and newborn screening pathway, including performance management. Each aspect of the screening pathway, along with failsafe points, is clearly outlined on a Map of Medicine pathway, which can be found at http://sct.screening.nhs.uk/carepathways

Details of the Screening Programme policies for antenatal and newborn screening can be found at http://sct.screening.nhs.uk/Policy.htm.

B Support for the Programme:

1. Education and training:
All those directly involved in the provision of antenatal and newborn screening information or services should have an induction to the Programme, and must undertake regular updating in line with continuing professional development guidance for their profession, and guidance specified by the UK NSC, which can be found at http://cpd.screening.nhs.uk/

All professionals providing newborn and antenatal screening information and services should have received the appropriate education for their roles and responsibilities and any specific tasks required in the area in which they work as specified by the UK NSC. This includes midwives, health visitors, primary care teams, medical practitioners and laboratory staff.

Additional training for more specific aspects of the programme such as specialist counselling for ‘couples at risk’ of an affected pregnancy is required. Professionals with this remit must attend the designated training course. More information can be found at http://sct.screening.nhs.uk/education

The UK NSC is working to provide improved and more extensive training about both the Screening Programme and care within routine undergraduate training and relevant postgraduate programmes. The aim is to ensure that students across the country – irrespective of local policy – are exposed to these programmes and conditions in recognition of the increasing size of the health need, and the national nature of the screening programmes.
Additional training for registrars in haematology, paediatrics and obstetrics is required to support the development of clinical capacity to meet the demands and expectations arising from the Programme.

Established haematologists, obstetricians and paediatricians also require appropriate training through continuing professional updates.

2. Information for women and the baby’s father:

Antenatal screening
All women and their babies’ fathers should receive information about antenatal screening options as early as possible in pregnancy, before they are asked to make any screening decision. All women and their babies’ fathers should be given an opportunity to discuss the decisions that they have made for antenatal screening with a professional who is appropriately trained and informed about the condition(s).

The information provided verbally and by other means should be consistent with the national requirements on standard reporting of results and language, such as the term ‘carrier’ and arrangements for the process of the Screening Programme.

All women and their babies’ fathers should be provided with accurate information on the likely timing of receipt of results following uptake of screening. This means ensuring that professionals have a clear understanding of the timing and arrangements for post-screening reporting in their local area.

Newborn screening
All women and their baby’s father with an infant born with a positive screening result (affected baby) should receive information about the result from a professional, who is appropriately trained and informed about the condition, and who has access to the relevant antenatal screening information. Women and their baby’s father should also be provided with relevant material, in acceptable formats and language to allow them to reflect on this information giving session. The family should be referred to an appropriate clinical network.

All women and the baby’s father with an infant who has a carrier result should be informed of the result, ideally face-to-face (this information should not be reported by laboratories as ‘normal’), receive relevant information and material about the result and, as a minimum, be offered access to an appropriately-trained health professional to discuss the result.

Information materials
A wide range of information has been developed in a range of formats and media including printed materials, information on the web, and DVDs. Materials developed to date can be accessed at http://sct.screening.nhs.uk/publications.htm.

The antenatal pre-screening leaflet Screening tests for you and your baby [88] developed by the National Screening Committee should be offered to all women at first contact prior to screening. This can be found at http://sct.screening.nhs.uk/public-resources

Translations about antenatal and newborn sickle cell and thalassaemia screening from above booklets are available in following languages:

- Arabic
- Bengali
- French Français
- Greek ελληνικά
- Gujarati
- Hindi
- Kurdish
- Polish język polski
- Portuguese português
- Romanian română
- Russian русский язык
- Simplified Chinese
- Somali
3. Support for women and their baby’s father:

Women must be informed of the genetic nature of screening for both the newborn and the antenatal programme and the possibility that the test could reveal the paternity of the pregnancy (see Appendix 6 for guidance on non-paternity).

Counselling of couples identified as ‘at risk’ (i.e. both parents are identified as a carrier of a significant haemoglobin variant, or a woman where the baby’s father is not available) must be undertaken by professionals with specialist knowledge who have undertaken relevant training in the genetics and natural history of haemoglobin disorders. Details of appropriate courses can be found at [http://sct.screening.nhs.uk/education](http://sct.screening.nhs.uk/education).

Women who are carriers of haemoglobin diseases, and whose baby’s father is unavailable for screening, should be offered prenatal diagnosis (if wanted) following appropriate counselling by a trained professional (as above). Women should also be informed that the sensitivity of the DNA procedures might be reduced for haemoglobin variants other than Hb S and C because the baby’s father information is unavailable (see the Handbook for Laboratories [10] at [http://sct.screening.nhs.uk/cms.php?folder=2493](http://sct.screening.nhs.uk/cms.php?folder=2493)).

Health professionals should work collaboratively and consistently with relevant organisations including the voluntary sector, service users, religious organisations and other appropriate agencies. This is to ensure that as far as possible, mothers and fathers are provided with support after screening that is most appropriate to their needs and wishes.

C Infrastructure requirements:

1. Newborn and maternity records:

All documentation used must record the offer of screening and consent for each test undertaken.

Routine paper and electronic records used must ensure that space is available to record family origin (based on Family Origin Questionnaire categories for the mother and baby’s father).

