East London
NHS Foundation Trust

# Antenatal Sickle Cell and Thalassaemia Screening Operational Policy

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| 2 | 14/11/2013 | Sekayi Tangayi | Service Manager/ Specialist Nurse | Revised |

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| 4 | Introduction | The local policy falls within the guidance issued by the National Programme Centre.1 | The local policy falls within the guidance issued by the Public Health England (PHE) NHS England Screening Programme Sickle Cell and Thalassaemia 2017 |
| 4 | Introduction | PHE - Sickle Cell & Thalassaemia Screening Programme. www.sct.screening.nhs.uk | PHE - Sickle Cell & Thalassaemia Screening Programme. www.gov.uk/phe/screening |
| 5 | Referral Criteria | Added new | All fathers-to-be, where antenatal screening show the mother to be a carrier |
| 5 | | | Guidelines for Newborn Blood Spot Sampling (2016) |
| 5 | Footnote | **Antenatal Haemoglobinopathy Screening & Follow-Up Policy East London NHS Foundation Trust & Newham University Hospital - April 2015 NHS Sickle Cell & Thalassaemia Screening Programme *Antenatal Screening Policy*** **http://www.sct.screening.nhs.uk/policy-antenatal** | www.gov.uk/phe/screening |
| 6 | National screening Policy | NHS Sickle Cell & Thalassaemia Screening Programme *Antenatal Screening Policy* http://www.sct.screening.nhs.uk/policy-antenatal | www.gov.uk/phe/screening |
| 6 | National screening Policy | NHS Sickle Cell & Thalassaemia Programme Laboratory Handbook 3rd Edition October 2012 http://sct.screening.nhs.uk/standardsandguidelines [Accessed February 2015] | www.gov.uk/phe/screening |
| 7 | footnote | **Antenatal Haemoglobinopathy Screening & Follow-Up Policy East London NHS Foundation Trust & Newham University Hospital - April 2015 NHS Sickle Cell & Thalassaemia Screening Programme *Antenatal Screening Policy*** | www.gov.uk/phe/screening |

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| | | **http://www.sct.screening.nhs.uk/policy-antenatal** | |
| 8 | footnote | **Antenatal Haemoglobinopathy Screening & Follow-Up Policy**<br>**East London NHS Foundation Trust & Newham University Hospital - April 2015**<br>**NHS Sickle Cell & Thalassaemia Screening Programme *Antenatal Screening Policy***<br><br>**http://www.sct.screening.nhs.uk/policy-antenatal** | www.gov.uk/phe/screening |
| 8-16 | footnote | **Antenatal Haemoglobinopathy Screening & Follow-Up Policy**<br>**East London NHS Foundation Trust & Newham University Hospital - April 2015**<br>**NHS Sickle Cell & Thalassaemia Screening Programme *Antenatal Screening Policy***<br><br>**http://www.sct.screening.nhs.uk/policy-antenatal** | www.gov.uk/phe/screening |
| 17 | | All incidents related to the antenatal screening pathway should be reported the NHS Sickle Cell & Thalassaemia Screening Programme Centre by email to PHE.screeninghelpdesk@nhs.net. | |
| 19 | | Laboratory (and counselling) support service | removed |

## 1 Executive Summary

The East London NHS Foundation Trust - Newham Community Services Sickle Cell and Thalassaemia Service, has a commitment, in conjunction with Newham University Hospital, to screen during the antenatal period for sickle cell, thalassaemia and other haemoglobin variants and follow up positive results for the Newham population. This is a very diverse community at high risk of inheriting haemoglobin conditions.

The aim of this policy document is to address the local screening proceedures and pathways for clients affected by these haemoglobin carrier states and conditions, and should be read in conjunction with the NHS Englang national standards and guidelines for the national antenatal sickle cell and thalassaemia screening programme.

### Changes to National Screening Program:

*The NHS Screening Program is now commissioned and hosted by NHS Public Health England (PHE). The NHS Sickle Cell and Thalassaemia (SCT) Screening Programme replace NHS Sickle Cell and Thalassaemia Screening Programme Standards October 2011 and have an implementation date of April 2017. There has been National Standard update from the 1st of February 2017. We have reviewed this guideline taking into consideration the National standard update .*

*The program has also moved please see contact details below:*

*PHE Screening, Floor 2 Zone B, Skipton House, 80 London Road, London SE1 6LH www.gov.uk/phe/screening Twitter: @PHE_Screening Blog: phescreening.blog.gov.uk*

*For queries relating to this document, please contact: phe.screeninghelpdesk@nhs.net*

## 2 Introduction

Sickle cell disease and thalassaemia major syndromes (alpha and beta) are genetically inherited blood disorders which mainly affect people whose ancestors are from Africa, Asia, the Middle East, the Mediterranean, and the Caribbean Islands, but is not exclusive to these populations.

People inherit these haemoglobin conditions in either a "carrier" or "diseased" state. Carriers have usually inherited a defective gene from one parent, and are considered "healthy carriers." It is important to identify carriers because the gene is passed from biological parent(s) to child. A couple who are both carriers of a significant haemoglobinopathy have a 1 in 4 or 25% chance, in every pregnancy, of having a child with a major haemoglobin condition.

An individual with a major condition has inherited a defective gene from both parents and generally has a chronic lifelong condition where significant health issues are anticipated. Regular treatment is required and there is limited chance of a cure. During pregnancy, prenatal diagnosis is offered, and the woman/couple may decide to accept termination of pregnancy for an affected fetus.

Individuals with sickle cell disease have a condition which expresses itself with bouts of severe pain; serious life-threatening infections; and varying degrees of chronic and acute anaemia. Medication to manage pain when it occurs, and prophylactically to reduce the risk of infections, is required throughout their lives. Individuals with thalassaemia major syndrome inherit a severe anaemia which requires them to have regular blood transfusions, 3-5 weekly, for survival.

It is also possible to inherit "benign" haemoglobin conditions which do not merit pre-natal diagnosis and termination of pregnancy is not offered. These conditions may cause intermittent health issues, but life-long treatment is not required. Such conditions include Hb DD, Hb EE, Hb CC,
and Hb H Disease.

