Public Health England

NHS

NHS public health functions agreement 2019-20

Service specification No.18
NHS Sickle Cell and Thalassaemia Screening Programme

NHS England and NHS Improvement

NHS public health functions agreement 2019-20

Service specification No.18 NHS Sickle Cell and Thalassaemia Screening Programme

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Promoting equality and addressing health inequalities are at the heart of NHS England and NHS Improvement values. Throughout the development of the policies and processes cited in this document, we have:

- Given due regard to the need to eliminate discrimination, harassment and victimisation, to advance equality of opportunity, and to foster good relations between people who share a relevant protected characteristic and those who do not share it (as required under the Equality Act 2010); and
- Given due regard to the need to reduce inequalities between patients in access to, and outcomes from, healthcare services and to ensure services are provided in an integrated way where this might reduce health inequalities (in accordance with the duties under sections 13G and 13N of the NHS Act 2006, as amended).

Contents

Service specification No.18

This is a service specification to accompany the 'NHS public health functions agreement 2019-20 (the 2019-20 agreement').

This service specification is to be applied by NHS England and NHS Improvement in accordance with the 2019-20 agreement. This service specification is not intended to replicate, duplicate or supersede any other legislative provisions that may apply.

Where a specification refers to any other published document or standard, it refers to the document or standard as it existed at the date when the 2019-20 agreement was made between the Secretary of State and NHS England and NHS Improvement Board, unless otherwise specified. Any changes in other published documents or standards may have effect for the purposes of the 2019-20 agreement in accordance with the procedures described in Chapter 3 of the 2019-20 agreement

Service specifications should be downloaded in order to ensure that commissioners and providers refer to the latest document that is in effect.

The 2019-20 agreement is available at www.gov.uk (search for 'commissioning public health').

All current service specifications are available at www.england.nhs.uk (search for 'commissioning public health').

Section 1: Purpose of Screening Programme

1.1 Purpose of the Specification

To ensure a consistent and equitable approach across England a common national service specification must be used to govern the provision and monitoring of the linked antenatal and newborn NHS Sickle Cell and Thalassaemia (SCT) Screening Programme.

The purpose of the service specification is to outline the service and quality indicators expected by NHS England and NHS Improvement (NHS E) for NHS E responsible population and which meets the policies, recommendations and standards of the NHS Screening Programmes.

The service specification is not designed to replicate, duplicate or supersede any relevant legislative provisions which may apply, e.g. the Health and Social Care Act 2008 or the work undertaken by the Care Quality Commission. The specification will be reviewed and amended in line with any new guidance as quickly as possible.

This specification needs to be read in conjunction with the following:

SCT Screening Programme Standards, 2018 http://sct.screening.nhs.uk/standards https://www.gov.uk/government/collections/nhs-population-screening-programme- standards

NHS Sickle Cell and Thalassaemia Programme Handbook 2017 https://www.gov.uk/government/publications/handbook-for-sickle-cell-and- thalassaemia-screening

Checks and audits to improve quality and reduce risks 2017 https://www.gov.uk/government/publications/sct-checks-and-audits-to-improve-quality- and-reduce-risks

Sickle Cell and Thalassaemia Handbooks for Laboratories 2017 https://www.gov.uk/government/publications/sickle-cell-and-thalassaemia-screening- handbook-for-laboratories

NHS Newborn Blood Spot Screening Programme service specification. https://www.england.nhs.uk/commissioning/wp-content/uploads/sites/12/2016/04/serv- spec-19-chld-blood-spot-screening.pdf

Specialised Haemoglobinopathy Services definition as set out in the Manual for prescribed specialised services

https://www.england.nhs.uk/wp-content/uploads/2017/10/prescribed-specialised-services-manual.pdf

The Specification for Specialised Services for Haemoglobinopathy Care (in development); it will be published at: https://www.england.nhs.uk/commissioning/spec-services/npc-crg/blood-and-infection-group-f/f05/

NICE clinical guideline 62. Antenatal care https://www.nice.org.uk/guidance/cg62

NICE Clinical Guideline 110. Pregnancy and Social Complex Factors: A Model for Service Provision for Pregnant Women with Complex Social Factors. https://www.nice.org.uk/guidance/cg110

Guidance and updates on KPIs https://www.gov.uk/government/collections/nhs- screening-programmes-national-data-reporting

NHS Screening Programmes guidance, Managing Serious Incidents in the English NHS

National Screening Programmes https://www.gov.uk/government/publications/managing-safety-incidents-in-nhs- screening-programmes

Recommended reading ‘Parents’ stories’ personal experiences of the SCT programme, 2017 http://www.sicklecellsociety.org/parents-stories/

1.2 Aims

The NHS SCT Programme aims to:

- ensure a high quality, accessible screening programme throughout England
- support people to make informed choices during pregnancy and ensure timely transition into appropriate follow up and treatment
- improve infant health through prompt identification of affected babies and timely transition into clinical care
- promote greater understanding and awareness of the conditions and the value of screening

1.2.1 Antenatal Sickle Cell and Thalassaemia Screening Programme

Objectives:

To offer timely antenatal sickle cell and thalassaemia screening to all women (and couples), to facilitate informed decision-making.

Outcome:

For those women accepting prenatal diagnosis (PND), at least 50% of PNDs to be performed before 12 weeks 6 days.

1.2.2 Newborn Sickle Cell Screening Programme

Objectives:

To identify babies born with conditions where early intervention is likely to be beneficial.

Outcome:

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

1.2.3 Linkage between Antenatal and Newborn Sickle Cell and Thalassaemia Screening Programme

Objectives:

- to promote an appropriate level of understanding about screening for these genetically inherited conditions among professionals involved with the programme
- to review the results from antenatal testing before, during and after the newborn test is offered, and to check that the results are congruent; and
- to prepare parents for their baby’s screening result

Outcome:

To minimise the adverse effects of screening.

1.3 Principles

- all individuals will be treated with courtesy, respect and an understanding of their needs
- all those participating in the sickle cell and thalassaemia screening programme will have adequate information on the benefits and risks to allow an informed decision to be made before participating
- the target population will have equitable access to screening
- screening will be effectively integrated across a pathway including between the different providers, screening centres, primary care and secondary care

1.4 Equality

Delivery of the screening programme contributes to reducing health inequalities and should include the following deliverables:

- screening should be delivered in a way which addresses local health inequalities, tailoring and targeting interventions when necessary
- a Health Equity Audit should be undertaken as part of both the commissioning and review of this screening programme; including equality characteristics, socio-economic factors and local vulnerable populations
- the service should be delivered in a culturally sensitive way to meet the needs of local diverse populations
- user involvement should include representation from service users with equality characteristics reflecting the local community including those with protected characteristics
- providers should exercise high levels of diligence when considering excluding people with protected characteristics in their population from the programme and follow equality, health inequality and screening guidance when making such decisions

The provider will demonstrate they have systems in place to address health inequalities and make sure there is equity of access to screening, subsequent diagnostic testing and outcomes. This will include, for example, how the services are designed to make sure that there are no obstacles to access on the grounds of the nine protected characteristics as defined in the Equality Act 2010.