Women and their baby’s father must be notified of all test results (antenatal and newborn) in writing, along with relevant materials as appropriate.

Maternal antenatal screening results should be recorded on the newborn screening blood spot card to assist linkage.

The antenatal report given to all women following the birth of their baby should contain all antenatal screening results for communication to primary care and to allow linkage to relevant newborn screening results and action where relevant. Sickle Cell and Thalassaemia screening results should be included within this report.

2. Laboratories:

Antenatal screening laboratories:

The antenatal screening laboratory must have the IT infrastructure to produce standard reporting as specified by the SCT Programme and to support the audit and monitoring requirements.

The laboratory should have links with the relevant newborn laboratory(ies) including named contacts, and should routinely report on all couples identified as ‘at risk’ along with other relevant antenatal information, to the newborn screening laboratory team through the local screening coordinator. (For further details please refer to Handbook for Laboratories [10] at [http://sct.screening.nhs.uk/cms.php?folder=2493](http://sct.screening.nhs.uk/cms.php?folder=2493)).

There must be a failsafe policy and arrangements in laboratory and maternity services for acting on and routinely reviewing positive screening results. Laboratories should work with maternity units to ensure that a
Guidance on local organisational arrangements for the screening programmes

Joint review of screen positive results across the linked antenatal and newborn pathway occurs on a regular basis. This could be a virtual clinic.

The antenatal screening laboratories are required to:
- Identify first antenatal blood ‘booking’ sample (and distinguish them from subsequent pregnancy, other non-pregnancy samples or later samples in the same pregnancy)
- Incorporate FOQ with antenatal booking bloods request
- Use SCT Programme approved paper FOQ
- Ensure that electronic FOQ provides adequate information (consult the SCT Programme)
- Link mother’s antenatal result to the baby’s father’s result

Following the Low Prevalence Antenatal Screening Status Codes Pilot (report available here: http://sct.screening.nhs.uk/cms.php?folder=2460) the SCT Programme is working on establishing technical feasibility of transfer of haemoglobinopathy antenatal status codes from laboratories to primary care. The SCT Programme is supporting and working towards enabling transfer of status codes to requesting sources.


Newborn screening laboratories:
The newborn screening laboratory is required to have the IT capacity to integrate with the central IT developments for newborn and antenatal screening; to report results in formats as specified by UK NSC and support newborn status code reporting; to support the development of routine notification of parents working with child health; and to support the audit requirements of the SCT Programme.

The laboratory must have named contacts from haematology and maternity services with all providers within its catchment area, and should routinely report newborn positive screening results, as recommended by the SCT Programme, in order to allow antenatal audit and monitoring.

Failsafe policies in laboratories and clinical networks must be in place, to check if all children with a positive screening result have been received by clinical network centres and act where they have not.

As stated in the model service specifications for clinical care (National Haemoglobinopathy Project http://sct.screening.nhs.uk/cms.php?folder=2558) newborn laboratories should report screen positive results locally for action and to the designated centre for a clinical network. The designated clinical centre is responsible for checking the baby has entered the care pathway


DNA laboratories:
Each DNA laboratory is required to be part of the National Genetics Testing Network including all the quality requirements of this network.

The laboratory is required to contribute to the national audit of prenatal diagnosis and to inform the review of antenatal screening, including feedback of results to maternity and paediatric services to assist with local audits. The laboratory is required to work with stakeholders to obtain completed outcome data on all PNDs.

D Clinical and specialist support

1. Managed clinical network arrangements:
The aim of the SCT Programme is to reduce childhood morbidity and mortality for children with sickle cell and thalassaemia. While the remit of the SCT Programme does not extend to care, it is recognised that screening will not achieve its outcomes if high quality care is not offered to children with sickle cell and thalassaemia. Experience from the USA shows that the main reason for the failure of a screening programme in terms of clinical outcomes is failing to ensure that identified infants are registered in a programme of treatment and care, or that having been registered, they are subsequently lost to follow-up.

The National Haemoglobinopathies Project, hosted by East Midlands Specialised commissioning group (http://sct.screening.nhs.uk/cms.php?folder=2558) is producing guidelines on the effective commissioning of high quality sickle cell and thalassaemia services. It is anticipated that specialist haemoglobinopathy services will be commissioned by the National Commissioning Board, and this guidance will support the process. It is anticipated these guidelines will include the following recommendations:

- All infants must be linked to a recognised specialist centre or network, which is managed by a named paediatrician/haematologist (with adequate backup arrangements) for annual review
- All infants should have arrangements for care and day-to-day management at a relatively local centre which has links to a specialist centre
- Regional and local centres for care should use the multi-professional guidance (http://sct.screening.nhs.uk/getdata.php?id=11164) and audit care against these standards
- Network arrangements should include reporting to the SCT Programme of routine audit requirements
- Newborn laboratories should report screen positive results locally for action and to the designated centre for a clinical network. The designated clinical centre is responsible for checking the baby has entered the care pathway.

There is now a National Definitions Set (No.37) for specialised haemoglobinopathy services, which can be found at (http://sct.screening.nhs.uk/news.php?id=10816).