Screening during the antenatal period, using a simple blood test, offers an opportunity to identify women who are haemoglobinopathy carriers, and subsequently allows screening of consenting biological fathers. Identified carrier women and couples "*at risk*" of having a baby with a major haemoglobin disorder, are able to receive information, advice and counselling in order to make choices for the pregnancy, including the decision to have further investigations if desired. The team at the Newham Sickle Cell and Thalassaemia Service work closely with the maternity department at Newham University Hospital, the laboratory services at Barts Healthcare, and liaise with primary care and other stakeholders, in order to provide quality care for this client group.

This document outlines the local processes for referral, appointments, and follow up of women who are carriers, as well as for identified "*at risk*" couples, to ensure that timely reproductive choices can be made for the pregnancy.

The local policy falls within the guidance issued by the Public Health England (PHE) NHS England Screening Programme Sickle Cell and Thalassaemia 2017 .[i]

[i] PHE - Sickle Cell & Thalassaemia Screening Programme. www.gov.uk/phe/screening

## 3 Aims & Objectives

- To offer counselling and follow up to support antenatal screening for sickle cell, thalassaemia and other haemoglobin variants, of all women booked within the maternity service at Newham University Hospital, Barking Birthing Centre, via the GP service or those who self-refer.
- To allow informed reproductive choice by identifying couples "*at risk*" of having affected infants at an early stage in pregnancy.
- Services include counselling, testing the baby's father, referral for prenatal diagnosis and termination of affected pregnancy, as well as support for women/couples who choose to continue a pregnancy where the baby has been diagnosed as having a major haemoglobin disorder.

## 4 Screening Criteria

- The GP, Midwife or Obstetrician offer screening for sickle cell, thalassaemia and other haemoglobin variants, via a blood test, when a woman attends to report a possible pregnancy.
- All women screened for haemoglobinopathy and identified as a carrier, or with a haemoglobin condition, or with an inconclusive result, are referred to the Sickle Cell and Thalassaemia team via Barts Healthcare Laboratory.
- The screening follow up service is available to all women booked within the maternity department at Newham University Hospital, Barking Birthing Centre or via the local GP service. Known "*at risk*" couples may also self-refer to the Newham Sickle Cell & Thalassaemia Centre.

## 5 Referral Criteria

- All women with confirmed carrier results;
- All fathers-to-be,where antenatal screening show the mother to be a carrier
- All women with inconclusive haemoglobinopathy screening results;
- *"At risk"* couples known to the sickle cell and thalassaemia service who self-refer.

## 6 Clients that do not meet the criteria

- Normal results – HbAA
- Women who are **23** weeks pregnant or over, can still be seen within the context of the antenatal genetic counselling service and offered screening for the baby's father, **BUT** prenatal diagnosis should not be offered, or referral made as the national "cut off" for termination of pregnancy is 24 weeks gestation.

## 7 National Screening Policy[ii]

The national screening programme has outlined the maternal conditions that must be detected during antenatal screening, and when identified require testing of the baby's father.[iii]

1. **Significant maternal haemoglobinopathies**
   - Hb SS
   - Hb SC
   - Hb $SD^{Punjab}$
   - Hb SE
   - Hb $SO^{Arab}$
   - Hb S/Lepore
   - Hb S/β thalassaemia
   - Hb S/δβ thalassaemia
   - β thalassaemia major/intermedia
   - Hb Lepore/β thalassaemia
   - Hb E/β thalassaemia
   - Hb H Disease (--/-α)

2. **Carrier states in mother**
   - Hb AS
   - Hb AC
   - Hb $AD^{Punjab}$
   - Hb AE
   - Hb $O^{Arab}$
   - Hb A Lepore
   - β thalassaemia carrier
   - δβ thalassaemia
   - $\alpha^0$ thalassaemia carrier (--/αα)
   - Hereditary persistence of fetal haemoglobin (HPFH)

3. **Any compound heterozygous state including one or more of the above carrier states.**

4. **Any homozygous state of the above carrier conditions.**

[ii] NHS Sickle Cell & Thalassaemia Screening Programme *Antenatal Screening Policy* http://www.sct.screening.nhs.uk/policy-antenatal 2011

[iii] NHS Sickle Cell & Thalassaemia Programme *Laboratory Handbook 3rd Edition October 2012* http://www.sct.screening.nhs.uk/policy-antenatal [Accessed February 2015]

## 8 Screening & Referrals to Newham Sickle Cell & Thalassaemia Service

All women booking for antenatal care should be offered a screening test for sickle cell, thalassaemia and other haemoglobin variants by the GP or midwife. This sample should be accompanied by completion of a Family Origin Questionnaire Form (FOQ)[Appendix 3] which assists the laboratory with processing results, and the specialist nurse with giving relevant information.

National policy[1] supported by NICE Guidelines[2] and NICE Quality Standards[3] state that women should be offered screening by 10 weeks of pregnancy in order to access timely follow up care.

Samples are sent to Barts Healthcare Laboratory from Newham University Hospital for processing in relation to haemoglobinopathies.

Newham Sickle Cell & Thalassaemia Service see women for counselling and offer screening of the baby's father if they have booked for antenatal care at

- Newham University Hospital;
- Barking birthing centre;
- via their local GP service;
- Or on occasion women/couples who self-refer to the service.

All carrier (trait), affected and inconclusive results are sent to the Newham Sickle Cell & Thalassaemia Service by email (via nhs.net) on a daily basis, for follow up.

Women who are **23** weeks pregnant or over, can still be seen within the context of the antenatal genetic counselling service and offered screening for the baby's father, **BUT** prenatal diagnosis is not be offered, or referral made, as the national "cut off" for termination of pregnancy is 24 weeks gestation.

Women who are Hb AA are **not** seen by the Sickle Cell & Thalassaemia Service. These results should be given to the woman by the midwife or GP.

## 9 Counselling Appointment

**Summary**

A comprehensive assessment of the woman using the RiO IT system needs to be carried out. This should include an assessment of the woman's physical and mental well being, ability to make informed decisions, as well as a review of her housing situation and any other relevant social issues. Information about the baby's father should also be included.

Women/couples are seen for antenatal genetic counselling by the Newham Sickle Cell & Thalassaemia Service between 8 weeks of pregnancy to term, and can be referred for prenatal diagnosis (PND) between 11-22 weeks of pregnancy. 24 weeks gestation is the national "cut off" for termination of pregnancy.

***If the baby's father is not available for screening, then appropriate information and advice should be given to the woman in regards to having prenatal diagnosis (PND) and a referral should be made if the woman chooses to do so.***

All information given and discussions should be documented:

- On the woman's RiO assessment which should be created prior to the counselling session;
- On the woman's/man's RiO progress notes;
- In the woman's handheld antenatal records.