The provider will have procedures in place to identify and support those persons who are considered vulnerable/ hard-to-reach, including but not exclusive to, those who are not registered with a GP; homeless people and rough sleepers, asylum seekers, gypsy traveler groups and sex workers; those in prison; those with mental health problems; those with drug or alcohol harm issues; those with learning disabilities, physical disabilities or communications difficulties. The provider will comply with safeguarding policies and good practice recommendations for such persons.

Providers are expected to meet the public sector Equality Duty which means that public bodies have to consider all individuals when carrying out their day-to-day work – in shaping policy, in delivering services and in relation to their own employees https://www.

gov.uk/equality-act-2010-guidance.

It also requires that public bodies:

- have due regard to the need to eliminate discrimination
- advance equality of opportunity
- foster good relations between different people when carrying out their activities

Personal informed choice

All screening is an individual choice. The UK NSC has published guidance for screening programmes in the 4 UK countries to follow. Everyone must be given the opportunity to make an informed choice about whether or not to be screened. The decision should be based on an understanding of:

- why they are being offered screening
- what happens during the test
- the benefits and risks of screening
- the potential outcomes (including types of result, further tests and treatment)
- what happens to their screening records

If someone is provided with the above information about the programme and chooses not to attend screening, then this is a valid choice and must be respected.

Women who are at risk of having a baby affected by SCT who accept PND and parents of babies who screen positive must be provided with information about data sharing with NCARDRS. Information must include how to opt out of the register for more information sees www.gov.uk/phe/ncardrs

Sharing personal information

Under the 2010 Equality Act, screening services are required to anticipate and prevent discrimination against people with learning disabilities.

The duty of care to share information can be as important as the duty to protect patient confidentiality. GPs and other health professionals should have the confidence to share relevant information with screening services in the best interests of their patients. For example, a GP may know that an individual with a learning disability requires accessible information about screening in easy read format or needs a longer than normal appointment slot.

See NHS England's information sharing policy for more detailed guidance.

PHE Screening's privacy notice has more information about how screening data is shared within the legal requirements, including those of the General Data Protection Regulation (GDPR).

Reasonable adjustments

Under the 2010 Equality Act, screening providers have a legal duty to make reasonable adjustments to make sure services are accessible to disabled people as well as everybody else.

Screening providers must follow the Accessible Information Standard by law. The standard

aims to make sure that people who have a disability, impairment or sensory loss are provided with information they can easily read or understand with support, so they can communicate effectively with health and social care services.

As part of the Accessible Information Standard, screening providers must do 5 things.

1. Ask people if they have any information or communication needs, and find out how to meet their needs.
2. Record those needs clearly and in a set way.
3. Highlight or flag the person's file or notes so it is clear that they have information or communication needs and how to meet those needs.
4. Share information about people's information and communication needs with other providers of NHS and adult social care, when they have consent or permission to do so.
5. Take steps to ensure that people receive information which they can access and understand, and receive communication support if they need it.

National accessible information materials

PHE Screening has published national easy read versions of screening information leaflets and screening appointment letter templates.

Local screening providers should use these national materials when inviting individuals for screening who have been identified as needing information in an easy read format.

Large print and audio (MP3) versions of standard information leaflets are also available to download from GOV.UK for people with sight loss. Local screening providers should send any individual requests for hard copy Braille versions of PHE Screening leaflets to the screening helpdesk.

Section 2: Scope of Screening Programme

2.1 Description of Screening Programme

The provider is expected to follow, guidance from the national screening programme as detailed in the standards and policies

To make sure there is national consistency the provider should:

- work to national screening standards
- provide data and reports against screening standards, key performance indicators (KPIs), and other measures as requested by the national screening programme
- provide data on screening outcomes as required by the national screening programme
- make sure appropriate governance structures are in place
- take part in quality assurance (QA) processes and implement changes recommended by QA including urgent suspension of services if required
- implement and monitor failsafe procedures and drive continuous quality improvement
- work with NHS England and NHS Improvement and the screening quality assurance service (SQAS) in reporting, investigating and managing screening safety incidents
- respond to national action/lessons for example, change of software, equipment or equipment supplier, new technologies
- make sure all health care professionals access appropriate training to maintain continuous professional development and competency
- use materials provided by the national screening programme, for example, leaflets, e-learning resources and operational guidance
- implement and support national IT developments

The linked screening programme comprises two parts: sickle cell and thalassaemia screening during pregnancy and sickle cell screening in the newborn period.

Newborn screening for sickle cell disease is offered to all babies in England and is integrated into the newborn blood spot test. See service specification No. 19: NHS newborn blood spot screening programme. https://www.england.nhs.uk/commissioning/wp- content/uploads/sites/12/2016/04/serv-spec-19-chld-blood-spot-screening.pdf

Antenatal screening for sickle cell, other haemoglobin variants and thalassaemia is offered to pregnant women early in pregnancy, (women should be tested by 10 weeks + 0 days of pregnancy) to identify women and couples who are at increased risk (1:4 or higher) of an affected pregnancy to offer them the choice of PND and the option of termination of an affected pregnancy or continuation of the pregnancy. For those women and couples at risk of having an affected baby PND to be offered by 12 weeks 0 days. PND test should ideally take place before 12 weeks 6 days of pregnancy.

The provider must ensure:

- that if risk status is known pre-pregnancy a prompt referral for counselling and offer of PND is made at the first appointment in pregnancy
- all pregnant women, are offered screening for sickle cell, other haemoglobin variants and thalassaemia
- where a woman is identified as affected or a carrier of significant haemoglobinopathy the baby's biological father is offered screening for sickle cell, other haemoglobin variants and thalassaemia, irrespective of their family origin
- women and couples at risk of having a baby with sickle cell disease or thalassaemia major are offered counselling and diagnostic tests for the baby.

2.2 Care pathway (figure 1)

Identify eligible population: All pregnant women are eligible for screening. Women should be tested at the first antenatal appointment by 10 weeks and 0 days of pregnancy. In each pregnancy, the family origin questionnaire should be completed and a routine full blood count taken and mean cell haemoglobin (MCH) and other red cell indices should be assessed. In rare circumstances molecular techniques may be used to confirm their haemoglobinopathy status.

