Antenatal Sickle Cell & Thalassaemia Screening Programme protocol (CG475)

Approval and Authorisation

<table>
<thead>
<tr>
<th>Approved by</th>
<th>Job Title or Chair of Committee</th>
<th>Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>Maternity Clinical Governance Committee</td>
<td>Chair, Maternity Clinical Governance Committee</td>
<td>4th March 2016</td>
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</tbody>
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Change History

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<th>Author</th>
<th>Reason</th>
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<td>June 2009</td>
<td>J. Young</td>
<td>Trust requirement</td>
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<td>April 2010</td>
<td>J. Young</td>
<td>Updating requirement</td>
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<tr>
<td>2.1</td>
<td>Sept 2010</td>
<td>J. Young</td>
<td>Appendix added &amp; section 8.0 amended</td>
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<td>April 2012</td>
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<td>Reviewed</td>
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<td>J Young</td>
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<tr>
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<td>J Young</td>
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Appendix A – Equality Impact Assessment Toolkit

This Protocol should be read in conjunction with the following:

☐ Antenatal Screening protocol (CG474)
1.0 Purpose
This document is the protocol for Antenatal Screening for Sickle Cell and Thalassaemia for the Royal Berkshire Hospital NHS Foundation Trust. It replaces any previous protocol and procedure documents.

The protocol is subject to annual review.

2.0 Function
The purpose of this protocol is to ensure adherence to The National Screening Committees Standards of practice for Sickle Cell and Thalassaemia Screening Programme.

2.1 Aims
- To offer early screening and diagnosis as indicated to all pregnant woman (preferably by 12+6 weeks of pregnancy) attending for antenatal care booked to deliver at the Royal Berkshire Hospital Maternity Unit.

- To provide adequate high quality information on the screening process to enable the woman and her partner to make an informed decision on whether to accept or decline the offer of screening.

- To ensure adequate support and counselling to all women with a confirmed diagnosis as the result of screening, who have chosen to end their pregnancy.

- For women with a confirmed diagnosis from further testing as a result of screening, who choose to continue their pregnancy, to provide optimal management for follow-on care during pregnancy and the subsequent newborn and childhood periods.

2.2 Objectives
- To identify and offer screening to all eligible women/couples, either pre-conceptually or at the booking appointment with Community Midwife. This is a High Prevalence Trust so all pregnant women are offered screening for Sickle Cell and Thalassaemia. (See section 3.1)

- To process and report on screening tests in a timely manner as detailed in the UK NSC working standards for the NHS Sickle Cell and Thalassaemia Screening Programme specific standards. (See sections 4 and 6)

- To provide adequate information and support for women with a confirmed diagnosis.
- At all stages in the screening process, to provide information in appropriate media that meets national standards. (See section 3.1 and 3.2)

- To facilitate choices in specific timescales, with appropriate counselling, of screening tests and Pre Nataal Diagnosis (PND), in screen positive women/couples that meet UK NSC working standards for the NHS Sickle Cell and Thalassaemia Screening Programme specific standards. (See sections 4 and 6).

- To promote an appropriate level of knowledge for all health professionals involved in the screening programme.

- To minimise the adverse effects of screening: anxiety, misunderstanding, inaccurate information, unnecessary investigation and follow-up, and inappropriate disclosure of patient specific information.

- To effectively audit and monitor the screening process and compliance with national standards and directives. To have in place systems for risk assessment and management of adverse incidents occurring during the screening process.

3.0 Content

3.1 Process of screening

- The population served by the Royal Berkshire Hospital NHS Foundation Trust has been identified as a high prevalence district for sickle cell. Therefore, all women booking for antenatal care will be offered the screening test for Sickle cell and Thalassaemia as early as possible in pregnancy and definitely at booking, with an accompanying FOQ. This will ensure there is equity and equality in the provision of screening in line with practice recommended by the UK NSC.

- All women will be offered appropriate screening in a timely manner irrespective of gestation to enable reproductive choice and to provide optimum pregnancy management.

- Women are offered testing with every pregnancy and therefore will receive appropriate pregnancy management.