The RiO assessment form should be updated:

- With the father's results when available;
- If the woman/couple choose to have PND, including the outcome.

**Process**

- Antenatal haemoglobinopathy results are received from Barts and the London Hospital Laboratory by the designated administrative staff at the Centre on a daily basis.

- All results should be reviewed within 48 hours of receipt, and the relevant process followed.

- The administrative team at the Newham Sickle Cell & Thalassaemia Service should check **all results** to determine whether the woman has been screened in a previous pregnancy.

- If the woman has been **screened in a previous pregnancy** then the following process should be followed
    - Has the baby's father from the previous pregnancy been screened? Are the paternal records available to the team at the Sickle Cell & Thalassaemia Service?
    - The administrative team should contact the woman by telephone to establish whether or not the woman is aware of her previous screening result? Is the baby's father the same as for her previous pregnancy?
    - If the baby's father is the same, will a counselling appointment be required?
    - If the baby's father is different then an appointment for counselling and partner screening is required.
    - The decisions made by the administrative team regarding women screened in a previous pregnancy should be reviewed by the specialist nurse responsible for the relevant counselling clinic.
    - The woman's RiO records should be updated to ensure that she is included in the screening cohort for statistical purposes.

- If there is no record that the woman has been screened in a previous pregnancy or she has not previously attended the Newham Sickle Cell & Thalassaemia Centre for counselling, the following process should be followed, within 48 hours of receiving the result.

    - The administrative team will contact the woman by telephone and inform her that her results have been received and she is being offered a counselling and baby's father screening appointment. ***Results should not be disclosed over the telephone.***

    - An appointment letter,[iv] haemoglobinopathy card, appropriate carrier leaflet[v,vi] and leaflet for the baby's father to explain the need for counselling and reason for needing a blood test[vii] should be sent to the woman, along with confirmation of her result and the date and time of the counselling session, by 1st class post.

[iv] The national programme centre has developed a generic letter that could be adapted for local use to invite women for counselling http://www.sct.screening.nhs.uk/guidance - (Appendix 5)

[v] NHS Sickle Cell & Thalsseamia Screening Programme Carrier Leaflets http://sct.screening.nhs.uk/professional-leaflets

[vi] Newham Sickle Cell & Thalassaemia Service Carrier Leaflets

[vii] Tests for Dads Leaflet & letter for men to encourage screening during antenatal period http://www.sct.screening.nhs.uk/guidance - http://sct.screening.nhs.uk/professional-leaflets - (Appendix 6)

- All letters and haemoglobinopathy cards should be reviewed and signed by the specialist nurse conducting the genetic counselling clinic before dispatch (or an alternative specialist nurse colleague if the named specialist nurse is unavailable).

- Complex results should be discussed with the named senior specialist nurse or haematology consultant (if the senior specialist nurse is unavailable) **prior** to the woman's/couples appointment.

- If the woman's result is inconclusive and DNA has been performed on the sample, then the administrative team should ensure that the result is available prior to counselling.

- Antenatal Genetic Counselling Clinics are held at least 2 days weekly at the Sickle Cell & Thalassaemia Centre.

- The admininstrative team should check whether or not an interpreter is required for the counselling session and book as necessary.

## 10 Antenatal Counselling - Confirmed Carrier Results

If the woman attends for counselling with the baby's father then consent for screening is obtained from him, prior to the counselling session.
The woman/couple are seen by a trained haemoglobinopathy specialist nurse and the following information should be obtained or included during the counselling session:

- The woman's result and it's significance to her.
- Any prior knowledge of haemoglobinopathies?
- Any family history of haemoglobinopathies?
- Relevant antenatal history (if not already known); alternately check any details that have already been recorded to ensure that they are correct.
- Obtain some background information on the baby's father.
- Significance of the woman's result for the baby.
- The need for screening the baby's father.
- The potential condition (s) that the baby may be at risk of inheriting (use the maternal results and the family origins of both parents to identify the conditions which the baby is **most likely** to inherit).
- Genetic inheritance pattern.
- Pathophysiology of the condition that the baby may be at risk of inheriting.
- Offer screening to the baby's father.
- Discuss the possible outcomes of the screening test.
- Discuss the process following screening, and reporting results to the couple.

If the baby's father has not yet received a screening information leaflet then one should be given to him during the counselling session.[v ii]

The blood request form sent with the paternal sample should include the man's demographic information as well as his family origins. The antenatal woman's details (NHS number, name, surname and date of birth), should also be included on the form to ensure that maternal and paternal results are linked by the Laboratory. The source of the sample should be written on the form so that the results are received back at the Sickle Cell & Thalassaemia Centre.

A letter summarising the counselling session should be sent to the GP and antenatal screening midwife within 24 hours of the counselling sesson.

## 11 Special Notes

***If the baby's father is unavailable or declines haemoglobinopathy screening then the woman can be referred for prenatal diagnosis without a paternal haemoglobinopathy screening result, in the same way as "at risk" couples. The test is done at University College London Hospital (UCLH) and results are returned to the Newham Sickle Cell & Thlassaemia Service.***

***If either the woman or the baby's father is a beta thalassaemia carrier then the woman/couple should be informed that if their baby is also a beta thalassaemia carrier then this will not be identified during newborn bloodspot screening and that arrangements should be made for their baby to be screened at 1 year of age or older.***

## 12 Follow up after paternal screening

All "partner" results should be reviewed by the specialist nurse who conducted the genetic counselling clinic and linked to the woman's results. "Partner" results should be made available to the Sickle Cell & Thalassaemia Service within 5 working days from the time of taking the blood sample. The admintrative team are responsible for chasing up progress and receipt of the partner results.

If there are any problems with obtaining results or they are not available within the recommended timescale, this should be reported to the nursing team lead who should escalate with the laboratory team within 48 hours. If the results are not received, this should be reported on Datix and the laboratory team notified directly.

## 13 Normal Results (Hb AA)

If the paternal results are normal then this should be conveyed to the couple by letter, accompanied by a haemoglobinopathy card for the man. If specifically requested, the results may in addition be conveyed by a telephone call. The couple should then be discharged from the Sickle Cell & Thalssaemia Service as no further appointments will be required.

## 14 Paternal Carrier Results (baby not "at risk" of inheriting a major haemoglobin disorder)

If the man is identified as a haemoglobinopathy carrier then the couple should be invited for a follow up counselling session and the results explained "face to face".