Provide information and take consent: personal informed choice and reference to the provision of written information including *Screening Tests For You And Your Baby* should be recorded in the maternity records.

Known at risk couples: women and couples who already know their carrier status must be offered prompt referral for counselling and offer of PND (Figure 2).

Low prevalence trust: Low prevalence trusts are those where less than 1% of the booking bloods received by the laboratory are screen positive. In low prevalence trusts a family origin questionnaire (FOQ) is used to assess the risk of either the woman or the baby's father being a carrier for sickle cell or other haemoglobin variants, or alpha thalassaemia major, to identify those who need testing for haemoglobin variants. The FOQ should be completed and sent to the laboratory with the sample.

For more information on the FOQ see https://www.gov.uk/government/publications/family-origin-questionnaire-sickle-cell-and- thalassaemia-screening

High prevalence trust: High prevalence trusts are those where greater than or equal to 2% of the booking bloods received by the laboratory are screen positive.

In high prevalence trusts all pregnant women are offered screening followed by a blood test for sickle cell and other haemoglobin variants. The FOQ must also be completed and sent to the laboratory with the sample to facilitate interpretation of results by laboratory staff and identify those at risk of alpha thalassaemia major.

Laboratories that are between these two cut-offs would be considered borderline and should continue to use their current algorithm.

For a list of high and low prevalence trusts and information on obtaining FOQ forms see https://www.gov.uk/government/publications/nhs-trusts-area-prevalence-for-sickle-cell-and-thalassaemia

Trust mergers

The high prevalence algorithm is viewed as the gold standard, low prevalence trusts are recommended to move to the high prevalence algorithm rather than the other way around.

There should be one prevalence algorithm per laboratory to avoid laboratories needing to follow two algorithms side-by-side. Refer to https://www.gov.uk/government/publications/nhs-trusts-area-prevalence-for-sickle-cell-and-thalassaemia.

Take sample and send to laboratory with completed family origin questionnaire (FOQ):

Information on the family origins of the woman and the baby's biological father must accompany the full blood count (FBC) and haemoglobinopathy screen sample. Family origins of both the woman AND the baby's biological father (where available) going back at least 2 generations (or more if possible) need to be assessed and documented.

Screening for sickle cell and thalassaemia should still be offered even if information about the baby's biological father is unavailable.

If a woman declines screening, explore the reasons for this and document.

In low prevalence trusts, if a sample is received in the laboratory without a completed FOQ there is a risk that the woman will not be screened for sickle cell disease and a risk of missing an affected pregnancy.

There should be a local policy in place to ensure that FOQ information on any women whose samples are received without an FOQ, are obtained.

Screening laboratories must be able to identify antenatal samples as distinct from other samples they receive and should be able to match these samples to a specific maternity service.

Management of results

Laboratory tests sample as per national policy and reports results as per local arrangements: All laboratories and maternity units should have a written policy and process for reporting results to all women, and for obtaining a blood sample from the baby's biological father for all carrier and/or affected women for screening.

Nothing abnormal detected: All women should be notified of their screening test result before or at the next antenatal visit, according to local protocol. The result should be recorded in the health records and on the newborn blood spot card.

Inconclusive results: Some haemoglobin variants are difficult to identify and analysis may take longer. Rather than waiting for a long period of time for a conclusive maternal result, offer screening to the baby's biological father and if nothing abnormal is detected on father's result, then the risk to the baby can be excluded.

Result for carrier or affected mothers: A healthcare professional informs parents of carrier/affected results in accordance with programme guidelines and local pathway. Women should be provided with information about their carrier status along with a leaflet specific to their carrier status: https://www.gov.uk/government/collections/adult-carriers-sickle-cell- thalassaemia-unusual-haemoglobin

Refer affected mothers to consultant care for clinical and obstetric management.

The programme recommends the maternity units keep a log of, and inform the newborn laboratory of, all women who are carriers or affected, including results from PND, and have

systems in place to check the screening result of babies born to screen positive women.

Offer screening to baby's biological father: the offer must be made in every pregnancy, irrespective of previous screening results.

Baby's biological fathers should be provided with written information: https://www.gov.uk/government/publications/tests-for-dads-sickle-cell-and-thalassaemia- screening

Baby's biological father available and gives consent.

Laboratory tests sample and reports results as per local arrangements.

Nothing abnormal detected on screening of baby's biological father: Offer information and advice about the possibility of the child being a carrier based on the mother's carrier or affected result.

Confirmed carrier result in both parents - refer at risk couple: Parents are referred for counselling by a trained healthcare professional. Written information should be provided along with written confirmation of carrier status.

https://www.gov.uk/government/publications/baby-at-risk-of-having-sickle-cell-disease-description-in-brief

https://www.gov.uk/government/publications/baby-at-risk-of-having-thalassaemia-description-in-brief

Baby's biological father not available or declines consent: If the baby's biological father is unavailable for testing or his haemoglobinopathy status is unknown, then a risk assessment should be done. The programme supports women who are carriers or affected being offered PND in this situation. PND for sickle cell disease can be undertaken without the baby's biological father's DNA.

Similarly, PND for thalassaemia can be undertaken without the baby's father's DNA, although the diagnosis cannot be given with as high a degree of certainty if samples from both parents are not known or tested.

Offer PND testing: Prior to the PND test always contact the PND laboratory to make arrangements for sending the sample. Fresh blood samples from both parents (where available) are to be sent to the DNA referral laboratory with appropriate consent for molecular analysis along with the PND sample.

Decline PND: Respect decision. The programme recommends that information on women who decline PND is recorded in the maternity notes.

Accept PND - contact PND laboratory to make arrangements for analysis: For additional information on DNA testing and contact details of PND laboratories see laboratory handbook at https://www.gov.uk/government/publications/sickle-cell-and-thalassaemia-screening- handbook-for-laboratories

Inform parents, community midwife and GP: The responsibility for reporting results is as per local arrangements / practice. To find out what your local arrangements for reporting results are, contact your local antenatal and newborn screening coordinator.

Carrier or normal result: A designated healthcare professional informs and counsels parents of PND results in accordance with local policy and in a timely manner as per Programme standards i.e. within 5 days of having the PND procedure.

