Public Health England

NHS

NHS Sickle Cell and Thalassaemia Screening Programme Standards

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Third edition

Public Health England leads the NHS Screening Programmes

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About Public Health England

Public Health England (PHE) exists to protect and improve the nation's health and wellbeing, and reduce health inequalities. It does this through world-class science, knowledge and intelligence, advocacy, partnerships and the delivery of specialist public health services. PHE is an operationally autonomous executive agency of the Department of Health.

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About PHE Screening

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

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1. Introduction

These revised national standards for the NHS Sickle Cell and Thalassaemia (SCT) Screening Programme replace NHS Sickle Cell and Thalassaemia Screening Programme Standards October 2011 and have an implementation date of April 2017. A summary of the main changes is on page 10. They should be read in conjunction with the standards for the NHS Newborn Blood Spot Screening (NBS) Programme.

The SCT programme aims to support health professionals and commissioners in providing high quality SCT screening services. This involves the development and regular review of quality standards against which data is collected and reported. The standards provide a defined set of measures that providers have to meet to ensure local programmes are safe and effective.

Quality assurance (QA) is the process of checking that these standards are met and encouraging continuous improvement. QA covers the entire screening pathway; from identifying who is eligible to be invited for screening, through referral and intervention where required/appropriate.

2. The NHS Sickle Cell and Thalassaemia Screening Programme

The UK National Screening Committee (UK NSC) has responsibility for setting screening policy. The NHS SCT antenatal and newborn screening programme screens for:

- genetic carriers for sickle cell, thalassaemia and other haemoglobin disorders
- sickle cell disease
- thalassaemia
- haemoglobin disorders

It offers screening to:

- all pregnant women
- fathers-to-be, where antenatal screening shows the mother is a genetic carrier
- all newborn babies, as part of the Newborn Blood Spot Screening Programme

Objectives and outcomes of the SCT antenatal programme:

- to offer timely antenatal sickle cell and thalassaemia screening to all women (and couples), to facilitate informed decision-making
- for those women accepting prenatal diagnosis (PND), 50% of prenatal diagnoses to be performed before 12 weeks + 6 days

Objectives and outcomes of the SCT newborn programme:

- to identify babies born with conditions where early intervention is likely to be beneficial
- to achieve the lowest possible childhood death rate and to minimise childhood morbidity from sickle cell diseases

The SCT programme has responsibility for implementing this policy and setting standards in England. It is a complex programme delivered by a range of different organisations working together. The service specification (No. 18) for providers is available as part of the public health functions exercised by NHS England.

The SCT programme aims to ensure that there is equal access to uniform and quality assured screening across England and that families are provided with high quality information so they can make an informed choice about SCT screening. Review of performance at a local level by population group may indicate inequity in whether or not women and babies enter, complete the screening pathway or access services within optimal timescales. Tools that can be used to help local services and commissioners consider how to improve equity of access are the NHS England's Equality Diversity System and PHE's Health Equity Assessment Tool.

3. Format of the standards

The format of the screening standards ensures stakeholders have access to:

- reliable and timely information about the quality of the screening programme
- data at local, regional and national level
- quality measures across the screening pathway without gaps or duplications
- a consistent approach across screening programmes
- data collection that is proportionate to the benefits gained

4. Scope and terminology

Standards

This document presents standards that assess the screening pathway and allow for continuous improvement. This enables providers and commissioners to identify where improvements are needed.

To clarify what is measured, each standard has:

- an objective: the aim of the standard
- a criteria: what is being assessed
- a measure: 2 thresholds (acceptable and achievable)

The acceptable threshold is the lowest level of performance which programmes are expected to attain to ensure patient safety and programme effectiveness.

The achievable threshold represents the level at which the programme is likely to be running optimally.

All programmes should aspire towards attaining and maintaining performance at the achievable threshold. All programmes are expected to exceed the acceptable threshold and to agree to 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. These thresholds, definitions and reporting levels are approved by PHE's Screening Data Group.

The standards are accompanied by clinical guidelines that should be followed to deliver high quality screening processes and to meet the standards (see section 9).