The National Institute for Clinical Excellence (NICE) Antenatal Care Guidelines (2008) recommends that women have access to maternity services at 8 -10 weeks of pregnancy to give them time to consider early screening options. This is also
Antenatal Sickle Cell & Thalassaemia Screening Programme protocol (CG475) March 2016


- **Minimum Standard**
  50% of women booking in early pregnancy are offered screening and tested by 10+0 weeks

- **Achievable Standard**
  75% of women booking in early pregnancy are offered screening and tested by 10+0 weeks

Screening should be preceded by the provision of written information, where possible in the woman's first language, and there should be access to additional information and support in other suitable formats. The UKNSC booklet ‘Screening tests for you and your baby’ should be given prior to obtaining a sample for screening. Where English is not the woman’s first language arrangements should be made to obtain a version in her first language from https://www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief, or an appropriate interpreter should be used to enable the woman to understand the information she is given. This is booked via Prestige Network either online rkh@prestigenetwork.com, or phone 01635 246700.

The offer of screening and the decision to accept or decline, and written information given, should be recorded in the women’s notes. This allows for auditable evidence that screening has been offered.

These standards are audited by Key Performance Indicators (KPI’s) on a quarterly basis

- **Minimum Standard**
  95% of women will have a documented offer or screening
  90% of samples submitted will be supported by a Family Origin Questionnaire (FOQ)

- **Achievable Standard**
  99% of women will have a documented offer of screening
  95% of samples submitted will be supported by a Family Origin Questionnaire (FOQ)

### 3.2 Results

All women will be informed of their screening test results. See Appendix 1

- Normal results are put onto the Eproa pathology results system by the Haematology Lab staff. These are accessed by the Community Midwife at
the 16 weeks appointment and relayed to the patient. The Community Midwife is responsible for documenting the results in the hand held notes.

- Variant results are transferred electronically from Haematology to the SCO who then contacts the woman with her result and offers an appointment to discuss partner screening. These are also entered onto Eproa as above. Women identified as having a haemoglobin variant or thalassaemia trait will be offered testing of the baby’s father. It may be relevant to discuss paternity issues.

  **Minimum Standard**
  100% of carrier results should be reported to women
  85% of carrier results should be provided verbally and in writing, with supporting materials

- **Achievable Standard**
  100% of carrier results should be reported to women
  95% of carrier results should be provided verbally and in writing, with supporting materials

Paternal testing will be offered to partners of all women with haemoglobin variants or thalassaemia.

The Screening Coordinator (SCO) will post an appointment to the women. If the woman attends for an appointment, a pathology form for partner blood is given, consent is obtained to put copy of the partners test result in woman’s maternity notes, stamped with Partner Result. If the partner attends the appointment the SCO will obtain a blood sample with consent at the time. The SCO sends appointments following the DNA policy. If no contact has been made by the woman after a minimum if two posted appointments the SCO informs the Newborn Screening lab via electronic linkage and informs the woman’s GP via post. Electronic linkage of maternal and paternal results are via the Haematology Database

- **Minimum Standard**
  70% of fathers of carrier women’s babies provided with information supported by appropriate materials

- **Achievable Standard**
  75% of fathers of carrier women’s babies provided with information supported by appropriate materials

Paternal consent/decline to release result data to ensure appropriate pregnancy management will be obtained prior to paternal screening.
3.3 **Identified Pregnancies at Risk – See Appendix 1**

All women/couples identified as having a pregnancy at risk will have the opportunity to discuss the result and options for further management by expert counsellors with decisions clearly documented.

- **Minimum Standard**
  - 50% of prenatal diagnosis performed by 12+6 weeks
  - 90% of PND results will be returned to the woman within 5 days of the PND procedure
  - 80% of requested terminations of pregnancy should be arranged within 5 days of the PND result

All women / couples should be offered counselling by expert counsellors pre and post PND if appropriate.

All at risk couples are seen jointly with Screening Co-ordinator and Haemoglobinopathies Nurse Specialist. All counsellors should be PEGASUS trained.

- **Achievable Standard**
  - 50% of women / couples counselled by 12 weeks
  - Decision documented in 90% of cases
  - 50% of prenatal diagnosis performed by 12+6 weeks
  - 90% of PND results will be returned to the woman within 5 days of the PND procedure
  - 100% of requested termination of pregnancy should be arranged within 5 days of the PND result

All women / couples should be offered counselling by expert counsellors pre and post PND if appropriate

All at risk couples are seen by the Screening Coordinator and Haemoglobinopathies Nurse Specialist. All counsellors should be PEGASUS trained.

- All parental decisions are documented in the hospital maternity notes
- SCO liaises with PND staff and Reference Laboratory re: timing of PND and special Lab requirements such as maternal and/or paternal blood samples to accompany CVS/amniocentesis sample
- Results from PND are faxed from the Reference Laboratory to the SCO who communicates with the woman, and organizes appropriate follow-up congruent with the woman’s wishes
There is a mechanism in place to inform the newborn screening laboratory of any identified parental haemoglobin variants. This is to enable seamless linkage of antenatal and newborn programmes to aid interpretation of newborn results and appropriate counselling and support for families.