Baby affected by a major haemoglobin disorder: Parents to be referred for counselling and follow up by trained healthcare professionals. There should be a local protocol in

place. Specialist midwives, Sickle Cell and Thalassaemia Centres and Regional Genetic Centres (RGCs) provide counselling services, information and advice for families with, or 'at risk' of, genetic conditions. For a list of specialist centres refer to:

http://www.sickle-thal.nwlh.nhs.uk/information/nationalsicklecellthalassaemiacentres.aspx.

Continue pregnancy; include results on blood spot card: Maternity units should notify newborn screening laboratories of women continuing with affected pregnancies and parental results are to be included on newborn blood spot card. Parents must be informed that their child will be referred to paediatrician / haematologist / specialist counsellor as per local policy and programme standards.

An alert form should be sent to the newborn screening laboratory. Maternity units to complete the short-term and long-term PND outcome form.

Links to newborn sickle cell screening covered by Newborn Blood Spot Programme: The result of an antenatal genetic test will be related to the results of a newborn baby's test for the same condition, so the linked programme provides a check between the mother and baby result.

The postnatal report given to women following the birth of the baby should contain all antenatal screening results for communication to primary care and allow linkage to newborn screening results where relevant. There should be systems in place to inform newborn screening laboratories of all antenatal screening and diagnostic results. There should be a named person in every maternity unit with responsibility to ensure that newborn screening laboratories are informed of carrier or affected women whose pregnancies are ongoing.

Discontinue pregnancy: Offer counselling and follow up support.

Women who miscarry or terminate their pregnancy following screening:

There should be a mechanism in place to ensure that women who subsequently miscarry or terminate their pregnancy receive their antenatal screening results to allow appropriate management and reproductive choice.

The postnatal report should contain all antenatal and PND screening results for communication to primary care.

Known at risk women should be given a direct contact to ensure early access to counselling in future pregnancies and given information on pre-implantation genetic diagnosis.

Figure 1: Sickle Cell and Thalassaemia (SCT) screening care pathway

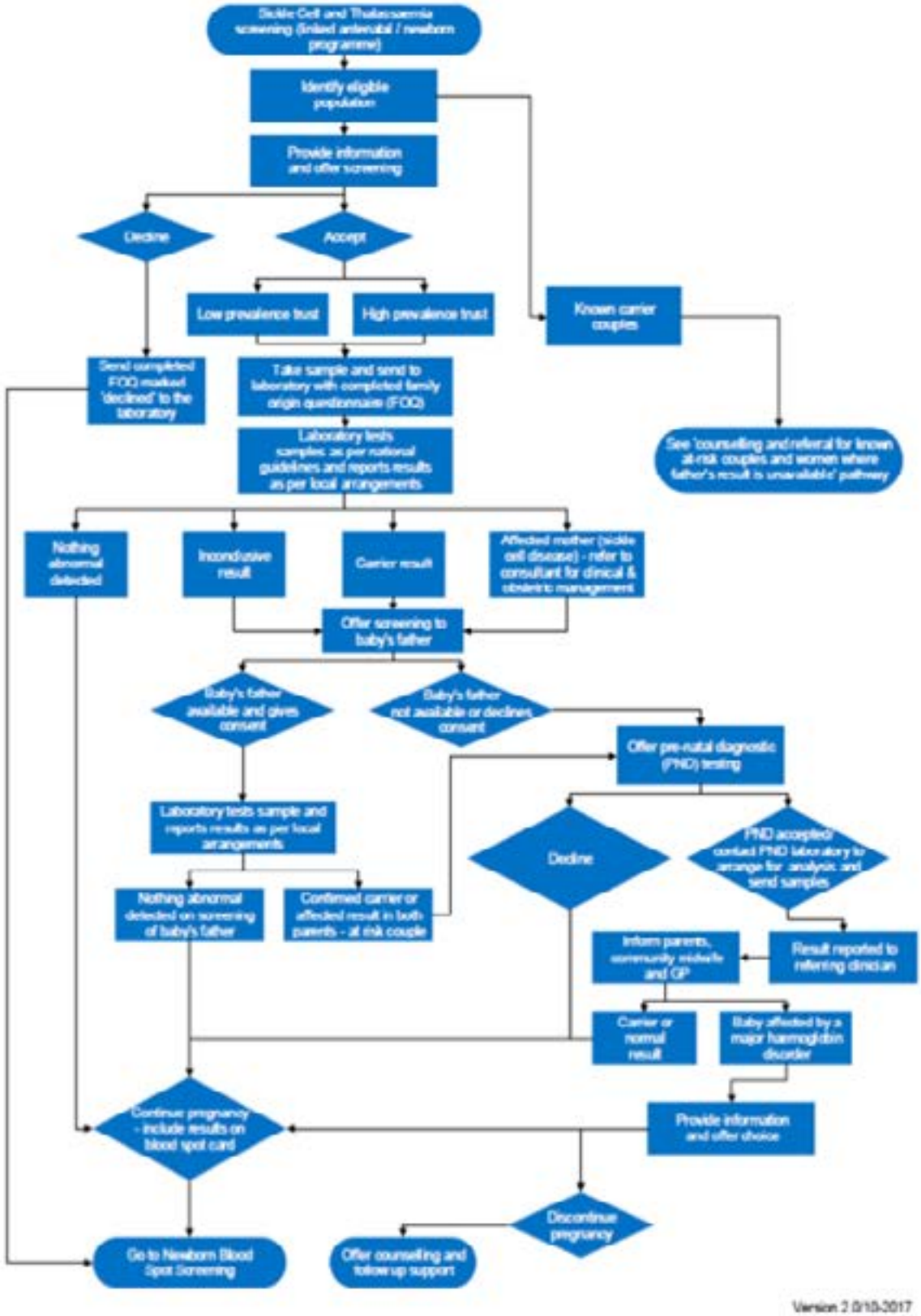
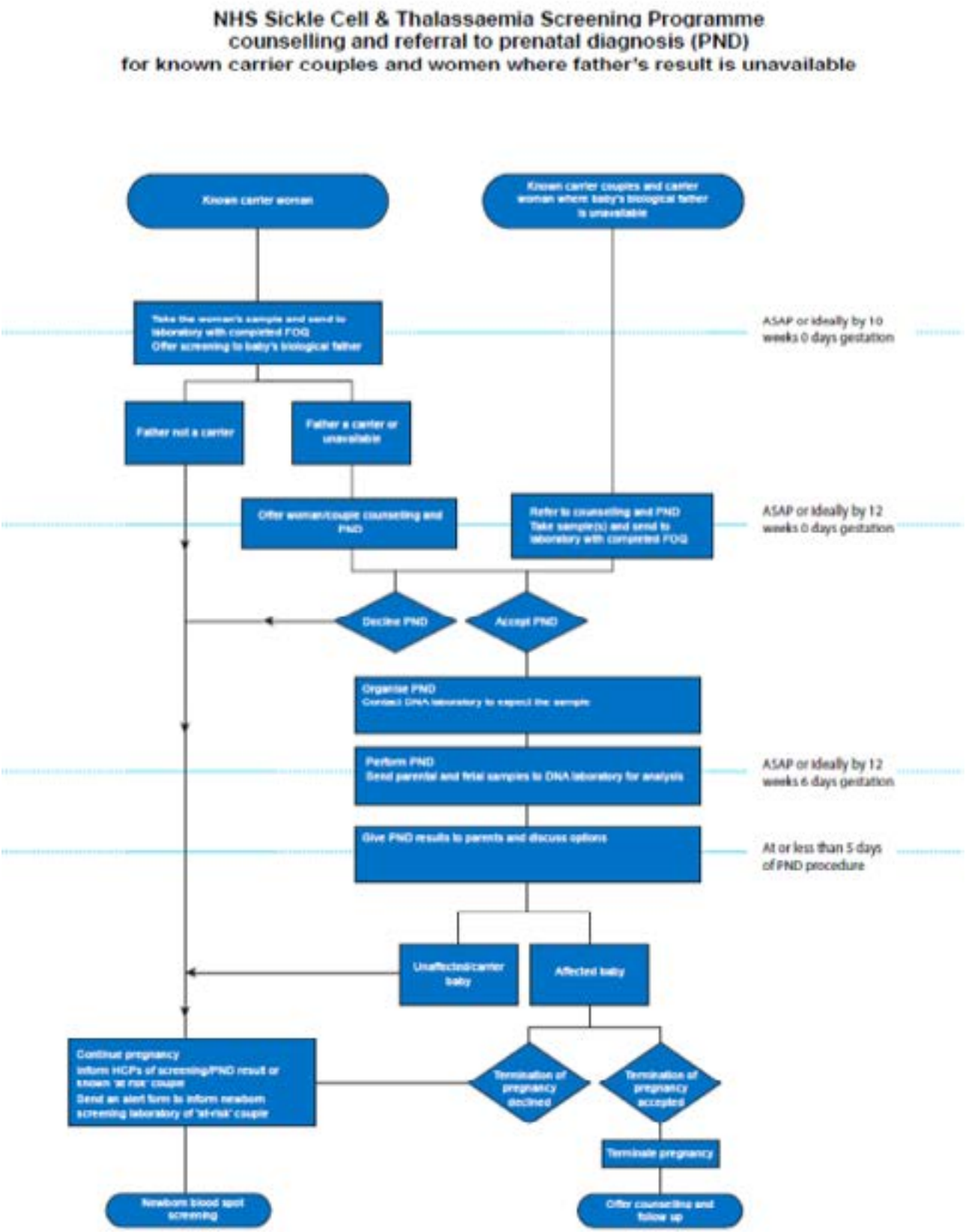