2. Specialist counselling arrangements:
There should be pre- and post-PND counselling arrangements by specialists with appropriate training for all units. Details of appropriate courses can be found at http://sct.screening.nhs.uk/training.

There should be formal arrangements to counsel women and the baby’s father with an affected infant by specialists with appropriate training.

3. Arrangements for prenatal diagnosis (PND) by chorionic villus sampling (CVS) or amniocentesis:
Women should have PND performed by, or be referred to, a recognised specialist centre which reports its rates of miscarriage as low.

PND samples (along with blood samples from both parents where available) should be sent to the recognised national prenatal diagnostic centres for processing and issue of the results. When women are offered PND, other chromosomal/genetic conditions that may be appropriate should be considered.

(Details of the referral process for samples are included in the DNA section of the Handbook for Laboratories [10] and the Map of Medicine pathway: http://sct.screening.nhs.uk/cms.php?folder=2493)

4. Specialist commissioning for preimplantation genetic diagnosis (PGD)
As a technique to help couples at risk of having a child with a serious genetic condition, PGD is very relevant to sickle cell and thalassaemia. Because of the cost associated with the use of this technique, and the fact that only highly specialized centre’s offer it, there may be a great deal of uncertainty in relation to when PGD would be appropriate.

An initiative was launched by the Genetics Consortium in the South East (covering London) to ensure equitable and appropriate use of PGD. A Preimplantation Genetic Diagnosis Clinical Group has been set up with a remit
Guidance on local organisational arrangements for the screening programmes

to consider individual cases for PGD, making sure that the genetic work-up has taken place and authorisation for funding has been requested. The group makes recommendations in relation to PGD based on expert clinical advice; however, the decision to fund treatments is agreed by each PCT.

Criteria have been developed for cases that would normally be recommended for treatment, but which do not have to be referred through the group. These are as follows:

- Female partner is under 40 years of age at time of referral
- Couple do not have a healthy child from the current relationship
- Couples who are carriers of translocation
- Cases with defined X linked disorders
- Cases for the following diseases: sickle cell, beta thalassaemia, cystic fibrosis, SMA, DMD, BMD, retinoblastoma and Alport syndrome
- When the laboratory in question has applied for CPA accreditation or received unconditional or conditional status
- Myotonic dystrophy (added 2007)
- X linked Pelizaeus Merzbacher (added 2008)

E Quality aspects of the programme

Quality Assurance aims to provide assurance that the Screening Programme is safe, effective and provides a good user experience – from identification of the eligible population through to entry into care. The SCT Programme works with UK NSC cross-programme groups on the principle that quality assurance arrangements should be made once across all programmes and not duplicated, resulting in increased efficiency and greater leverage than a QA system based on individual programmes. More information on cross-programme QA initiatives can be found here at http://www.screening.nhs.uk/qa-group.

These work streams need to link clearly with all cross-programme quality assurance initiatives and are currently under development.

1. Audit and monitoring:
   Each newborn programme will be required to produce an annual report on the process and outcome of the programme over the previous year, including information on coverage, timeliness and entry into care.

   Each maternity unit/associated provider trust will be required to produce an annual report on the process and outcome of the programme over the previous year.

   Each DNA laboratory will be required to produce an annual report and contribute data – including outcome data.

   All services will be required to provide relevant data to the SCT Programme for audit and monitoring, including nationally-agreed Key Performance Indicators (http://www.screening.nhs.uk/kpi), to ensure screening programmes are achieving the overall aims and objectives.

   The SCT Programme will be required to collate all relevant annual reports in order to:

   - review the outcome of the SCT Programme nationally
   - identify units that have been unable to comply with national reporting requirements
   - report on the progress of the SCT Programme in achieving national standards.
Information strategy

Whilst it is accepted that there is much in common in the information requirements for UK NSC embedded screening, the programmes have in practice developed in different ways and timescales. This has led to some inconsistencies of approach and incompatibility of definitions. An over-arching information strategy is needed to ensure cohesion of approach and language, commonality of system designs, general availability of basic components to build systems and the establishing of principles that underpin individual programme detail.

Information requirements should support all aspects of the screening programmes, so to better inform commissioning, to allow benchmarking, to enable surveillance and look back, to facilitate linkage of screening data to outcome data and to support initiatives on equity of access to services [92] http://sct.screening.nhs.uk/getdata.php?id=10962.

Three separate segments of information requirements for screening programmes can be identified:

- Managing people through the process of screening
- Failsafe systems / cohort identification
- Monitoring the overall outcome of screening

2. External Quality Assessment:

Cross-programme quality assurance processes across the Antenatal and Newborn Screening Programmes, including the SCT Programme, are under development. It is anticipated there will be a joined up process between the cross-programme quality assurance assessments and the programme specific issues, allowing no gaps or duplications between the processes. While the cross-programme quality assurance processes are being developed, we have outlined the SCT Programme-specific quality assurance processes that should be in place:

- All screening laboratories are required to take part in National EQA schemes and to share their results with the SCT Programme
- All screening laboratories to be CPA accredited and take part in the CPA screening assessment (currently under development)

3. Failsafe arrangements:

Each local Trust needs to have arrangements in place for a failsafe policy – with designated individuals from the relevant professional disciplines responsible for each aspect of the care pathway – to ensure all people who want screening are tested, receive their results and have them acted on in a timely manner. This should be distinct from the care pathway and responsibilities of professionals who follow up on particular actions for individual users of the service. It is acknowledged that in the absence of robust IT systems this is challenging. Suggested failsafe points are included on the Map of medicine screening pathways (http://sct.screening.nhs.uk/cms.php?folder=2493). Processes should ensure regular review by specified individuals and with clear accountability to ensure that the process of care operates smoothly and in a timely manner. For the Newborn Programme, this includes having a system to ensure that there is a review of positive screening (affected) infants.