- The Screening Coordinator informs the Newborn Screening Laboratory of any identified couples via an alert form
- At risk couples who have an affected fetus determined through PND and any at risk couple who have declined PND should be offered a capillary blood test from the neonate soon after birth. This blood is sent to JRH via RBH Haemoglobinopathy lab (ext 7756)

There are recognised professional responsibilities to support women and their partners throughout the screening process, and actions to be taken in the management of adverse events, adhere to QA assurances, as identified in the local care pathway:

- Standard Operating Procedure, Haemoglobinopathy Screening by High Performance Liquid Chromatography (HPLC) using The Biorad Variant 2
- Guideline on Antenatal Screening
- Guideline on Detection and Follow-up of Fetal Anomaly Guideline
- Maternity Clinical Governance and Risk Management Strategy and Policy

### 3.4 Education and training

The policy for education and training for the Royal Berkshire Hospital NHS Foundation Trust reflects National Guidance.

- There is provision of an education and training programme for all healthcare professionals involved in the screening process, which where appropriate utilises tools and media provided by the United Kingdom National Screening Committee (UKNSC).
- All newly employed healthcare professionals involved in the screening process will have training in the Trust’s sickle cell and thalassaemia screening programme as part of their induction programme.
- All healthcare professionals involved in the screening process will undertake update sessions on sickle cell and thalassaemia screening.
- All education and training provided should be evaluated and audited, where appropriate utilising tools and media provided by the UKNSC.
- Annual review of training needs to ensure that training is developed and delivered to meet the needs of all staff involved in antenatal screening programmes.
Information for healthcare professionals will be disseminated through the Screening Newsletter, update sessions and the Annual report.

4.0 Definitions

4.1 Aetiology
Sickle cell disease and Thalassaemia disorders, also known as haemoglobinopathies or haemoglobin disorders, 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.

They mainly affect individuals who have originated from Africa, Asia, the Caribbean, the Middle East, and the Mediterranean, but because of migration and subsequent integration, can affect any ethnic group.

There are two types of haemoglobin disorders categorised as qualitative or quantitative.

Thalassaemias are quantitative disorders, - alpha (α), beta (β), delta beta (δβ) and haemoglobin Lepore, and affect the quantity of haemoglobin produced. This results in partial or no production of the globin chains that form the structure of haemoglobin within the red blood cells. Thalassaemia major can result in severe anaemia. Regular treatments, including blood transfusions and chelation therapy are required from a very early age to enhance quality of life and long-term survival.

Sickle cell is a qualitative disorder affecting the quality of haemoglobin produced. The significant haemoglobin variants are haemoglobin S (sickle), C, D (Punjab), E and O (Arab). This condition is characterised by a chronic haemolytic anaemia, vaso-occlusion, and pain of varying intensity, subsequent organ damage, chronic ill health and reduced life expectancy. Sickle cell disease has a high mortality and morbidity rate in young children if left untreated.

4.2 Conditions screened for:
Significant maternal haemoglobinopathies should be detected by antenatal screening and are important for maternal care:

- Hb-SC
- Hb-S/β thalassaemia
- β thalassaemia intermedia
- Hb H disease
- Hb-SD Punjab
- Hb-SE
- Hb-SOArab
- Hb-S/Lepore
- Hb E/β thalassaemia
- (β thalassaemia major would be clinically apparent)
- All variants (excluding alpha thalassaemia) are reported and partner testing requested
- Alpha thalassaemia variant partner testing would depend on ethnicity.

4.3 **Maternal conditions requiring partner testing:**
- Conditions in (1)
- Carrier states in mother
  - Hb-AS
  - Hb-AC
  - Hb-AD Punjab
  - Hb-AE
  - Hb-AOArab
  - Hb-A Lepore
  - β thalassaemia trait
  - δβ-thalassaemia trait
  - αo thalassaemia trait
  - HPFH
  - Any compound heterozygote state including one or more of the above conditions
  - Any homozygous state of the above conditions
- All variants (excluding alpha thalassaemia) are reported and partner testing requested
- Alpha thalassaemia variant partner testing would depend on ethnicity

