Neonatal Haemoglobinopathies - Paediatric Full Clinical Guideline – Derby only

Reference no: NIC SS 14/Oct 20/v003

1. Introduction

- Newborn blood spot screening (NBSS) forms part of the process of the national linked antenatal and neonatal screening programme for haemoglobinopathies and sickle cell disease.
- The overall aim of screening is to achieve early detection, referral and treatment of babies affected by sickle cell and other haemoglobin disorders, in order that the lowest possible childhood death and morbidity rates occur (NHS Sickle Cell and Thalassaemia Screening Programme [NHS SCATSP], 2017). The NBSS is not intended to capture all carrier babies or all other haemoglobin variants, but to identify babies born with conditions where early intervention is likely to be beneficial (NHS SC&TSP Standards, 2017). See below which conditions are screened for via NBSS.
- About 1 in 2000 babies born in the UK has sickle cell disease (SCD), which is a serious inherited blood disease and require specialist care throughout their lives
- The first time an untreated child with sickle cell disorder is symptomatic may be fatal. Ideally prophylactic penicillin would be started before the child is 2 months old (NHS SC&TP, 2012).
- Babies born 'at risk' of inheriting a major haemoglobin disorder (where prenatal diagnosis has not been performed) enter into a different care pathway at birth, which is discussed later in the guideline.

2. Conditions screened for

- 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.
- Sickle cell disease and thalassaemia disorders, or other haemoglobinopathies, are autosomal recessive conditions affecting the structure or synthesis of haemoglobin
- Infants are at risk of inheriting these disorders only if both parents are carriers and/or suffer from the disease.
- Carrier states of sickle cell and other haemoglobinopathies may also be diagnosed, but although β thalassaemia major may be detected on newborn screening, the carrier state is unlikely to be identified.

3. Screening

- When using this guideline please cross-reference to the full Derby Teaching Hospital NHS Foundation Trust (DTHFT) Antenatal Screening Guideline (H11) for further information on the antenatal screening process and identification of high risk couples, and to the Newborn Blood Spot Guideline (N7). Both are available on FLO.
- Although the NBSS Programme strongly recommends all babies be screened, parents are entitled to accept or decline either all or any of the tests.

- Parents should have received information during pregnancy about the NBSS and been counselled if they are a high risk couple.

- Where high risk couples have been identified in the antenatal period via maternal & father of baby screening, the ANSC completes a *'Notification of couples at risk of having a baby with sickle cell & thalassaemia'* alert form and emails a scanned copy to: newborn.screening@nhs.net

- Where a high risk couple have been offered, but declined prenatal diagnosis, the ANSC (in addition to the newborn screening alert) partially complete a blood test request form and files in the baby notes for a venous sample to be taken at birth and sent to Oxford Reference lab for DNA analysis.

- Parents should have received 'Screening tests for you & your baby' booklet (Public Health England (PHE, 2017)), which includes information on routine NBSS:

    - How screening can help babies with sickle cell and other haemoglobinopathy disorders
    - How the sample is taken
    - When the parents should receive the result
    - That screening may identify babies who are carriers
    - That screening is not 100% accurate

- A discussion with the parents should be documented along with the parental acceptance/decline

- If parental consent is given, the screening may be performed according to detailed guidance in 3 publications from PHE:

    Guidelines for Newborn Blood Spot Sampling (2016)
    Newborn Blood Spot Screening in the UK; Health Professional Handbook (2012)
    Newborn Blood Spot Screening in the UK; Standards (2017)

- Blood spot samples are stored for at least 5 years, potentially for re-testing, re-analysis, or research. In the future a situation may arise where researchers would contact the family directly. If parents would not wish this, NO RESEARCH CONTACT should be written on the sample.

- All samples should be posted within 24 hours and record made of the date of sample and postage

- Any maternal variant should be documented on the NBSS card at the time of taking the sample.

4. Decline of screening

If screening is declined, this should be recorded in the maternity records and Personal Child Health Record, and the NBSS card should be sent to the laboratory stating DECLINED HAEMOGLOBINOPATHY SCREEN. The GP and Health Visitor (HV) should be informed in case the baby becomes unwell at any stage and the parents should be given contact details for further discussion and/or change of mind. (If other, or all, conditions for screening are declined the NBSS card should indicate this, and the GP and HV notified accordingly).