Exclusions

The following standards and information are not included in this document:

1. Structural standards

These describe the structure of the programme and must be fully met. An example of a structural standard is ‘parents are provided with approved information on SCT screening’. Structural standards are included in screening service specifications and monitored through commissioning and other QA routes. Providers and commissioners should review the service specifications to ensure structural standards are met by all screening programmes.

2. Laboratories offering screening for the Sickle Cell and Thalassaemia Screening Programme must also be accredited by the UK Accreditation Service (UKAS) to ISO. ‘Medical laboratories – Requirements for quality and competence (ISO 15189) or be CPA accredited and actively transitioning towards ISO 15189.

3. Information on clinical outcomes

The SCT programme reports data on the pregnancy outcomes of screen positive women who accept prenatal diagnosis and newborn outcomes from screen positive babies. Outcome data is collected by National Congenital Anomolies and Rare Disorder Registration Service (NCARDRS).

5. Screening pathway

The standards are based on 10 generic themes that assess the whole pathway:

Themes	Related standards
1. Identify population (to accurately identify the population to whom screening is offered)	Standard 1: Antenatal coverage
2. Inform (to maximise informed choice across the screening pathway)	Standard 2: Timeliness of antenatal screening test Standard 5: Timely offer of PND to women at risk of having an affected infant Standard 6: Timeliness of PND
3. Coverage/uptake (to maximise uptake in the eligible population who are informed and wish to participate in the screening programme)	Standard 1: Antenatal coverage *Also Public Health Outcome Framework Indicator 2.20iii Sickle cell and thalassemia screening: coverage*
4. Test (to maximise accuracy of screening test from initial sample or examination to reporting the screening result)	Standard 3: Completion of family origin questionnaire (FOQ) Standard 4: Antenatal screening test turnaround times
5. Diagnose (to maximise accuracy of diagnostic test)	
6. Intervention/treatment (to facilitate high quality and timely intervention in those who wish to participate)	Standard 5: Timely offer of PND to women at risk of having an affected infant Standard 6: Timeliness of PND Standard 7: Timely reporting of PND results
7. Outcome (to optimise individual and population health outcomes in the eligible population)	Standard 8: Timely reporting of newborn screen positive results Standard 9 Timely receipt into Haemoglobinopathy Centres
8. Minimisation/harm (to minimise potential harms in those screened and in the population)	Standard 2: Timeliness of antenatal screening test Standard 5: Timely offer of PND to women at risk of having an affected infant Standard 8: Timely reporting of newborn screen positive results
9. Staff: education and training (to ensure that the screening pathway is provided by a trained and skilled workforce, with the capacity to deliver screening services as per service specification)	
10. Commissioning/governance (to ensure effective commissioning and governance of the screening programme)	

6. Relationships between standards and key performance indicators (KPIs)

KPIs are a subset of standards which focus on areas of particular concern. In general, once a KPI consistently reaches the achievable level, the KPI is withdrawn. This allows entry of another KPI to focus on additional areas of concern or a change to the threshold of the existing standard to promote continuous improvement.

SCT has 3 KPIs derived from standards 1 to 3 and NBS KPIs from standards 1a, 1b and 6

7. Reporting standards

SCT standards are reported annually and KPIs are reported quarterly (unless they are small numbers). The SCT programme coordinates an annual collection and analysis of standards data from antenatal, PND and newborn screening laboratories. The organisations collating the data are responsible for ensuring the data is accurate, timely and complete.

Specific details for reporting are provided for each standard in the template.

PHE is responsible for ensuring that reports on important aspects of screening are available at various geographies (for example local authority) to enable population-based oversight.