Figure 2:


NHS Sickle Cell & Thalassaemia Screening Programme
counselling and referral to prenatal diagnosis (PND)
for known carrier couples and women where father's result is unavailable
Known carrier woman
Known carrier couples and carrier woman where baby's biological father is unavailable
Take the woman's sample and send to laboratory with completed FOG
Offer screening to baby's biological father
ASAP or ideally by 10 weeks 0 days gestation
Father not a carrier
Father a carrier or unavailable
Offer woman/couple counselling and PND
Refer to counselling and PND
Take sample(s) and send to laboratory with completed FOG
ASAP or ideally by 12 weeks 0 days gestation
Decline PND
Accept PND
Organise PND
Perform PND
Send parental and fetal samples to DNA laboratory for analysis
ASAP or ideally by 12 weeks 6 days gestation
Give PND results to parents and discuss options
At or less than 5 days of PND procedure
Unaffected/carrier baby
Affected baby
Continue pregnancy
Inform HCPs of screening/PND result or known 'at risk' couple
Send an alert form to inform newborn screening laboratory of 'at-risk' couple
Termination of pregnancy declined
Termination of pregnancy accepted
Terminate pregnancy
Newborn blood spot screening
Offer counselling and follow up

Newborn Sickle Cell Disease Screening within Newborn Blood Spot Screening Programme

The pathway for newborn screening for sickle cell is an integral part of the NHS Newborn Blood Spot Screening Programme see Service Specification No. 19: NHS Newborn Bloodspot Screening Programme https://www.england.nhs.uk/commissioning/wp- content/uploads/sites/12/2016/04/serv-spec-19-chld-blood-s pot-screening.pdf and Guidelines for Newborn Blood Spot Sampling.

Specific issues for newborn screening for sickle cell disease:

- registered health care professionals check antenatal results (including results from baby's biological father) and family history and record antenatal results on the blood spot card
- a pre-transfusion sample to screen for sickle cell disease is taken on all babies admitted to a neonatal unit. The blood spot card should be marked "pre-transfusion"
- the "pre-transfusion" blood spot card should be stored with the baby's medical record in line with local protocols and dispatched to the newborn screening laboratory together with the day 5 sample if the baby has received a blood transfusion in the interim
- as a failsafe, transfused babies who did not have a pre-transfusion sample taken can be tested for sickle cell disease using DNA analysis. Such samples are sent by the newborn screening laboratory to the DNA laboratory

Newborn screening results in one of four outcomes:

- condition not suspected: parents are informed of the result
- baby is identified as a carrier: results are reviewed against maternal and paternal results where these are available (to assist communication and identify any cases where misdiagnosis or non-paternity could be an issue); then parents are informed by a trained health care professional, ideally by face-to-face discussion, or by letter with offer of a face-to-face session
- inconclusive result: additional sample may be required
- condition suspected: immediate clinical referral initiated and parents informed of the result face to face. All screen positive results must be given to parents by a trained healthcare professional, by 28 days of age. The baby must enter care by 90 days of age

Newborn screening laboratory scientists and clinicians responsible for the care of screen positive infants should use PHE's Newborn Outcome System to refer screen positive infants from screening into treatment services.

This system allows users to view the status of patients along the care pathway and alerts clinicians when important milestones are breached; it will automate the gathering and reporting of newborn outcome data and KPIs and integrate with the National Haemoglobinopathy Registry (NHR) and with NCARDRS.

2.3 Roles and accountability through the screening pathway

The linked NHS SCT screening programme is dependent on systematic specified relationships between stakeholders. Stakeholders include staff from maternity units, the antenatal, newborn and pre-natal diagnosis screening laboratories, diagnostics laboratory and genetics services, child health records departments, and specialist sickle cell and thalassaemia services, i.e. 'the screening pathway'.