If they are not "*at risk"* of having a baby with a major haemoglobin disorder (sickle cell disease or thalassaemia major syndrome), but the baby could inherit a haemoglobin disorder which does not require long-term treatment, for example a condition such as Hb EE; Hb CC; Hb DD; Hb C/Beta thalassaemia, Hb H Disease, this should be explained and the couple reassured. A paternal haemoglobinopathy card and appropriate carrier leaflet should be given.

Prenatal diagnosis is **not** required for any of these conditions and referral will not be accepted by University College London Hospital.

## 15 Antenatal Counselling – Couples "at risk" of having a baby with a major haemoglobin disorder

If the paternal result shows that the couple are "*at risk"* of having a baby with a major haemoglobin disorder (sickle cell disease, thalassaemia major syndrome) then the couple should be:

- Offered an urgent follow up counselling appointment to explain the risk to their baby, details about the condition that their baby could inherit and the options for the pregnancy. An explanatory leaflet should be sent with the appointment or given after attendance at the counselling session.

- Informed of the risk of miscarriage associated with having prenatal diagnosis whether or not the baby is affected by a major haemoglobin disorder.

- Offered an appointment for prenatal diagnosis.

- Urgently referred to University College London Hospital (UCLH) if they do decide to have prenatal diagnosis. An appointment will be sent directly to the woman/couple.[viii]

Prior to faxing the referral to UCLH, the specialist nurse at Newham Sickle Cell & Thalassaemia Service should liaise with the maternity department /antenatal screening coordinator to ensure that

- the woman/couple do not require PND for any other condition, such as Down's Syndrome.
- all the woman's results such as Hepatitis B & C; full blood count; HIV status, blood group and rhesus factor are available for inclusion on the referral form.

If prenatal diagnosis is declined then the couple should be informed of the process for screening their baby after birth and how they can/will obtain the result, and then discharged from the service to await baby's results.

The Newborn Screening Laboratory at Central Middlesex Hospital should be informed of all "at risk" couples that continue the pregnancy whether or not they have had PND.[ix]

## 16 Following Prenatal Diagnosis (PND)

Following PND, the initial result is given to the woman/couple by the team at UCLH. On receipt of the result at Newham Sickle Cell & Thalassaemia Service, the specialist nurse should contact the woman to ensure that she has a complete understanding of the result and what this means for her baby, and discuss the options for her pregnancy.

- If the baby **does not** have a major haemoglobin disorder (is a haemoglobinopathy carrier or Hb AA), then no further appointments or follow up is required during pregnancy. A record of the woman/couple results and the baby's due date should be

[viii] Referral Form University College London Hospital should be used
[ix] *West Midlands "At Risk Pregnancy" Alert Form* - Appendix 10

kept so that the newborn screening result is confirmed and conveyed to the couple as soon as it is available.

- If the couple have PND and the baby is affected, and they **choose to** have a termination of pregnancy, then the specialist nurse needs to inform the GP so that he can facilitate a referral for this procedure to be carried out. Ongoing counselling support should be offered by the specialist nurse.

- If the couple have PND, the baby is affected and they **choose not to** have a termination of pregnancy then the Newham Sickle Cell & Thalassaemia Service will offer ongoing support to the woman/couple. Some parents may wish to speak to the Paediatrician or a family with a child who has the same condition. The parents should be informed of the newborn screening result as soon as possible after receipt by the Sickle Cell & Thlassaemia Centre.

All outcomes from PND should be documented on the woman's RiO assessment form and progress notes. A copy of the results from UCLH should also be uploaded and attached to her records.

The Newborn Screening Laboratory at Central Middlesex Hospital should be informed of all "at risk" couples with an affected baby that continue the pregnancy.[ix]

**Special Screening Situations**

## 17 Women who book late for pregnancy

Women who are **23** weeks pregnant or over, can still be seen within the context of the antenatal genetic counselling service and offered screening for the baby's father. **BUT** prenatal diagnosis should not be offered or referral made as the national "cut off" for termination of pregnancy is 24 weeks gestation.

If they do not attend the first appointment offered, then any subsequent follow up should be in the general carrier counselling clinic.

## 18 Woman with a major haemoglobin disorder[x;xi]

Women who have a major haemoglobin disorder and are pregnant are considered "high risk" and should always be booked for joint obstetric/haematologist care. The specialist nurse should ensure that joint care by Obstetrician & Haematologist is initiated.

The woman should be offered an appointment for counselling regarding care of her condition during pregnancy, as well as for genetic counselling and screening of the baby's father.

## 19 Woman with inconclusive results

If the woman has an "inconclusive" haemoglobinopathy result this should be discussed with the named senior specialist nurse. If she is unavailable, then contact should be made with the consultant haematologist *prior* to the counselling appointment. The administrative staff should chase up the final report and inform the specialist nurse when this is available.

A counselling appointment should be offered

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[x] Royal College of Obstetricians and Gynaecologists (2014) *Thalassaemia in Pregnancy, Management of Beta* (Green-top Guideline 66) https://www.rcog.org.uk/en/guidelines-research-services/guidelines/gtg66/

[xi] Royal College of Obstetricians and Gynaecologists (2011) *Sickle Cell Disease in Pregnancy, Management of* (Green-top Guideline 61) Reviewed December 2014 https://www.rcog.org.uk/en/guidelines-research-services/guidelines/gtg61/

- If the final result is unavailable prior to the counselling appointment, this should be explained to the woman and partner testing should proceed.
- If the final result is available then this should be explained to the woman, and partner testing offered where relevant.
- If any further investigations are required then blood should be taken as requested by the laboratory.
- If the "partner" result is Hb AA then there is no risk to the baby of inheriting a major haemoglobin disorder.
- If the "partner" also has a haemoglobinopathy, then this should be discussed with the senior specialist nurse and a risk assessment for the baby made, prior to any follow up appointment for the couple.

## 20 Women with suspected alpha plus thalassaemia or iron deficiency anaemia[xii][xiii]

The screening result for women who may be alpha plus thalassaemia carriers or have iron deficiency anaemia may be indistinguishable.

Women who are suspected alpha plus thalassaemia carriers are not at high risk of having a baby with a major haemoglobin disorder, and the recommendation from the National Programme Centre is that there is no requirement for re-screening, follow up or partner screening in relation to haemoglobinopathy risk.