8. Other resources to support providers and commissioners

This document focuses on standards to enable providers and commissioners to continuously improve the quality of the screening programme. Additional operational guidance is available in the following documents:

Service specification (No. 18) NHS Sickle cell and thalassaemia screening
Handbook for sickle cell and thalassaemia screening
Laboratory handbooks
Guidelines for Newborn Blood Spot Sampling (2016)

9. Summary of changes

General changes:

Standard	Changes
Standard 1: Antenatal coverage	Former standard AO1aii changed to new format
Standard 2: Timeliness of antenatal screening test	Former standard AP1 changed from timeliness of 'offer' to timeliness of 'test'
Standard 3: Completion of family origin questionnaire (FOQ)	Former standard AO1aiii threshold increased
Standard 4: Antenatal screening test turnaround times	Former standard AO2; part 2 changed to new format
Standard 5: Timely offer of prenatal diagnosis (PND) to women at risk of having an affected infant	New standard
Standard 6: Timeliness of prenatal diagnosis (PND)	Former standard AO1b changed to new format
Standard 7: Timely reporting of prenatal diagnosis (NBS) results	Former standard AP3 changed to new format
Standard 8: Timely reporting of newborn screen positive results	Former standards NP3 changed to new format
Standard 9: Timely receipt into Haemoglobinopathy Centres	Former standards NP4; (part 2) changed to new format and NP4 (part 1) deleted

10. The SCT standards

Standard 1: Antenatal coverage

Rationale	To provide assurance that screening is offered to all eligible women and each woman accepting screening has a screening result. Timely information on screening coverage is important to identify trends and monitor the effectiveness of service improvements. Coverage is a measure of the delivery of screening to an eligible population. Low coverage might indicate that: • not all eligible women were offered screening • those offered screening are not accepting the test • those accepting the test are not being tested
Objective	To maximise the impact of the screening programme in the eligible population
Criteria	The proportion of pregnant women eligible for screening who are tested
Definitions	tested women / Eligible women — expressed as a percentage Numerator: 'tested women' is the total number of 'eligible women' for whom a screening result is reported, including: • known at risk couples referred directly for prenatal diagnosis (PND); repeat testing must not delay referral Denominator: 'eligible women' is the total number of pregnant women booked for antenatal care during the reporting period, or presenting in labour without previously having booked for antenatal care, excluding: • women who miscarry between booking and testing • women who opt for termination between booking and testing • women who transfer out between booking and testing, i.e. do not have a result • women who transfer in who have a result from a screening test performed elsewhere in this pregnancy
Performance thresholds	Acceptable level: ≥ 95.0% Achievable level: ≥ 99.0%
Mitigations/ qualifications	Requires matched cohort data
Reporting	Reporting focus: maternity service Data source: maternity service Responsible for submission: maternity service
Reporting period	Quarterly; data to be collated between 2 and 3 months after each quarter end Deadlines: 30 September (Q1), 31 December (Q2), 31 March (Q3), 30 June (Q4)

Standard 2: Timeliness of antenatal screening test

Rationale	To identify carrier and affected women by 10 weeks + 0 days of pregnancy to allow the baby's biological father to be offered testing and to offer of PND to women at risk of having an affected infant by 12 weeks + 0 days of pregnancy
Objective	To maximise the opportunity for informed choice
Criteria	Proportion of women tested by 10 weeks + 0 days gestation
Definitions	<table><tr><td>women tested by 10 weeks + 0 days gestation</td><td rowspan="2">expressed as a percentage</td></tr><tr><td>women for whom screening sample received at laboratory</td></tr></table> Numerator: 'women tested by 10 weeks + 0 days gestation' is the total number of pregnant women for whom a screening sample was received in the laboratory and for whom an antenatal sickle cell and thalassaemia screening result was available (though not necessarily communicated to the woman) by 10 weeks + 0 days gestation (≤70 days) Denominator: 'women for whom screening sample received at laboratory' is the total number of pregnant women for whom an antenatal sickle cell and thalassaemia screening sample was received at the laboratory during the reporting period excluding full blood count samples where the request is other than antenatal screening Calculation of gestational age, may be based on last menstrual period or ultrasound scan
Performance thresholds	Acceptable level: ≥ 50.0% Achievable level: ≥ 75.0%
Mitigations/ qualifications	Does not need to be matched cohort
Reporting	Reporting focus: maternity service Data source: antenatal screening laboratory Responsible for submission: maternity service
Reporting period	Quarterly; data to be collated between 2 and 3 months after each quarter end Deadlines: 30 September (Q1), 31 December (Q2), 31 March (Q3), 30 June (Q4)