The outcome of these processes should be included in annual reports.

It should be noted that the acceptable and achievable level of standards assume failsafe systems are in place. There should be specified people responsible for ensuring failsafe systems are working. If failsafe systems are not in place then the acceptable level for coverage of screening and use of the Family Origin Questionnaire at 100%.
4. Serious incident reporting and learning lessons:
The role of local, regional and national teams in investigating serious incidents in screening programmes is outlined in *Managing Serious Incidents in the English NHS National Screening Programmes, Guidance on behalf of the UK National Screening Committee (UK NSC), Version: 4.0, June 2010* (http://www.screening.nhs.uk/quality-assurance). The SCT Programme is working with the UK NSC to develop arrangements for serious incident reporting to the SCT Programme, which are not onerous, blaming, or designed to replace local investigation. This process aims to ensure problems highlighted about aspects of the screening programme that need central action or guidance can be rapidly identified, lessons learnt and changes made to avoid similar problems elsewhere in the country.

Incidents and serious incidents should be reported and investigated locally, and the SCT Programme should also be informed about incidents and serious incidents to ensure lessons are learnt in a timely manner and appropriate national action is taken if required.

5. Performance monitoring and management:
Performance monitoring of the programmes will be required at an appropriate local level and will need good links with commissioners. The Service Frameworks (http://www.screening.nhs.uk/quality-assurance#fileid9864) outline the performance that commissioners should expect from their providers [91]. There remain questions about cross-boundary issues and the need for nationally-approved IT systems in place.

An overall understanding of the way the Programme is operating will combine reviews of data at a **national level** for items where this is relevant (such as outcome of screen positive cases, training and education, performance of individual laboratories, and communication issues) with **local level information** and monitoring on issues of coverage, timeliness of process, care pathways and failsafe arrangements and their adequacy and sentinel events. The level at which monitoring will routinely occur is specified in the newborn, antenatal and linked standards.
Section 11
Laboratory requirements
Quality improvement covers the screening journey from offer of testing through to entry into care, as well as the user experience, equity, governance and commissioning. The minimum laboratory criteria requirements have been revised to include quality improvement across the whole screening pathway. These changes will be incorporated into the 3rd Edition of the Handbook for Laboratories, which is due to be published in 2012.

Antenatal screening laboratories are required to meet the following minimum criteria:

1. The laboratory must be appropriately accredited with a nationally-approved accreditation scheme such as Clinical Pathology Accreditation UK (Ltd), now formally part of the United Kingdom Accreditation Service (UKAS).
2. A senior member of the laboratory staff (at the level of medical consultant or clinical scientist consultant) must be responsible for the haemoglobinopathy screening service, with defined lines of accountability and authority for all laboratory aspects of the service.
3. The laboratory must adopt the testing algorithm, defined by the National Screening Programme, to determine those pregnancies at risk of sickle cell disease or thalassaemia. This testing algorithm sets out the conditions to be tested for and the analytical methods that can be used.
4. The laboratory must adopt the guidelines for the standardised reporting of antenatal screening results as defined by the National Screening Programme. These guidelines set out the wording to be used on laboratory reports in response to defined analytical data.
5. The laboratory must have a standard operating procedure for the antenatal sickle cell and thalassaemia screening service, describing the process of laboratory testing from initial receipt of the specimen to despatching of the report.
6. There must be a documented risk management policy for the laboratory aspects of the haemoglobinopathy screening service. This should describe the steps in the testing protocol where mistakes could occur, and the procedures that have been implemented to minimise the risk of the mistake occurring.
7. The laboratory must participate in an accredited External Quality Assessment Scheme (EQAS) appropriate for antenatal sickle cell and thalassaemia screening (eg. UKNEQAS), and must be able to demonstrate satisfactory performance as defined by the criteria specified by the EQA scheme organisers.
8. A report or interim report must be provided within 3 working days of receipt of a specimen by the laboratory.
9. The laboratory must participate in audit at local and regional level, with the effectiveness of the screening programme being published in a local annual report.
10. The laboratory must be willing to release information on screening performance, external quality assurance and CPA assessments to any appropriate monitoring group of the National Screening Committee and the NHS SCT Programme Centre, and be open to peer review visits and inspection by the commissioners or their representatives at any time, by mutual agreement.
11. The sensitivity of the testing protocol must be such that at least 95% of carriers/affected individuals with thalassaemia and the significant Hb variants are detected.
12. There must be a failsafe policy and arrangements in laboratory and maternity services for acting on and routinely reviewing positive screening results. Laboratories should work with maternity units to ensure that a joint review of screen positive results across the linked antenatal and newborn pathway occurs on a regular basis. This could be a virtual clinic.
13. The Handbook for Laboratories gives lines of responsibility. For both the newborn and the antenatal programmes, working together as a linked programme, this includes a requirement for a formal independent process, such as a regular audit meeting, for the review of all screen positive results and action taken to follow up screen positive cases. This should be undertaken on a regular basis (at least annually) by specified individuals and with clear accountability, to ensure that processes of care operate smoothly.
Quality improvement covers the screening journey from offer of testing through to entry into care, as well as the user experience, equity, governance and commissioning. The minimum laboratory criteria requirements have been revised to include quality improvement across the whole screening pathway. These changes will be incorporated into the 3rd Edition of the Handbook for Laboratories, which is due to be published in 2012.