In East London NHS Foundation Trust, particularly given the family origins of the local population groups, these inconclusive results should be referred to the GP, in writing, for investigations to exclude iron deficiency anaemia. All further follow up is via the GP.

## 21 Woman with suspected alpha zero thalassaemia[xiii]

The Newham Sickle Cell & Thalassaemia Service have been given the responsibility to follow up women who are ***suspected alpha zero thalassaemia carriers*** as the laboratory at Barts Healthcare do not have access to information regarding family origins. Women should be routinely offered a counselling appointment.

The local laboratory policy requests partner testing and a blood sample from the woman for the following investigations[xii]

- Full blood count,
- Ferritin levels,
- Hematenics,
- DNA testing to confirm the result.

If both couples are at RISK

This differs from the national policy and high prevalence screening algorithm.[Appendix 4] "Partner" testing should be offered if the man has family origins from China (including Hong Kong), Taiwan, Thailand, Cambodia, Laos, Vietnam, Indonesia, Burma, Malaysia, Singapore, Philippines, Cyprus, Greece, Sardinia, Turkey, or uncertain family origins.

## 22 Women who have had fertility treatment

If a woman has had fertility treatment then it is important to establish the source of both egg and sperm to assess the potential risk of the baby inheriting a haemoglobin disorder.

[xii] NHS Sickle Cell & Thalassaemia Screening Programme (2012) *Laboratory Handbook* http://www.sct.screening.nhs.uk
[xiii] High Prevalence Antenatal Screening Algorithm - Appendix 4

If both egg and sperm are from the baby's biological parents then the risk to the baby can be assessed as for any other carrier woman/couple.

If the sperm or the egg has been donated to the couple then it is not possible to do a risk assessment of the pregnancy based on the parental screening results. If the donor sperm and/or egg have been screened for a haemoglobinopathy and the results are available then this can be discussed with the couple.

If no screening results are available for either donor sperm or egg then the process for dealing with this situation should be locally determined with discussions between maternity services, specialist nurses and consultant haematologist.

## 23 Women who have had a bone marrow transplant

Women who have had a bone marrow transplant will not show their own haemoglobin genotype on routine testing. DNA will be required to confirm their haemoglobin genotype. In the interim, until the woman's result is known, testing the baby's father should proceed. Follow up will be based on both parental results

## 24 Woman who does not attend counselling appointment (DNA)[xiv]

The woman should be routinely contacted by telephone, prior to the first appointment being booked, to agree the date and time of the counselling session. A letter should be sent via first class post to confirm this appointment. If she does not attend the appointment then her GP and screening midwife should be informed by letter.

A subsequent appointment should be booked by letter, which should include information about the fact that she will be discharged from the service if she fails to attend.

If the woman does not attend the second appointment then she is discharged from the service. Failure to attend should be documented on RiO system and GP & maternity services should be informed of non-attendance with the option to re-refer client as necessary.

## 25 Woman who cancels counselling appointment

If the woman cancels her counselling appointment the administrative team should check

- The reason for cancellation (for example, previous counselling? Personal reasons?).
- The woman's earliest convenience for re-booking her appointment, and reschedule the counselling session.
- If she does not attend the rescheduled appointment then follow the process as for a woman who DNA their appointment.

## 26 Audit & Monitoring - Data collection

Data is collected at three levels to support the antenatal sickle cell and thalassaemia screening programme.

## 27 National Screening Committee (Quarterly Key Performance Indicators)[xv]

[xiv] East London NHS Foundation Trust (2012) *Policy for Service Users Who Fail to Attend Appointments in Children, Young People, Sexual and Reproductive Health Services and CAMHS*

Key Performance Indicator Data (KPI) is collected quarterly (June, September, December and March) for the National Screening Committee. The Sickle Cell & Thalassaemia Screening Programme requires a report on three areas of activity related to antenatal screening

- **ST1** - Screening coverage and uptake.
  **Data source**: Maternity Services.

- **ST2** - Timely offer of screening and availability of results by 10 weeks gestation.
  **Data source**: Antenatal Screening Laboratory.

- **ST3** - Completion of Family Origin Questionnaire (FOQ) Appendix 3
  **Data Source**: Antenatal Screening Laboratory.

## 28 NHS Sickle Cell & Thalassaemia Screening Programme Annual Performance Appendix 8/9,[xvi]

Antenatal Screening Data is collected by the National Sickle Cell & Thalassaemia Screening Programme for the financial year, April - end of March. Appendix 8 An Annual Data Report is collated and the information is published on the Programme Centre website.

Prenatal Diagnosis Data is collected by the National Sickle Cell & Thalassaemia Screening Programme and the data is published in the Annual Data Report. Appendix 9

## 29 East London NHS Foundation Trust & Newham Sickle Cell & Thalassaemia Centre

Monthly Activity Data is collated from the RiO IT System on activity at the Newham Sickle Cell & Thalassaemia Service.

## 30 Incident Reporting[xvii]

All incidents related to the antenatal screening pathway should be reported the NHS Sickle Cell & Thalassaemia Screening Programme Centre by email to PHE.screeninghelpdesk@nhs.net.

Incidents should also be reported using the national incident form to National Patient Safety Agency.[xviii]

Incidents should also be reviewed locally by using the Trust agreed pathway for investigation and reporting.

The purpose of incident reporting is to

- Review all incidents and apply the lessons learned;
- Use the incidents to inform an improvement in services;
- Use the incidents to support national and local policy development;
- Use the incidents to support development of national and local training resources.