Standard 3: Completion of family origin questionnaire (FOQ)

Rationale	To interpret screening results in high prevalence areas and to identify women at higher risk to be offered further testing in low prevalence areas [1]
Objective	To maximise accuracy of screening test
Criteria	Proportion of samples that arrive in the antenatal laboratory accompanied by a completed FOQ
Definitions	number of antenatal samples with completed FOQ / number of antenatal samples, expressed as a percentage Numerator: 'number of antenatal samples received in the laboratory with completed FOQ' Denominator: 'number of antenatal samples' received by the laboratory A completed FOQ must use the national template (paper or electronic format), and must include: • at least one box for the mother or options for 'declined to answer' or 'don't know' selected • at least one box for the father or options for 'declined to answer' or 'don't know' selected • gestational age or gestational age 'not known' recorded
Performance thresholds	Acceptable level: ≥ 95.0% Achievable level: ≥ 99.0%
Mitigations/ qualifications	Does not need to be matched cohort Laboratories that serve more than one maternity service must report by each maternity service
Reporting	Reporting focus: maternity service Data source: antenatal screening laboratory Responsible for submission: maternity service
Reporting period	Quarterly; data to be collated between 2 and 3 months after each quarter end Deadlines: 30 September (Q1), 31 December (Q2), 31 March (Q3), 30 June (Q4)

Standard 4: Antenatal screening test turnaround times

Rationale	To report screening outcomes promptly to help to achieve the offer of PND by 12 weeks + 0 days gestation
Objective	To maximise the opportunity for informed choice
Criteria	Proportion of results reported within 3 working days
Definitions	<table><tr><td>number of antenatal results reported ≤ 3 working days number of antenatal samples</td><td>expressed as a percentage</td></tr></table> Numerator: 'number of antenatal results reported ≤ 3 working days' on receipt of sample in the laboratory including: • interim reports if there is likely to be a delay in producing a final report e.g. recommending the baby's father testing • samples that cannot be processed due to poor sample quality or incomplete FOQ Denominator: 'number of antenatal samples' received in the laboratory • count receipt of sample (day 1) when the specimen is received in the reception in the first laboratory
Performance thresholds	Acceptable level: ≥ 90.0% Achievable level: ≥ 95.0%
Mitigations/ qualifications	Poor samples and incomplete FOQs are included because a report must be issued to request a new sample/more information
Reporting	Reporting focus: antenatal screening laboratory Data source: antenatal screening laboratory Responsible for submission: antenatal screening laboratory
Reporting period	Annually for samples received in the laboratory in the previous financial year Deadline: 30 June

Standard 5: Timely offer of prenatal diagnosis (PND) to women at risk of having an affected infant

<table>
<tr><td>Rationale</td><td>There is a known association between gestation at screening offer and uptake of PND, with the early offer of screening being associated with greater uptake of PND [2], [3], and [4]. The majority of PND currently takes place after 12 weeks + 6 days [5]. Approximately half of women at risk of having an affected infant decline PND; gestational age at time of decline is not known</td></tr>
<tr><td>Objective</td><td>To maximise the opportunity for women at risk of having an affected infant to make informed and timely reproductive choices</td></tr>
<tr><td>Criteria</td><td>Proportion of at risk women offered PND by 12 weeks +0 days gestation</td></tr>
<tr><td>Definitions</td><td>
<table>
<tr><td>Number of at risk women offered PND by 12 weeks + 0 days
Number of at risk women</td><td>Expressed as a percentage</td></tr>
</table>
Numerator: ‘Number of at risk women offered PND by 12 weeks + 0 days gestation’

Denominator: ‘Number of at risk women’