4.4 **Potentially significant disorders in the fetus that should be detected with PND**
- Hb-SS
- Hb-SC
- Hb-SD Punjab
- Hb-SE
- Hb-SOArab
- Hb-S/Lepore
- Hb-S/β thalassaemia
- Hb-S/δβ thalassaemia
- Hb-β/Lepore
- Hb Bart’s Hydrops Fetalis (αo/αo)
- β thalassaemia major (except cases with silent or near silent maternal phenotype)
- Hb E/β thalassaemia

The antenatal screening programme for sickle cell and thalassaemia requires close collaboration between community and hospital based antenatal services, primary care, laboratory services and specialist services.
5.0 Consultation

This protocol has been written in consultation with the following:

✓ Consultant Haematologist
✓ Haemoglobinopathies Nurse Specialist
✓ Lead Biomedical Scientist
✓ Consultant Obstetricians
✓ Public Health Lead
✓ Regional Screening Coordinator for the South East of England

6.0 Dissemination/Circulation

By Clinical Governance Committee

7.0 Monitoring of Effectiveness and Compliance

This protocol will be monitored for effectiveness by regular audit and presentation of these audits to Local and Regional interested parties:

- The Local Antenatal and Newborn Screening Group is responsible for establishing appropriate links with the laboratories to establish mechanisms for collection and reporting of data, in line with national proforma, for Sickle Cell & Thalassaemia screening.

- Data on number of tests, gestation at time of test, and uptake of screening supplied by Haematology Lab. Data on numbers of carriers identified, partner screens, at-risk couples and prenatal diagnoses collated and supplied by SCO

- The Local Antenatal and Newborn Screening Group is responsible for ensuring appropriate links are in place with regional quality management groups and the trust complies with regional arrangements for performance management

- The Trust will produce quarterly KPI figures and an annual audit report, pertaining to the previous fiscal year, to the NHS PHE and CCG
### 8.0 Local contacts

<table>
<thead>
<tr>
<th>Role</th>
<th>Contact Details</th>
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| Antenatal Screening Co-Coordinator | Jeanne Harris 01183228507  
Newborn Screening Co-Coordinator | Jo Young 01183227292 |
| Haemoglobinopathy Nurse Specialist | |
| Chair of Local Antenatal and Newborn Screening Group | Dr Suruchi Arora |
| Lead Obstetricians | Mr Mark Selinger  
Miss Samantha Low |
| Lead Haematologist | Dr Asif Khan |
| Lead Paediatrician | Dr C. Golding Ass Spec Reg |
| Lead Biomedical Scientist | David Wheeler / Susan Brown |
| Regional Cytogenetics Department | The Churchill Hospital, Oxford |
| National Haemoglobinopathy Reference Laboratory | The Churchill Hospital, Oxford, Dr Shirley Henderson 01865 225298 |
| NHS PHE | Voluntary sector representatives |
| Trust Bereavement Midwife | Kate Flack |

This service agreement can be translated on request to Jane Burnett patient Information Manager (0118 322 8706).

### 9.0 References

1. Antenatal and Newborn Screening programmes: NHS Sickle Cell and Thalassaemia Screening Programme 2011
3. Antenatal and Newborn Screening programmes: NHS Sickle Cell and Thalassaemia Screening Antenatal Screening Policy
4. Antenatal and Newborn Screening programmes: PEGASUS: Professional Education for Genetic Assessment and Screening.


10.0 Auditable standards
1. All pregnant women will be offered screening for anaemia. This screening will take place at booking, at 28 and 34 weeks. Haemoglobin results ≥ 11.0 at first contact or ≥ 10.5 from 28 weeks will be discussed at next antenatal appointment. All women will be informed of haemoglobin results < 11.0 at first contact or < 10.5 from 28 weeks and advised to start iron supplementation within 2 weeks of the result being given to the maternity services. The date of test taken, result, patient informed and action taken will be documented in the “investigation and results of test” page in the maternal health care record.

2. All pregnant women will be offered testing for blood group and rhesus status at booking and at 28 weeks. Results will be discussed at next antenatal appointment. All non-sensitised women who are rhesus D-negative will be advised to have routine anti-D prophylaxis. The date of test taken, result, patient informed and action taken will be documented in the “investigation and results of test” page in the maternal health care record.

3. All pregnant women will be offered screening for haemoglobin variants and thalassaemias at booking. The action plan will be stated in the laboratory result form and the SCO will follow up all cases of variant detected results. The date of test taken, result, patient informed and action taken will be documented in the “investigation and results of test” page in the maternal health care record.
Appendix 1 – Screening Test Results flowchart

AN Haemoglobinopathy for Sickle Cell & Thalassaemia

Pregnant woman screened preferably by 10/40. Counsellled by CMW. FOQ.