Newborn screening laboratories are required to meet the following minimum criteria:

1. The laboratory must be appropriately accredited with a nationally-approved accreditation scheme such as Clinical Pathology Accreditation UK (Ltd), now formally part of the United Kingdom Accreditation Service (UKAS).

2. The workload of the newborn screening laboratory should exceed 25,000 specimens per year (ideally 50,000), to give appropriate economies of scale and confidence in the interpretation of abnormal results.

3. A senior member of the laboratory staff (at the level of medical consultant or clinical scientist consultant) must be responsible for the haemoglobinopathy screening service, with defined lines of accountability and authority for all laboratory aspects of the service.

4. The initial screening test must be performed using high-performance liquid chromatography (HPLC), isoelectric focusing (IEF), or a method giving comparable results – with a confirmatory test for positive results being performed on the original blood spot, using a different technique from the initial screening test.

5. Screening laboratories must use the appropriate screening status codes when interfacing with the child health record systems, and collect the recommended data fields for the annual return required for monitoring and audit purposes.

6. There must be a documented risk management policy for the laboratory aspects of the haemoglobinopathy screening service. This should describe the steps in the testing protocol where mistakes could occur, and the procedures that have been implemented to minimise the risk of the mistake occurring.

7. The laboratory must participate in an accredited External Quality Assessment Scheme (EQAS), appropriate for neonatal screening (eg. NEQAS), and must be able to able to demonstrate satisfactory performance as defined by the criteria specified by the EQA scheme organisers.

8. Appropriate internal quality control procedures must be undertaken and documented, eg. recording of reagent lot numbers, recording of turnaround time for reports, results of internal quality control specimens, etc.

9. The laboratory must participate in audit at local and regional level, with the results of the newborn screening programme being published in a local annual report. There must be links established with the antenatal screening laboratories to audit the effectiveness of both arms of the National Screening Programme.

10. The laboratory must be willing to release information on screening performance, external quality assurance and CPA assessments to any appropriate monitoring group of the National Screening Committee and the NHS SCT Programme Centre, and be open to peer review visits and inspection by the commissioners or their representatives at any time, by mutual agreement.

11. The sensitivity of the screening process (offer, test and repeat test) must exceed 99% detection for Hb-SS and Hb-SC and 95% detection for other conditions.

12. Failsafe policies in laboratories and clinical networks must be in place, to check if all children with a positive screening result have been received by clinical network centres and act where they have not.

13. As stated in the model service specifications for clinical care (National haemoglobinopathy commissioning project http://sct.screening.nhs.uk/cms.php?folder=2558 ) newborn laboratories should report screen positive results locally for action and to the designated centre for a clinical network. The designated clinical centre is responsible for checking the baby has entered the care pathway

14. The Handbook for Laboratories gives lines of responsibility. For both the newborn and the antenatal programmes, working together as a linked programme, this includes a requirement for a formal independent process, such as a regular audit meeting, for the review of all screen positive results and action taken to follow up screen positive cases. This should be undertaken on a regular basis (at least annually) by specified individuals and with clear accountability, to ensure that processes of care operate smoothly.
Quality improvement covers the screening journey from offer of testing through to entry into care, as well as the user experience, equity, governance and commissioning. The minimum laboratory criteria requirements have been revised to include quality improvement across the whole screening pathway. These changes will be incorporated into the 3rd Edition of the Handbook for Laboratories, which is due to be published in 2012.

DNA laboratories are required to meet the following minimum criteria:

1. The laboratory must be a member of the UK Genetic Testing Network (UK GTN) and comply with the quality criteria laid down by the UK GTN Steering Group.

2. The laboratory must be accredited by an appropriate body, eg. Clinical Pathology Accreditation UK (Ltd). and for those laboratories awaiting inspection, accreditation must be achieved within 18 months.

3. A senior member of the laboratory staff (at the level of medical consultant or clinical scientist consultant) must be responsible for the DNA analytical service, with defined lines of accountability for all laboratory aspects of the service.

4. The laboratory must agree to collect a minimum dataset of information for monitoring purposes, as well as appropriate patient information for a computerised national database associated with sickle cell diseases and thalassaemias. It must have the IT capacity to support standard reporting and audit requirements of Programme. The minimum dataset includes pregnancy outcome data.

5. The laboratory must have a standard operating procedure for the DNA work associated with the SCT Programme, describing the process of laboratory testing from initial receipt of the specimen to dispatching of the report.

6. There must be a documented risk management policy for the laboratory aspects of the DNA service. This should describe the steps in the testing protocol where mistakes could occur, and the procedures that have been implemented to minimise the risk occurring.