At risk women includes:
• those with a one in four chance or higher of the fetus being affected by a serious haemoglobin disorder (mother and biological father results known)
• women who are carriers or affected with a clinically significant haemoglobin variant where the haemoglobinopathy status of the baby’s biological father is unknown
• pregnancies by donor egg or sperm where the haemoglobinopathy status of the donor is unknown and the biological partner is a carrier or affected with a clinically significant haemoglobin variant
</td></tr>
<tr><td>Performance thresholds</td><td>Acceptable level: ≥ 50%
Achievable level: ≥ 75%</td></tr>
<tr><td>Mitigations/ qualifications</td><td>None</td></tr>
<tr><td>Reporting</td><td>Reporting focus: maternity service
Data source: maternity service and specialist haemoglobinopathy counsellors
Responsible for submission: maternity service</td></tr>
<tr><td>Reporting period</td><td>Annually for women offered in the previous financial year
Deadline: 30 June
A new KPI with quarterly data collection will be piloted in 2017</td></tr>
</table>

Standard 6: Timeliness of prenatal diagnosis (PND)

Rationale	There is a known association between gestation at PND offer and uptake, with the early offer being associated with greater uptake of PND. Advanced gestational age may limit reproductive choices [2], [3], [4].
Objective	Timely intervention and choice in procedure for those who accept PND
Criteria	Proportion of PND tests performed by 12 weeks + 6 days gestation
Definitions	number of women who have PND by 12 weeks + 6 days gestation / number of women who have PND — expressed as a percentage Numerator: 'number of women who have PND by 12 weeks + 6 days gestation' Denominator: 'number of women who have PND'
Performance thresholds	Acceptable level: ≥ 50.0% Achievable level: ≥ 75.0%
Mitigations/ qualifications	None
Reporting	Reporting focus: maternity service Data source: PND laboratory Responsible for submission: PND laboratory
Reporting period	Annually for women tested in the previous financial year Deadline: 30 October

Standard 7: Timely reporting of prenatal diagnosis (PND) results to parents

Rationale	To provide information about living with and supporting an affected child and if chosen, to ensure timely referral for termination of pregnancy
Objective	Maximise informed choice
Criteria	Proportion of results received within 5 working days of PND procedure
Definitions	number of women who receive their result ≤ 5 working days of PND test / number of women who have PND — expressed as a percentage Numerator: 'number of women who receive their result ≤ 5 working days of PND test' Denominator: 'number of women who have PND'
Performance thresholds	Acceptable level: ≥ 70.0% Achievable level: ≥ 90.0%
Mitigations/ qualifications	None
Reporting	Reporting focus: maternity service Data source: maternity service and counselling services Responsible for submission: maternity service
Reporting period	Annually for women tested in the previous financial year Deadline: 30 June

Standard 8: Timely reporting of newborn screen positive results

Rationale	To provide timely results. This includes providing information about the screening result, living with and supporting an affected child, and the care pathway
Objective	To ensure parents of screen positive infants receive results at ≤ 28 days of age
Criteria	Proportion of parents informed of newborn screen positive results at ≤ 28 days of age
Definitions	<table><tr><td>number of newborn infants with screen positive results for whom parents are given results by ≤ 28 days of age</td><td rowspan="2">Expressed as a percentage</td></tr><tr><td>number of newborn infants with screen positive results</td></tr></table> Numerator: 'number of newborn infants with screen positive results reported to parents at ≤ 28 days of age' Denominator: 'number of newborn infants born within the reporting period, with screen positive result' Specified conditions to be detected in newborn screening: HbSS, HbSC, HbS/β thalassaemia (S/β+, S/β°, HbS/δβ, HbS/γδβ, S/Lepore), HbS/DPunjab, HbS/E, HbS/OArab, HbS/HPFH, Hb S with any other variant and no Hb A, and other clinically significant Haemoglobinopathies likely to be detected as by-products of newborn screening including β thalassaemia major, Hb E/β thalassaemia, and β thalassaemia intermedia
Performance thresholds	Acceptable level: ≥ 90.0 % Achievable level: ≥ 95.0%
Mitigations/ qualifications	Detection of thalassaemia is not part of the programme but we expect beta thalassaemia major to be detected as a by-product and the same standards for communicating results to parents and enrolment into care apply
Reporting	Reporting focus: • SHC geographical area of responsibility • haemoglobinopathy centre (nursing or medical) responsible for giving results • newborn screening laboratory Data source: organisation responsible for giving results Responsible for submission: newborn screening outcomes system
Reporting period	Annually for infants born in the previous financial year Deadline: June 30