5. Pathway for communicating results

- All NBSS screen negative or haemoglobinopathy carrier results should be routinely reported to parents by 6 weeks of age
- Any baby born to high risk couples should already be known to the Sheffield Newborn Screening Laboratory
- Once the neonatal sample has been sent to the lab then the Antenatal Screening Coordinator (ANSC) must be informed by DTHFT Specialist Haematology Lab.

- Please then confirm by email to dhft.antenatalandnewbornscreeningRDH@nhs.net

- Copy into the email:
  Antenatal Screening Coordinator Tracy Doucas tracy.doucas@nhs.net
  SpMW cover for ANSC Henriette Green - henriette.green@nhs.net
  Senior BMS- gail.ford1@nhs.net
  Haematology Specialist Nurse Emma Bush – emma.bush1@nhs.net
  Consultant Paediatrician Claire Weights - claire.weights1@nhs.net,
  Consultant Haematologist Caroline Harvey- caroline.harvey13@nhs.net
  Haemoglobinopathy CNS Phone or email Oxford Reference lab to let them know the newborn sample has been sent
- The ANSC should then book the baby into Dr Weights' clinic using code CLW4A or CLW5P for 3-4 week time. Please email paedclinicchanges@nhs.net and ask for a screening appointment and for a patient letter to be sent. If there are any problems then please contact Dr Weights.

- Ensure parents of potential screen positive infants have an appointment receive results by ≤ 28 days of age

- The ANSC will also update the Maternity HBE referral spreadsheet with the baby details.

- Once the result is back from Oxford, the Lab will inform the ANSC via phone. This report will be scanned into CITO/Lorenzo.

- The ANSC to update the Maternity HBE referral spreadsheet with the results.

- If the baby is affected with a major haemoglobin disorder then they will already have an appointment with Dr Weights and will be seen in clinic in a timely manner. Results SHOULD NOT be phoned through to parents. Dr Weights will discuss the results with the parents in clinic. If appropriate the ANSC, Health Visitor and Haemoglobinopathy CNS will also attend the clinic appointment.

- If the baby not does have a haemoglobin disorder or is a carrier then the ANSC will call the parents with the good news and file a copy the Oxford report in the baby notes.

- The ANSC must also cancel the outpatient appointment via email on paedclinicchanges@nhs.net.

- If the baby is identified by NBSS or the Oxford Neonatal DNA Results as a carrier the Haemoglobinopathy CNS will send out a letter, card and booklet to parents. This also applies to normal results from the Oxford DNA Reports.

6. References (including any links to NICE Guidance etc.)

NHS Sickle Cell and Thalassaemia Screening Programme (NHS SCATSP) (2017). Standards for the Linked Antenatal and Newborn Programme. PHE
https://www.gov.uk/government/publications/standards-for-sickle-cell-and-thalassaemia-screening

NHS Sickle Cell and Thalassaemia Screening Programme (NHS SCATSP) (2012). Information for healthcare professionals
https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/402643/HCP_leaflet.pdf

PHE (2017a) Screening tests for you and your baby booklet
https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/622333/Screening_tests_for_you_and_your_baby_information_booklet.pdf

PHE (2017b) Newborn blood spot screening programme standards
https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/585415/Newborn_Blood_Spot_Screening_Programme_Standards.pdf

PHE (2016) Guidelines for Newborn Blood Spot Sampling
https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/511688/Guidelines_for_Newborn_Blood_Spot_Sampling_January_2016.pdf

https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/520749/Guidelines_for_Newborn_Blood_Spot_Sampling_quick_guide_May_2016.pdf

PHE (2014). Health professional handbook. A guide to Newborn Blood Spot Screening:
https://www.gov.uk/government/publications/health-professional-handbook-newborn-blood-spot-screening

7. Documentation Controls

<table>
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Contact for Review	This should match the author if different please state who the contact is by Job Title
Lead Executive Director Signature	

Neonatal Haemoglobinopathies - Summary Guideline