7. The laboratory must participate in an accredited External Quality Assessment Scheme (EQAS) appropriate for the DNA referrals for sickle cell and thalassaemia screening (eg. UKNEQAS), and must be able to demonstrate satisfactory performance.

8. A report or interim report must be provided within five working days of receipt of a DNA specimen by the laboratory for those clinical cases where the result is needed urgently for prenatal diagnosis, and within two weeks for less urgent cases. This will allow counselling for women within five days of diagnostic testing.

9. The laboratory must participate in audit at local, regional and national level, to assess the effectiveness of the National Screening Programme.

10. The laboratory must be willing to release information on laboratory performance to any appropriate monitoring group of the National Screening Committee, and be open to inspection by the commissioners, or their representatives, at any time.

11. The sensitivity of the testing process must exceed 99% sensitivity for Hb-SS, & SC and beta thalassaemia major at-risk or affected pregnancies.

12. There must be failsafe arrangements to be in place. This includes checking all results are received and acted upon (positive and negative) in a timely manner.
Section 12
Appendices

1. A linked Antenatal and Newborn Screening Programme
2. High prevalence antenatal screening algorithm
3. Low prevalence antenatal screening algorithm
4. Audit standards for newborn clinical care
5. Contacts and electronic links
6. Guidance concerning non-paternity issues
The NHS SCT Programme is the first service in the world to aim to link antenatal and newborn screening. Here we explain what we mean by a linked programme, why this provides benefits for patients and the NHS, and how this is breaking new ground for the NHS.

What do we mean by a linked programme?
At its simplest, it means that we are striving to link results from antenatal tests undergone by parents-to-be with their baby’s test result.

More broadly, our goal is to ensure that:

- for every test a person takes, the result is accessible throughout their life, whenever and wherever it is needed
- every step of the screening process is informed by results from the previous step
- the NHS sets up systems and procedures that enable different health professionals to access people’s test results when and where they are needed
- the process of following people through screening provides the NHS with an automatic check that no-one has slipped through the net, and that procedures have been followed properly.

Why have we pioneered the concept of a linked programme?
Linking results of parents and babies is particularly important for us because we are a genetic screening programme. In fact, we are the first national genetic screening programme in the NHS.

Our Antenatal Screening Programme is set up to see whether there is a risk that parents might pass on an inherited disease to their baby. Clearly where we are tracking inherited diseases through a family, it is really important to be able to link the test results of mother, father and baby.

More widely, linking results also helps us to talk to the family at large. With genetic conditions, if one family member is a carrier, it is likely that other people are carriers too. If the NHS is able to store and access all results, it means we can try to have important conversations with other family members such as siblings, aunts, uncles or cousins who might be at risk of passing on a condition to their own baby. For example, if a GP knows that one person is a carrier, they can ask them if other people in their family have been tested and explain why this might be important - particularly if they are planning to have a family.

In summary, genetic conditions can affect whole families over time and can involve a number of health professionals in different settings. A linked programme can help us to ensure that everyone’s results are accessible throughout their lifetime so that the appropriate information and care can be provided when and where it is needed.

What are the benefits to patients and to the NHS?

- Reducing anxiety and giving a chance to access information
Linking results from parents and babies is very important in reducing anxiety. If parents know from their own antenatal tests that there is a risk that their baby might inherit a condition or be a carrier, they can be prepared for that result. They have a chance to find out all the relevant information and to access support and advice. A linked programme should mean that their baby’s test result is never a shock out of the blue. Research shows that this prior knowledge makes a big difference for parents [18].

- Avoiding unnecessary repeat testing
The test for sickle cell and thalassaemia only needs to be done once in a person’s life. By having people’s test results available throughout their lives, it should mean that no-one should have an unnecessary repeat test - saving anxiety for patients and costs for the NHS.

- Ensuring accuracy in interpretation of results
Having access to parental test results can aid health professionals in making an accurate assessment of the baby’s result. This is because a baby’s test result may indicate a range of haemoglobin disorders with different health outcomes. For example, a preliminary result of FS could indicate either SS, S-Beta or S-HPFH - all of which...
have different prognoses. In these circumstances, knowing the parents’ results helps to identify which condition the baby has.

- Ensuring that nobody falls through the screening net
Linking results provides an automatic ‘failsafe’ system that helps us ensure we don’t miss people - for example if they move house or do not attend an appointment. It also helps us to establish systems for quality improvement - ie. measuring how well we are performing against our standards and objectives, and whether we are providing value for money for the NHS.

Why this is breaking new ground for the NHS
Offering genetic screening within mainstream NHS services is, in itself, breaking new ground. We have needed to invest in substantial education programmes for both the public and health professionals to understand issues such as inheritance, risk and the choices that may arise. In addition, genetic screening poses many challenges for the NHS in terms of changing roles for health professionals and the way that services are set up and delivered.

The nature of our linked programme is one such major challenge. This is for two key reasons. Firstly, it means establishing dialogue between many different professionals who may not routinely speak to each other, such as GPs, midwives, health visitors, laboratory staff and specialist doctors like paediatricians and haematologists. Secondly - and even more challenging - is the fact that the IT systems in these different settings often do not communicate with each other.