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[xv] UK National Screening Committee (2014) *Key Performance Indicators for Screening 2014/15 Version 1.1.14*

[xvi] NHS Sickle Cell & Thalassaemia Screening Programme (2015) *Annual Data Report 2013-14* www.sct.screening.nhs.uk

[xvii] Healthcare staff incident reporting http://www.nrls.npsa.nhs.uk/report-a-patient-safety-incident/healthcare-staff-reporting/

[xviii] Healthcare staff incident reporting form https://www.eforms.nrls.nhs.uk/staffreport/

## Contacts

### Newham Sickle Cell & Thalassaemia Centre- for queries contact

19-21 High Street South
East Ham
E6 6EN
Tel : 0208 821 0800 Fax 0208 821 0800
Sending referrals & Results - Email team on : elt-tr.sickleandthal@nhs.net
Specialsit lead Nurse Advise
Sekayi Tangayi – email sekayi.tangayi@nhs.net
Tel 07984 320 2190

**Bart's Healthcare** Laboratory processes all antenatal haemoglobinopathy samples and send the positive results to the Newham Sickle Cell & Thalassaemia Centre.-

**Contact number** 0203 246 0342 **or** 0203 246 6116

**Contact persons** are:
Katiri Sophia: email address is Sophia.Katiri@bartshealth.nhs.uk.

**NB** there is no fax method for contacting Bart's Healthcare Laboratory, only email requests are accepted.

### Prenatal Diagnosis Centre

University College London Hospital (UCLH)
Haemoglobinopathies Genetics Centre
Ground Floor, 86-96 Chenies Mews
London
WC1E 6HX

**Contact person for bookings & referrals:**
Dimitri Theodotou email address is Dimitri.theodotou@ uchl.nhs.net
**Tel:** 020 3447 9458 **Fax:** 020 3487 9864
Direct only for PND specialist advise-
Call Dr Mary Petrou on 0780 885 8215 or Email : mary.petrou@uchl.nhs.uk

### Newham University Hospital

Glen Road
Plaistow
London
E13 8SL
**Local Antenatal Screening Co-ordinator:**
**Maternity Booking Tel:** 020 7363 8433
For quieries or failsafe checklists send Email to - munewham@nhs.net

**Old Blood Results request – Newham Laboratory**
Tel: 020 7363 8732

### Barts & the London Consultant Haematologists

Dr Olivia Kreze-07825759155-
Secretary :
Tel: 0207 363 9413 fax :0207 363 8098

Dr Paul Telfer -07906311482 -
Secretary : Maureen Finneran email: Maureen Finneran@bartshealth.nhs.uk

Tel 0203 246 0352 fax 0203 246 0351

Dr Banu Kaya-07905265005
Secretary : Maureen Smith email: Maureen smith@bartshealth.nhs.uk
Tel : 0203 246 0338 fax:0203 246 0351

**NHS Sickle Cell & Thalassaemia Screening Programme**
2nd Floor
Skipton House,
80 London Road,
London,
SE1 6LH

**Programme Manager:** Cathy Coppinger
**Email:** PHE.screeninghelpdesk@nhs.net

# Antenatal Care Pathway - Haemoglobinopathies

Appendix 2

**High Prevalence**

Offer screening to all women for sickle cell, thalassaemia & other haemoglobin variants (irrespective of family origin)

Women screened by GP, Newham University Hospital, or Barking Birthing Centre or women may self refer to service. Sample processed at Barts & the London and results sent to Newham Sickle Cell & Thalassaemia Service

**Process**

**Screen at 8-10 weeks**

**Normal Result**

Woman Informed by maternity services

*Confirmation of result in writing*

**Inconclusive Result**

Inform woman. Assess risk, Counselling, **Offer partner screening (if appropriate)** (DNA testing if ***couple*** possible alpha zero)

**Carrier result**

Inform woman with confirmation in writing, HbO card & carrier information. Counselling appointment.

**Offer partner screening**

**Affected**

**Sickle Cell Disorder**

High Risk pregnancy – Haematology & Obstetric care essential. Counselling.

**Offer Partner Screening**

**Attended Appointment**

Follow up as appropriate

**DNA 1st Appointment**

Resend appointment x 1

**Inform GP & maternity service of attendance or DNA**

**Partner available for screening**

**Partner unavailable for screening**

**Partner Normal result**

Inform woman and partner in writing and send HbO card

**Partner Carrier result**

Give partner result with confirmation in writing, leaflet & HbO card

**Refer "At risk" couple**

Specialist counselling for woman or "at risk" couple by a trained individual

**Offer Diagnostic Testing (PND)**

**Decline diagnostic testing**

Inform Newborn Screening Laboratory at CMH

**Accept diagnostic testing**

CVS - ideal (11 weeks gestation)

Amniocentesis

Cordocentesis

Carrier or "normal for gestation" result [Inform Newborn Screening Laboratory]

Baby affected by a major haemoglobin disorder (sickle cell disease or thalassaemia major)

**Continue Pregnancy**

Newborn Bloodspot Screening

**Discontinue Pregnancy**

Offer counselling & follow up support by trained individual

**To be completed by 12+6 weeks of pregnancy**

# Family Origin Questionnaire

NHS Screening Programmes

Sickle Cell & Thalassaemia

If using a pre-printed label please attach one to each copy

Hospital Name .....
Hospital No .....
NHS No .....
Estimated Delivery Date .....
Surname .....
Forename .....
Date of Birth .....
Add1 .....
Add2 .....
Post Code .....

Screening test declined ☐

Do you want to give a reason why declined?

Yes .....

No ☐

REPORT DESTINATION *(eg Community Midwife, GP, Antenatal Clinic, Obstetrician)* .....

## What are your family origins?

*Please tick all boxes in ALL sections that apply to the woman and the baby's father*

| | Woman | Baby's father |
|---|---|---|
| **A. AFRICAN OR AFRICAN-CARIBBEAN** (BLACK) | | |
| Caribbean Islands | ☐ | ☐ |
| Africa (excluding North Africa) | ☐ | ☐ |
| Any other African or African-Caribbean family origins (please write in…) | | |
| **B. SOUTH ASIAN** (ASIAN) | Woman | Baby's father |
| India or African-Indian | ☐ | ☐ |
| Pakistan, Bangladesh | ☐ | ☐ |
| Sri Lanka | ☐ | ☐ |
| **C. SOUTH EAST ASIAN** (ASIAN) | Woman | Baby's father |
| China including Hong Kong, Taiwan, Singapore | ☐ # | ☐ # |
| Thailand, Indonesia, Burma | ☐ # | ☐ # |
| Malaysia, Vietnam, Philippines, Cambodia, Laos | ☐ # | ☐ # |
| Any other Asian family origins (please write in…) (e.g. Caribbean-Asian) | | |
| **D. OTHER NON-EUROPEAN** (OTHER) | Woman | Baby's father |
| North Africa, South America etc | ☐ | ☐ |
| Middle East (Saudi Arabia, Iran etc) | ☐ | ☐ |
| Any other Non-European family origins (please write in…) | | |
| **E. SOUTHERN & OTHER EUROPEAN** (WHITE) | Woman | Baby's father |
| Sardinia | ☐ # | ☐ # |
| Greece, Turkey, Cyprus | ☐ # | ☐ # |
| Italy, Portugal, Spain | ☐ | ☐ |
| Any other Mediterranean country | ☐ | ☐ |
| Albania, Czech Republic, Poland, Romania, Russia etc | ☐ | ☐ |
| **F.* UNITED KINGDOM** (WHITE) *refer to chart at the back* | Woman | Baby's father |
| England, Scotland, N Ireland, Wales | ☐ | ☐ |
| **G.* NORTHERN EUROPEAN** (WHITE) *refer to chart at the back* | Woman | Baby's father |
| Austria, Belgium, Ireland, France, Germany, Netherlands | ☐ | ☐ |
| Scandinavia, Switzerland etc | ☐ | ☐ |
| Any other European family origins, *refer to chart* (please write in) (e.g. Australia, N America, S Africa) | | |
| * Hb Variant Screening Requested by (F) and/ or (G) | ☐ | ☐ |