NHS England and NHS Improvement will be responsible for ensuring that the pathway is robust.

The provider will be expected to fully contribute to ensuring that systems are in place to maintain the quality of the whole screening pathway in their organisation.

This will include, but is not limited to:

- provision of robust co-ordinated screening that ensures all parties are clear of their roles and responsibilities, so that there is clarity of handover of responsibility throughout all elements of the screening pathway
- ensuring that midwifery services are supported to facilitate early booking and testing for maternity care within primary, community and hospital care settings
- developing joint audit and monitoring processes
- operating an escalation process for screening incidents
- agreeing and documenting roles and responsibilities relating to all elements of the screening pathway across organisations including CHRDs
- agreeing joint checks and mechanisms where required to ensure safe and timely processes across the whole screening pathway (see SCT: checks and audits to improve quality and reduce risks https://www.gov.uk/government/publications/sct-checks-and-audits-to-improve-quality-and-reduce-risks)
- contributing to any of NHS E and public health screening lead initiatives in screening pathway development in line with NHS Screening Programmes expectations
- providing or seeking to provide robust electronic links with relevant organisations
- links with primary, secondary and tertiary care
- the need for robust IT systems across the screening pathway

All providers should have the following posts in place

- screening midwife/coordinator (and deputy) to oversee the screening programme and act as a link between others members of the SCT multidisciplinary team
- health care professionals/SCT counsellors to provide genetic risk assessment and counselling.

2.4 Commissioning arrangements

Sickle cell and thalassaemia screening services will be commissioned by NHS E alongside specialised services where appropriate. Commissioning the sickle cell and thalassaemia screening pathway involves commissioning at different levels which may include NHS E, CCGs and directly by maternity services.

2.5 Links between screening programme and national programmes expertise

The role of PHE Screening

The national screening team in Public Health England (PHE Screening) provides expert advice and support to NHS Screening Programme. It does those things which make sense to do once rather than by each individual screening service. This includes:

- developing and monitoring standards
- producing public information leaflets
- quality assurance of local screening services
- enabling accessible training and education

Providers should subscribe to the PHE Screening blog for the latest national news and updates. National documentation and guidance is published on GOV.UK.

Section 3: Delivery of Screening Programme

3.1 Service model summary

See section 2.2 care pathway above for further details

As with all newborn and antenatal screening the process of the offer of screening is largely embedded within the routine maternity and newborn pathway and not as a separate service.

There are key points about this programme which make it different, and also are relevant to effective commissioning. The key points for the SCT Programme are:

- the importance of early testing in pregnancy to enable women to exercise choice
- recognition of the impact of lifetime genetic information. As an increasing proportion of women and their partners are aware of their carrier status before pregnancy, the choice of direct access for counselling and offer of PND rather than routine pregnancy care should be available
- the interface between maternity, laboratories, specialist counselling service and specialist diagnostic services
- the importance of timely and reliable communication by newborn screening laboratories of screen positive results to the local clinician/and the designated sickle cell and thalassaemia centre to ensure that affected babies enter the clinical care pathway
- linkage with primary care and CHRDs

All elements of the screening pathway should be delivered by appropriate staff and to national standards and guidelines.

3.2 Programme co-ordination

The provider will be responsible for ensuring that the part of the programme they deliver is co- ordinated and interfaces seamlessly with other parts of the programme with which they collaborate, in relation to timeliness and data sharing.

The provider will ensure that they have a screening midwife/coordinator (and deputy) in place to oversee the screening programme, supported by appropriate administrative support to ensure timely reporting and response to requests for information. Where there is only one named co-ordinator, the provider will ensure that there are adequate cover arrangements in place to ensure sustainability and consistency of programme.

The provider and NHS England and NHS Improvement should meet at regular intervals to monitor and review the local screening pathway. The meetings should include representatives from programme co- ordination, clinical services, laboratory services, service management and screening QA.

3.3 Governance and Leadership

The provider will:

- cooperate with and have representation on local oversight arrangements as agreed with NHS England and NHS Improvement commissioners
- identify a Trust director who is responsible for the screening programme
- ensure internal clinical oversight and governance by an identified clinical lead and a programme manager. The clinical lead has overall clinical responsibility for the programme across the pathway.
- provide documented evidence of clinical governance that includes:
    - compliance with the NHS Trust and NHS England and NHS Improvement information governance/records management
    - user involvement, experience and complaints
    - failsafe procedures
    - compliance with checks, audits and failsafe procedures https://www.gov.uk/government/publications/sct-checks-and-audits-to-improve- quality-and-reduce-risks
- ensure that there is regular monitoring and audit of the screening programme, and as part of the organisation's clinical governance arrangements, the board is assured of the quality and integrity of the screening programme
- produce an annual report of screening services, which is signed off by the board
- ensure the programme is delivered by trained workforce that meet national requirements

3.4 Definition, identification and invitation of cohort/eligibility

The target population to be offered screening antenatally is all pregnant women, and the biological fathers of babies whose mothers are carriers or affected.

In each pregnancy, the family origin questionnaire should be completed and a routine full blood count taken and mean cell haemoglobin (MCH) and other red cell indices should be assessed.

Women and couples who already know their carrier status should be offered direct referral to counsellors/antenatal screening coordinators for assessment of the couple risk status and PND.

The target population to be offered sickle cell screening as part of the NHS Newborn Blood Spot Screening Programme is all newborn babies and infants moving in to the country up to one year of age.

The provider will make every effort to offer screening and personal informed choice vulnerable and hard- to-reach groups within the eligible population. This includes babies born abroad who move into the country up to one year of age.

3.5 Location(s) of programme delivery

The provider will ensure appropriate accessible service provision for the population whilst assuring that all locations where SCT screening occurs fully comply with the policies, standards and guidelines referenced in this service specification.

3.6 Days/hours of operation

The provider will ensure that days and hours of operation are sufficient to meet the national programme standards.

3.7 Entry into screening programme

Antenatal: through GPs or direct referral into maternity services/SCT specialist centres. Whilst there is nothing specific in the GP contract regarding the SCT programme, general practitioners have a key role in ensuring that pregnant women referred to them are referred on as soon as possible to midwifery services and for holding previous antenatal results and results of newborn screening.

All maternity units should provide a publically available and advertised direct dial and/or email address for women, GPs and midwives.

Newborn: all newborn babies and infants moving in to the country under a year of age.