Mainly because of IT difficulties, a comprehensive programme is still an aspiration for us rather than a reality in every aspect. However, it is an extremely important aspiration and one that, when realised, will be immensely valuable for the NHS. Increasingly, patients expect doctors to know their past test results and to advise them on that basis. And - as genetic medicine advances - it is likely that other genetic tests will be able to benefit from the advances in linkage that we are developing.

And finally - a bigger vision of a linked programme
In the bigger vision, we move beyond links within the screening process to ensuring that screening is linked to care. This has always been a fundamental part of our philosophy. If screening identifies babies who have these serious conditions, then it is only ethical to ensure that they have high quality and accessible care – and that the transition from screening to care is managed smoothly and effectively.

The other vital part of this bigger picture is testing before pregnancy. Clearly it is stressful for parents to find out that they are carriers when they are already expecting a child. Since the test is a simple blood test that can be done at any time, it makes sense for young people to find out their carrier status before they start a family. Although there is no formal NHS screening programme outside pregnancy, we have been working to raise awareness that people can have this test at any time from their GP or from a local sickle cell and thalassaemia centre. These education programmes have been targeted at populations at higher risk of being carriers of these conditions.

In summary then, the widest vision of a linked programme embraces the whole journey - from informing at-risk populations about testing before pregnancy, to antenatal and newborn screening and, where relevant, to moving on into the care system. Our goal - albeit not one we can achieve immediately - is that results at each stage of this journey should be linked and available throughout a person’s life.
Appendix Two: Testing algorithm for laboratory screening in HIGH PREVALENCE areas (RF = Report Format)

* Refer analytical results to consultant for an opinion on the need for a clinical referral. Possible role of website/telephone advice.

** Consider at high risk if any ethnic origins in China (including Hong Kong), Taiwan, Thailand, Cambodia, Laos, Vietnam, Indonesia, Burma, Malaysia, Singapore, Philippines, Cyprus, Greece, Sardinia, Turkey, or if ethnic/ family origin is uncertain/ unknown. Reconsider low risk couples if fetal anaemia/ hydrops seen on ultrasound scanning or if family history of hydrops fetalis.
Appendix Three: Testing algorithm for laboratory screening in LOW PREVALENCE areas (RF = Report Format)

**Refer analytical results to consultant for an opinion on the need for a clinical referral. Possible role of website/telephone advice.**

**High risk if any ethnic family origins in China (including Hong Kong), Taiwan, Thailand, Cambodia, Laos, Vietnam, Burma, Malaysia, Singapore, Indonesia, Philippines, Cyprus, Greece, Sardinia, Turkey or if ethnic/family origin uncertain/unknown. Reconsider low risk couples if fetal anaemia/hydrops seen on ultrasound scanning or if family history of hydrops fetalis.**

***Low risk or high risk as determined by the family origin questionnaire. Note - If partner is in high risk ethnic group, test the mother’s sample regardless of her family origins.
These proposed standards are taken from the current clinical guidelines and are based on current evidence-based practice, accepted good practice and knowledge of current resources [67].

**Penicillin prophylaxis:**

i. 90% of infants should have been offered and prescribed Penicillin V (or alternative) by 3 months.

ii. 99% of infants should have been offered and prescribed Penicillin V (or alternative) by 6 months.

iii. Any parental refusal should be documented.

**Pneumococcal immunisation:**

i. 95% should be given Pneumovax (polysaccharide antigen) at 2 years of age (24-27 months) and five-yearly thereafter.

**TCD scanning:**

i. 90% of children with SCD (Hb SS and HbS/0 thalassaemia) should be offered annual TCD scans from the age of 2 to 16 years by 2012.

ii. 99% of SHT centres should have the capability of offering annual TCD scans to children with SCD by 2011.

**Follow-up and failsafe arrangements:**

The Specialist Health Team (SHT), in conjunction with local paediatric units, should have continuing responsibility for all children with SCD identified by the Newborn Screening Programme, and should maintain a list.

100% of babies identified are to be registered by eight weeks by a designated healthcare professional at the paediatric unit of the local acute hospital. This local acute hospital must have links into an SHT to ensure that any major complications, together with an annual review, can be managed by appropriate multidisciplinary teams at the specialist centre.

All SHTs and Local Health Teams (LHTs) should have robust follow-up arrangements to identify and follow up any child who does not attend their hospital appointments. They should also have the capability to track children who have moved out of the area in order to make appropriate handover arrangements.

**Data Collection:**

i. Anonymous data for 95% of children with SCD under the age of 5 should be submitted to the NHS SCT Programme – please note that this data does not require consent.

ii. 90% of children with SCD (whose parents have been offered information about the National Haemoglobinopathy Register (http://www.nhr.nhs.uk/) and have given consent for the child’s details to be included on the NHR) should have had their details entered on to the Registry.

**Failsafe arrangements**

The sickle cell centres, in conjunction with local paediatric units, should have continuing responsibility for children with sickle cell disease identified on the Newborn Screening Programme, and maintain a list. Sickle cell centres and local hospitals should have robust follow-up arrangements to identify and follow up any child who does not attend their hospital appointments, and should have the capability of tracking children who have moved out of area in order to make appropriate handover arrangements.