# Higher risk for alpha zero thalassaemia

| | Woman | Baby's father |
|---|---|---|
| **H. DON'T KNOW** adoption/unknown ancestry | ☐ | ☐ |
| donor egg/sperm | ☐ | ☐ |
| bone marrow transplant | ☐ | ☐ |
| **I. DECLINED TO ANSWER** | ☐ | ☐ |
| **J. ESTIMATED DELIVERY DATE** (please write in if not above) | | |
| **K. GESTATION AT TIME OF TEST** | | |

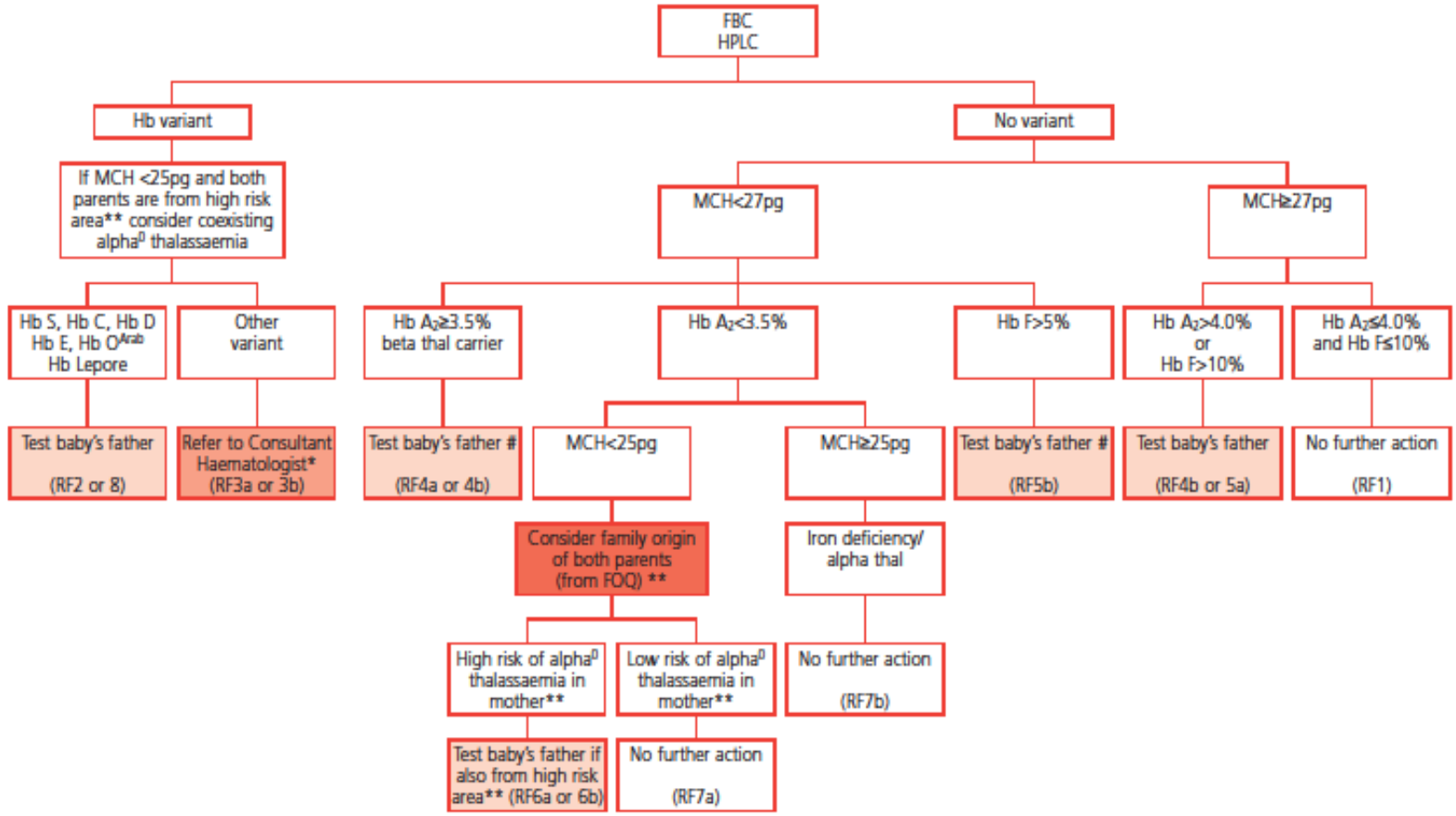
All women need to be informed that routine analysis of blood may identify them as a thalassaemia carrier. In **low prevalence** areas OFFER haemoglobin variant screening to all women if they or the baby's father have answers in any yellow box. In **high prevalence** areas OFFER haemoglobin variant screening to all women irrespective of answers, ie. if they or the baby's father have answers in white and yellow boxes A - I.

Signed _____ Print Name _____ Job Title _____ Date _____

(By Health Care Professional Completing the Form)

The TOP (white) copy of this form must be attached securely to the laboratory antenatal booking request form and sent to the laboratory with the antenatal blood samples, the second (pink) copy is to be retained in the patient's maternity notes, third (yellow) copy to go into hospital notes or where appropriate. The completion of this form is an ESSENTIAL part of the screening programme for sickle cell & thalassaemia.

# Testing Algorithm for High Prevalence Areas




* Refer analytical results to consultant for an opinion on the need for a clinical referral or consult the laboratory support service helpline.

** Consider at high risk if any ethnic origins in China (including Hong Kong), Taiwan, Thailand, Cambodia, Laos, Vietnam, Indonesia, Burma, Malaysia, Singapore, Philippines, Cyprus, Greece, Sardinia, Turkey, or if ethnic/family origin is uncertain/unknown.