3.8 Working across interfaces

The screening programme is dependent on strong working relationships (both formal and informal) between professionals and organisations along the screening pathway. These include maternity services, the screening and PND laboratories, SCT counsellors, health visiting and specialist haematology clinical services.

Accurate and timely communication and handover across these interfaces is essential to reduce the potential for errors and ensure a seamless pathway for service users. It is essential that there remains clear named clinical responsibility at all times and at handover of care the clinical responsibility is clarified.

The provider will be responsible for ensuring that the pathway is robust. For their part the provider will ensure that appropriate systems are in place to support an interagency approach to the quality of the interface between these services. This will include, but is not limited to:

- ensuring that midwives are supported to facilitate early booking for maternity care within all care settings
- agreeing and documenting roles and responsibilities relating to all elements of the screening pathway across organisations
- providing strong clinical and managerial leadership and clear lines of accountability
- developing joint audit and monitoring processes
- working to nationally agreed programme standards and policies
- agreeing jointly on what failsafe mechanisms are required to ensure safe and timely processes across the whole screening pathway and implement checks and audits to improve quality and reduce risks

- contribute to any NHS E Screening Lead's initiatives in screening pathway development in line with NHS Screening Programmes expectations
- develop an escalation process for screening safety incidents
- The Specification for Specialised Services for Haemoglobinopathy Care (in development). Treatment services are under review, however this will have no impact on the screening care pathway. It will be published at: https://www.england.nhs.uk/commissioning/spec-services/npc-crg/blood-and-infection-group-f/f05/

3.9 Testing (laboratory service, performance of test by individuals)

Antenatal, newborn and DNA referral laboratories are expected to follow the policy guidance and standards laid out in '*Sickle Cell and Thalassaemia: Handbook for Laboratories*' and meet the programme standards.

3.10 Results giving

Screening results should be explained to women by appropriately trained staff and recorded in the woman's health records/IT systems.

See section 2.2 for further detail.

Newborn screening results should routinely be transferred to primary care in a standard format.

3.11 Transfer of and discharge from care obligations

Based on the generic screening objectives of the programme the antenatal screening pathway ends for:

- pregnancies resulting in a live birth when the antenatal (including baby's biological father and PND) screening results are included on the blood spot card
- women opting for termination of pregnancy when the woman is counselled appropriately following PND

Based on the generic screening objectives of the programme the newborn screening pathway ends for:

- condition not suspected and carrier results when parents and GPs are informed of the result
- screen positive results when the parents are informed of the result and the baby is seen and tested and diagnosis confirmed by a clinician and registered in the designated clinical network. More detail is available in the Specification for Specialised Services for Haemoglobinopathy Care (in development); it will be published at: https://www.england.nhs.uk/commissioning/spec-services/npc-crg/blood-and-infection-group-f/f05/

3.12 Public information

PHE Screening uses published best practice processes to develop public information leaflets. It also works with NHS Digital to ensure that information on the NHS.UK website

for the public is accurate.

Providers must:

- use the public information leaflets from PHE Screening at all stages of the screening
- pathway
- involve PHE in the development of any local awareness campaigns
- not duplicate clinical information on local websites
- involve PHE if they want to move from providing printed leaflets to online sources of information

Using the leaflets provided by PHE ensures accurate messages about the risks and benefits of screening and any subsequent surveillance or treatment are provided. PHE Screening must be consulted and involved before developing any other supporting materials.

Providers must involve PHE in the development of local publicity campaigns to ensure accurate and consistent messaging, particularly around informed choice, and to access nationally- developed resources. For local awareness campaigns, local contact details must be used so that the national screening helpdesk is not overburdened.

Local provider websites must not duplicate clinical information about screening but should be restricted to contact and logistical information. Links should be provided to the national information on NHS.UK (http://www.nhs.uk/Livewell/Screening/Pages/screening.aspx or the relevant programme page) and GOV.UK (https://www.gov.uk/topic/population-screening-programmes or the relevant programme page).

To support PHE Screening to carry out regular reviews of the national screening public information leaflets and online content, providers are encouraged to send PHE Screening the results of any local patient surveys which contain feedback on these national resources.

Ordering leaflets

Providers can order leaflets developed by PHE Screening for free for core screening purposes.

Leaflets are regularly updated so providers should not order more than 3 months' supply, or stockpile leaflets, as they could become out of date and need to be destroyed. Leaflets for non-core activities, such as local health promotion purposes, can be bought from the national print provider.

PHE can only provide one leaflet per person per screening episode. A screening episode is defined as an invitation (with any subsequent reminders) for a particular screening test. People who are referred for further assessment following a screen should get a single copy of the appropriate follow-up leaflet.

Antenatal and newborn screening is treated as a single episode, so women should get a single copy of Screening Tests For You and Your Baby to last the entire antenatal and newborn period. This means that duplicate copies should not be provided with reminder letters or if people lose or forget their leaflet. They should be signposted to electronic sources of information instead.

3.13 Exclusion criteria

Newborn:

- babies stillborn or who died before day 5
- children over 1 year of age.

3.14 Staffing, Education and Training

In accordance with NHS Screening programmes standards and protocols the provider will ensure that there are adequate numbers of competent and appropriately trained staff in place across the screening pathway to deliver the screening programme in line with best practice programme and laboratory guidelines.

Qualifications will be specific to staff delivering the service across the care pathway. Staff must demonstrate competence (which is linked to training).

The provider will have in place a workforce plan designed to maintain a sustainable programme, especially where increase in birth rate are predicted and/or where there are difficulties in the recruitment of appropriately qualified healthcare staff.

Providers are responsible for funding minimum training requirements to maintain an effective screening workforce including appropriate annual CPD in line with programme requirements, for example a screening study day or completion of national NHS Screening Programme e-learning.

Providers must facilitate screener training in line with programme requirements as detailed in each NHS screening programme specification. Providers should ensure training has been completed satisfactorily and recorded and that they have a system in place to assess on- going competency.

The provider will ensure that counsellors / antenatal screening coordinators for the sickle cell and thalassaemia screening programme meet Sickle Cell & Thalassaemia counselling competences https://www.gov.uk/guidance/sickle-cell-and-thalassaemia-screening-education-and-training.

PHE screening makes available a variety of education and training for NHS screening staff. Evidence based, up-to-date e-learning resources, study days and courses can be accessed here https://www.gov.uk/guidance/nhs-population-screening-education-and-training

In addition each screening programme will have specific guidance for the initial training and ongoing learning for screeners. This learning should be facilitated, supported and monitored by local screening providers. In line with professional regulations individuals have a responsibility to ensure their practice is up-to-date and evidence based. Local programmes can use the national programme training guidance and resources to support this.