Standard 9: Timely receipt into haemoglobinopathy centres

Rationale	To ensure timely and appropriate management, newborn infants with positive screening results must attend a haemoglobinopathy centre (medical) by 90 days of age.
Objective	To optimise individual and population health outcomes in newborn infants born with conditions where early intervention is likely to be beneficial
Criteria	Proportion of newborn infants with a positive screening result followed up and entered into care within 90 days of age
Definitions	<table><tr><td>number of newborn infants with screen positive result seen by ≤ 90 days of age number of newborn infants with screen positive result</td><td>Expressed as a percentage</td></tr></table> Numerator: 'number of newborn infants with screen positive result seen at a haemoglobinopathy centre (medical) ≤ 90 days of age' Denominator: 'number of newborn infants born within the reporting period, with screen positive result' Screen positive results: specified conditions to be detected in newborn screening: HbSS, HbSC, HbS/β thalassaemia (S/β+, S/β°, HbS/δβ, HbS/γδβ, S/Lepore), HbS/DPunjab, HbS/E, HbS/OArab, HbS/HPFH, Hb S with any other variant and no Hb A, and other clinically significant Haemoglobinopathies likely to be detected as by-products of newborn screening including β thalassaemia major, Hb E/β thalassaemia and β thalassaemia intermedia. Effective timeframes penicillin prophylaxis should start by 90 days of age in children with sickle cell disease [6], infants with significant thalassaemia do not require penicillin prophylaxis but are still expected to be seen by 90 days of age
Performance thresholds	Acceptable level: ≥ 90.0% Achievable level: ≥95.0%
Mitigations/ qualifications	None
Reporting	Reporting focus: • specialist haemoglobinopathy centre with responsibility for geographical area (in development) • haemoglobinopathy centre (medical) responsible for care • newborn screening laboratory Data source: haemoglobinopathy centre (medical) responsible for care Responsible for submission: newborn screening outcomes system
Reporting period	Annually for infants born in the previous financial year

	Deadline: 31 July

11. Abbreviations

CCG	clinical commissioning group
CHIS	child health information system
FOQ	family origin questionnaire
KPI	key performance indicator
NBS	newborn blood spot
PHE	Public Health England
PND	prenatal diagnosis
QA	quality assurance
SCD	sickle cell disease
SCT	sickle cell and thalassaemia
SHC	specialist haemoglobinopathy centre
UK NSC	UK National Screening Committee

12. Glossary

A glossary can be found within the document *PHE screening key performance indicators for 2016 to 2017*

The glossary defines terms that are consistent across NHS screening programmes. The scope of each defined term as it applies to a particular screening programme is detailed separately for each screening programme.

13. References

[1] Dyson, S.M., et al., Ethnicity questions and antenatal screening for sickle cell/thalassaemia [EQUANS] in England: a randomised controlled trial of two questionnaires. Ethnicity & Health, 2006. 11(2): p.169-189

[2] Modell, B., et al., Informed Choice in Genetic Screening for Thalassaemia during Pregnancy: Audit from a National Confidential Inquiry. BMJ: British Medical Journal, 2000. 320 (7231): p. 337-341

[3] Neuenschwander, H. and B. Modell, Audit of process of antenatal screening for sickle cell disorders at a north London hospital. BMJ, 1997. 315(7111): p. 784

[4] Greengross, P., et al., Outcomes of universal antenatal screening for haemoglobinopathies. Journal of Medical Screening, 1999. 6(1): p. 3

[5] NHS Sickle Cell and Thalassaemia Screening Programme
Data Report 2015 to 2016: Trends and performance analysis

[6] Gaston, M.H., et al., Prophylaxis with Oral Penicillin in Children with Sickle-Cell-Anemia - a Randomized Trial. New England Journal of Medicine, 1986. 314(25): p.1593-1599