* There was previously an additional standard, stating that 95% of infants should have completed the primary Prevenar (conjugate pneumococcal vaccination) course by 15 months. This was withdrawn because PCV13 is now given universally to all babies.
Appendix Five: Contacts and electronic links

National & international websites & organisations with haemoglobinopathy, screening or genetic information

**Screening**
NHS Sickle Cell & Thalassaemia Screening Programme: http://sct.screening.nhs.uk
National Screening Committee: http://www.screening.nhs.uk
Newborn Screening: http://newbornbloodspot.screening.nhs.uk
Newborn Screening (Scotland): http://www.nsd.scot.nhs.uk/services/screening/newbornscreening/index.html
Newborn Screening (Northern Ireland): http://www.dhsspsni.gov.uk/index/phealth/php/screening/nbbscreening.htm

**Resources to support haemoglobinopathy screening and counselling**
Accessible Publishing of Genetic Information: http://www.chime.ucl.ac.uk/APoGI/
Brent Sickle Cell & Thalassaemia Centre: http://www.sickle-thal.nwlh.nhs.uk/
PEGASUS Network: http://www.pegasus.nhs.uk/
DIPEX: www.dipex.org.uk

**Genetics**
Genetic Alliance UK: http://www.gig.org.uk
Human Genetics Commission: http://www.hgc.gov.uk
British Society for Human Genetics: http://www.bshg.org.uk
National Newborn Screening and Genetics Resource Center (USA): http://genes-r-us.uthscsa.edu/

**Haemoglobins Data Bases**
http://globin.cse.psu.edu
http://globin.cse.psu.edu/html/huisman/variants

**Miscellaneous**
Association of Genetic Counsellors: http://www.agnc.co.uk
Sickle Cell Resources & Information: http://www.scinfo.org
European Network for Rare and Congenital Anaemias: http://www.enerca.org
Thalassaemia International Federation: http://www.thalassaemia.org.cy
UK Forum on haemoglobin disorders: http://www.haemoglobin.org.uk/

**National Voluntary Organisations**
**Sickle Cell Society**
54 Station Road, Harlesden, London NW10 4UA
Tel: 020 8961 7795; Fax: 020 8961 8346
Website: http://www.sicklecellsociety.org
Email: info@sicklecellsociety.org

**UK Thalassaemia Society**
19 The Broadway, Southgate, London N14 6PH
Tel: 020 8882 0011; Fax: 8882 8618
Website: http://www.ukts.org
Email: office@ukts.org
All genetic tests, including screening, undertaken during pregnancy and the newborn period will always include the possible issue of non-paternity. This needs to be considered when offering screening tests, and when inviting the woman’s ‘partner’ or ‘baby’s father’ for testing.

The role of health professionals is not to judge the situation but to provide clarity and discretion around genetic screening test results – particularly as there is no consensus on the rate of non-paternity in the population [93].

Obtaining this unsolicited information creates an ethical dilemma about whether to pass the information on, and to whom. Often the revelation of non-paternity may be detrimental to established relationships [94].

## Antenatal screening

When a carrier woman is identified during antenatal screening, the issue of non-paternity should be considered when offering screening to the ‘baby’s father’. It should be highlighted to the mother that the correct person to be screened is the ‘baby’s father’ so that the pattern of genetic inheritance in the baby can be correctly assessed. When discussing results the need for discretion is essential.

## Newborn screening for sickle cell diseases

Testing the newborn baby occasionally produces unexpected information (eg. evidence of non-paternity), and health professionals must be alert to the need for discretion in pursuing family studies and in discussing results with the mother/parents.

### Non-paternity may be suspected when

- a baby with a major haemoglobin disease has a carrier mother but the ‘father’ is not a carrier;
- a baby identified as a carrier has neither their mother nor their father identified as a carrier.

However, even when screening results seem to suggest non-paternity, alternative explanations must also be considered, for example:

- One parent carries a variant that cannot be detected by usual screening methods, eg. an unstable variant or a silent form of beta thalassaemia;
- There was an error
  a) with labelling a sample,
  b) in the laboratory,
  c) or in reporting the results;
- A parent’s identity may have been stolen by another person;
- The couple may have used assisted reproductive methods (artificial insemination by donor egg/sperm);
- The baby may have developed a De Novo mutation, which although rare is possible;
- A very premature baby may not yet have developed any of their adult haemoglobin.

### Guidance

Despite the above possibilities, the risk of non-paternity remains and this needs to be handled carefully if relationships and family units are not to be disrupted. It is not in the interests of anyone (professional or parent) to cause a division in the relationship by revealing this information [95].

The professional needs to remain non-judgemental while considering the following actions:

1. Review the antenatal and newborn screening process to establish whether or not an error may have occurred at any stage of the pathway;
2. Explore the possibility of non-paternity with the mother, preferably on her own in private, without her partner present;
3. Agree with the baby’s mother how the situation will be dealt with if non-paternity is indeed a possibility;
4. Offer a re-test (in the first instance) to:
   a. mother;
   b. baby
5. If indicated, re-screen the father;
6. Carefully document results and communicate these only to those professionals who need the information to support the family.
Section 13
References
References

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14. Balmer, S., Quality management for screening: report to the National Screening Committee: Nuffield Institute for Health.


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