Courses have been developed in collaboration with King's College London (KCL).

The genetic risk assessment and counselling module and SCT screening: specialist counselling update are intended to provide a benchmark of professional competence to health professionals involved in counselling women and couples 'at risk' of having a child with a haemoglobinopathy.

3.15 User involvement

The provider(s) will be expected to:

- demonstrate that they regularly seek out the views of service users, families and others in respect of planning, implementing and delivering services
- demonstrate how those views will influence service delivery for the purposes of raising standards
- make results of any user surveys/questionnaires available to NHS E and screening QA teams on request

3.16 Premises and equipment

The provider will:

- ensure that suitable premises and equipment are provided for the screening programme
- have appropriate polices in place for equipment calibration and electronic safety checks, maintenance, repair and replacement in accordance with manufacturer specification to ensure programme sustainability
- ensure that equipment meets the European Council Directive, enforced by the Medicines and Healthcare Regulatory Agency, to ensure that it is safe and effective to use

Electronic provision of the FOQ information with the antenatal order for sickle cell and thalassaemia screening is recommended in order to ensure complete data with every sample.

3.17 Safety & safeguarding

The provider should refer to and comply with the safety and safeguarding requirements as set out in the NHS standard contract. As an example, please see link below for 2016/17 NHS standard contract http://www.england.nhs.uk/nhs-standard-contract/.

Section 4: National standards, risks and quality assurance

The provider will:

- meet the acceptable national screening standards and work towards attaining and maintaining the achievable standards adhere to specific professional standards and guidance https://www.gov.uk/government/collections/nhs-population-screening-programme-standards
- maintain a register of risks, working with NHS England and NHS Improvement and quality assurance teams within Public Health England to identify key areas of risk in the screening pathway, and make sure these points are reviewed in contracting and peer review processes
- participate fully in national quality assurance (QA) processes which includes:
    - submitting agreed minimum data sets and reports from external quality assurance schemes
    - undertaking ad-hoc audits and reviews as requested
    - completing self-assessment questionnaires / tools and associated evidence
    - responding to SQAS recommendations within agreed timescales providing specified evidence
    - producing with agreement of commissioners of the service an action plan to address areas for improvement that are identified in recommendations
- all screening laboratories must
    - be accredited by the UK Accreditation Service (UKAS) to ISO. 'Medical laboratories – Requirements for quality and competence (ISO 15189)' or be actively working towards ISO 15189 and UKAS accreditation
    - participate in EQA schemes accredited to ISO. 'Conformity assessment. General requirements for proficiency testing schemes (ISO 17043)'
    - meet the screening programme quality assurance requirements mapped to ISO 15189
    - and use ISO 15189 accredited reference laboratories that are UKAS accredited for the relevant screening activities requested by the referring laboratory

The UK Accreditation Service (UKAS) will look at both ISO 15189 and the screening requirements on behalf of PHE Screening Quality Assurance and disclose these assessments to PHE Screening

- operate and evidence
    - check points that track individuals through the screening pathway

    - identify, as early as possible, individuals that may have missed screening, where screening results are incomplete or where referral has not happened
    - have process in place to mitigate against weakness in the pathway
- have arrangements in place to refer individuals to appropriate treatment services in a timely manner and these should meet national screening standards
- demonstrate that there are audited procedures, policies and protocols in place to make sure the screening programme consistently meets programme requirements
- comply with guidance on managing safety incidents in national screening programmes and NHS England and NHS Improvement serious incident framework
- ensure business continuity - business continuity plans must be in place where required
- ensure sub-contracts and/or service level agreements with other providers meet national standards and guidance

Service improvement:

The provider will develop and agree with commissioners a CSIP (continual service improvement plan) in cases where national recommendations and/or screening standards are not fully met. The CSIP will include the following:

- action plans specifying changes and improvements that will be made during the contracting period
- defined timescales for actions
- roles and responsibilities for actions
- performance issues highlighted by the commissioners
- concerns raised by service users.

New technologies:

New technologies should not be used for screening unless approved by the UK National Screening Committee.

Section 5: Data and intelligence

The collection, analysis and comparison of good quality data is critical for the all NHS screening programmes in England.

PHE Screening aims to develop a consistent approach to data collection and reporting across all screening programmes and is committed to making sure that 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

Performance thresholds are selected to align with existing screening standards and service objectives; 1 or 2 thresholds are specified.

The acceptable threshold is the lowest level of performance which screening services are expected to attain to assure patient safety and service effectiveness. All screening services should exceed the acceptable threshold and agree service improvement plans to meet the achievable threshold. Screening services not meeting the acceptable threshold are expected to put in place recovery plans to deliver rapid and sustained improvement.

The achievable threshold represents the level at which the screening service is likely to be running optimally. All screening services should aspire to attain and maintain performance at or above this level.

5.1 Key performance indicators (KPIs) and screening standards

The provider should adhere to the requirements as specified on following web pages:

KPIs: “Reporting data definitions” at https://www.gov.uk/topic/population-screening-programmes/population-screening-data-key-performance-indicators

Screening standards: https://www.gov.uk/government/collections/nhs-population-screening-programme-standards

Please note that indictor definitions are updated regularly and you should always obtain the most recent version available.

5.2 Data collection, monitoring and reporting

Providers should:

- ensure that appropriate systems are in place to support programme delivery including audit and monitoring functions
- continually monitor and collect data regarding its delivery of the service
- comply with the timely data requirements of the national screening programmes and regional quality assurance teams. This will include the production of annual reports. The most up to date dataset can be accessed from the national screening programme website
- collect and submit:

- Annual data returns by: -
    i. all antenatal laboratories,
    ii. all newborn laboratories
- (iii) the DNA laboratories (PND) including data on pregnancy outcomes for those women who have undergone PND
- (v) two DNA laboratories (newborn screening) for babies who have had a blood transfusion

Other:

For quality and monitoring, data from women at risk of having a baby affected by SCT who accept PND and data on babies who screen positive are shared with NCARDRS.

5.3 Public Health Outcomes Framework (PHOF)

PHE Screening contributes to “PHOF indicator 2.20 – National Screening Programmes”. Each screening programme reports on one or more sub-indicators.

The SCT Programme contributes to the Public Health Outcomes Framework. Indicator iii: The percentage of pregnant women eligible for antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available at the day of report.

Key deliverable: The acceptable level should be achieved as a minimum by all services

- acceptable ≥ 95.0%
- achievable ≥ 99.0%

See: https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/545605/PHOF_Part_2.pdf