Positive result

Electronic notification from lab to SCO

Not attend

Appointment sent by couple by SCO

Repeat appointment sent

Attend

GP informed by SCO

High risk form sent to NBBS lab by SCO

Non-attendance

File in notes - non-attendance

Routine Day 5 NBBS

Affected

Unaffected

Affected

Continue

TOP

Haemoglobinopathy
CNS counselling

SCO

Accepted

Partner negative, SCO to check result

SCO file in maternity notes

Partner positive, SCO to check result

Couple counselled by CNS and SCO

Offered PND by SCO

Declined

File in notes

File in notes

Capillary blood at delivery to be sent to Oxford

February 2016

Royal Berkshire NHS Foundation Trust

This document is valid only on the date Last printed 22/03/2016 13:23:00
Appendix 2 – Management flowchart

Affected

Woman registered with GP/Midwife

Booking appointment CMW
Screening Bloods Taken

Affected found with routine screening bloods

SCO Referral

Cons referral

Follow Sickle Cell disease in pregnancy policy
RBH Haemoglobinopathy guidance March 2010.pdf

Disclosed at booking status affected

Cons referral by CMW

Follow Sickle Cell disease in pregnancy policy
RBH Haemoglobinopathy guidance March 2010.pdf

Jo Young (November 2013)
Appendix A – Equality Impact Assessment Toolkit

The questions in the table below should guide in assessing equality relevance/conducting screening.

For each of the six equality categories, ask the questions in the table below.

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<td>Do different groups have different needs, experiences, issues and priorities in relation to the proposed protocol?</td>
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<td>Is there potential for or evidence that the proposed protocol will not promote equality of opportunity for all and promote good relations between different groups?</td>
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<tr>
<td>Is there potential for or evidence that the proposed protocol will affect different population groups differently (including possibly discriminating against certain groups)?</td>
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<td>N</td>
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<td>N</td>
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<td>Is there public concern (including media, academic, voluntary or sector specific interest) in potential discrimination against a particular population group or groups?</td>
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Equality Impact Assessment Summary of Findings

Name of Protocol: Protocol for sickle Cell and Thalassaemia Screening Programme

Write short notes to explain why you have drawn your conclusions including any evidence (of whatever type) that you have to support your assessment.

Do different groups (age, disability, race, sexual orientation, gender, religion or belief) have different needs, experiences, issues and priorities in relation to the proposed protocol?

There is no current evidence to suggest any group would have different needs, experiences, issues or priorities in relation to this service agreement

Is there potential for or evidence that the proposed protocol will not promote equality of opportunity for all and promote good relations between different groups (age, disability, race, sexual orientation, gender, religion or belief)?

Do different groups have different needs, experiences, issues and priorities in relation to the proposed protocol?

There is no current evidence to suggest any group would have different needs, experiences, issues or priorities in relation to this service agreement

Is there potential for or evidence that the proposed protocol will not promote equality of opportunity for all and promote good relations between different groups (age, disability, race, sexual orientation, gender, religion or belief)?

Write short notes to explain why you have drawn your conclusions including any evidence (of whatever type) that you have to support your assessment.

Do different groups have different needs, experiences, issues and priorities in relation to the proposed protocol?

There is no current evidence to suggest any group would have different needs, experiences, issues or priorities in relation to this service agreement

Is there potential for or evidence that the proposed protocol will not promote equality of opportunity for all and promote good relations between different groups (age, disability, race, sexual orientation, gender, religion or belief)?

Author: Jo Young
Job Title: Newborn Screening Co-ordinator
Policy Lead: Group Director Urgent Care
Location: Policy hub/ Clinical/ Maternity/ Antenatal/ CG475

This document is valid only on the date Last printed 22/03/2016 13:23:00
This service agreement will promote equality, ensuring all users of this service are offered the screening test

Is there potential for or evidence that the proposed protocol will affect different population groups (age, disability, race, sexual orientation, gender, religion or belief) differently (including possibly discriminating against certain groups)?

The proposed service agreement offers equal care to all population groups

Is there public concern (including media, academic, voluntary or sector specific interest) in potential discrimination against a particular population group or groups (age, disability, race, sexual orientation, gender, religion or belief)?

There is no current evidence to suggest there will be any public concerns

Based on the information set out above I have decided that a full equality impact assessment is/is not necessary.

Name, Job title and signature: Jo Young, Newborn Screening Co-ordinator

Department: Antenatal Clinic, Maternity

Date: February 2016