\# In all cases consider coexisting $\alpha^0$ thalassaemia if both parents are from a high risk area and MCH <25pg.

Reconsider low risk couples if fetal anaemia/hydrops seen on ultrasound scanning or if family history of hydrops fetalis.

# Template letter for inviting women for antenatal genetic counselling

Dear (insert woman's name)

## Antenatal sickle cell and thalassaemia screening

**Your test result is:** (insert result)

The substance in your blood that carries oxygen around your body is called haemoglobin. You had a blood test recently to check whether you carry a gene for unusual haemoglobin. The test result shows that you are a (insert result) carrier. Being a carrier means that you are healthy. You do not have a condition that requires any treatment and will never develop one.

Enclosed is a leaflet explaining about being a (insert result) carrier.

This result could have implications for your baby if the baby's biological father also carries a gene to make unusual haemoglobin. **For this reason we are inviting you and your baby's father to an appointment to discuss your test result. At that time, we will offer the baby's father a quick and simple screening blood test for unusual haemoglobin.**

Your joint appointment is on (insert date /time/place)

If either of you are unable to attend this appointment, please call (name and number) to let us know and to arrange another appointment.

We have enclosed a letter and a leaflet for your baby's father. Please could you pass this on to him? When considering his test, it is important to know:

- Your baby's father will not know if he is a carrier unless he has this specific blood test
- If both parents are carriers, there is a 1 in 4 (25%) chance that your baby could inherit a haemoglobin disorder
- There are many different types of haemoglobin disorders - some more serious than others. The most serious conditions are sickle cell disease and thalassaemia major

If you have any questions please do not hesitate to contact us.

Yours sincerely,

July 2013 V1.0

# Template letter for inviting the baby's father for antenatal genetic counselling and screening

Dear (insert father's name here)

**Your invitation to screening for sickle cell and thalassaemia**

Recent blood tests done antenatally have shown that the mother of your baby carries a gene for unusual haemoglobin. Haemoglobin is the substance in your blood that transports oxygen around your body.

**We are inviting your baby's mother *(or if possible insert her name)* to an appointment to discuss this result. We would also like to invite you to attend at the same time so that we can offer you a screening blood test. This quick and simple test will give you important information about whether you are a carrier and if this could affect the health of your unborn baby.**

**Your appointment is at:**
**Date, time, place**

If you cannot make this appointment and would like to arrange another time or if you would like to talk to us confidentially please call (HCP name and number)

Your blood test will show if you are also a carrier for unusual haemoglobin. If both parents are carriers, there is a 1 in 4 (25%) chance that your baby could inherit a haemoglobin disorder. Some of these disorders are serious – and include conditions such as sickle cell disease and thalassaemia major.

You can decide whether or not to have this blood test but we strongly recommend that you read the enclosed leaflet, *Tests for dads*. This explains about the test and why it is important for your baby.

When thinking about the blood test, it is important to note:

- If you are a carrier, you are usually well. You will not know your carrier status unless you have had a specific blood test
- Your baby can only inherit a haemoglobin disorder if both you and your baby's mother are carriers
- It is a simple blood test lasting just a few minutes. Your results are confidential to you and your baby's mother. They will only be used for the health care of you and your baby and will not be passed to any other organisation

Yours sincerely

July 2013 V1.0

# At Risk Couples

NHS
Screening Programmes

## Inheritance risk table

| Father \ Mother | Hb S | β thal | δβ thal | Hb Lepore | Hb E | Hb $O^{Arab}$ | Hb C | Hb $D^{Punjab}$ | HPFH | Not identified as a carrier |
|---|---|---|---|---|---|---|---|---|---|---|
| Hb S | ■ | ■ | ▒ | ▒ | ▒ | ■ | ■ | ■ | ▒ | |
| β (beta) thal | ■ | ■ | ■ | ■ | ■ | ▒ | | | | |
| δβ (delta beta) thal | ▒ | ■ | ▒ | ■ | ▒ | ▒ | | | | |
| Hb Lepore | ▒ | ■ | ■ | ■ | ▒ | ▒ | | | | |
| Hb E | ▒ | ■ | ▒ | ▒ | | | | | | |
| Hb $O^{Arab}$ | ■ | ▒ | ▒ | ▒ | | | | | | |
| Hb C | ■ | | | | | | | | | |
| Hb $D^{Punjab}$ | ■ | | | | | | | | | |
| HPFH | ▒ | | | | | | | | | |
| Not identified as a carrier | | | | | | | | | | |

Table of parental carrier state combinations that give rise to the risk of a baby with significant sickle cell disease or beta thalassaemia major

**Key:**

- ■ Serious risk - refer couple for counselling - prenatal diagnosis to be offered
- ▒ Less serious risk - refer couple for counselling - further investigation may be required
- ☐ Minimal risk

(Table based on work of Prof. B. Modell)

# National Data Collection

## Data for Antenatal Sickle Cell and Thalassaemia Screening Programme

**High Prevalence Laboratories**

**Annual collection - 1 April 2013 to 31 March 2014**

NHS Screening Programmes
Sickle Cell & Thalassaemia

| Laboratory details | | | | |
|---|---|---|---|---|
| **Trust name:** | | | | |
| **Laboratory name** (Dept. and Hospital): | | | | |
| **Maternity Unit:** | | | | |

| Laboratory contact details | | | | |
|---|---|---|---|---|
| **Contact person:** | *Name:* | | | |
| | *Telephone:* | | *Email:* | |
| **Haematologist responsible (incl. tel. no.):** | | | | |
| **Lab lead for SCT screening:** | *Name:* | | | |
| | *Telephone:* | | *Email:* | |

*Please state if this is the same person as specified under "Contact person"*

| Sample referrals | |
|---|---|
| **Samples sent away for initial analysis:** | |
| **Samples for initial analysis received:** | |

*If received, complete a separate form for all referring labs - See guidance notes*

| | |
|---|---|
| **Data collection period if different from 1st April 2013 - 31st March 2014:** | |

***Please click on the data items to view the appropriate guidance for each***

| | Data item | Value | Comments |
|---|---|---|---|
| HP1 | Booking bloods received | | |
| HP2 | Booking bloods tested by 10 wks (KPI ST2) | | |
| HP3 | Booking bloods with FOQ (KPI ST3) | | |
| HP4 | Not tested - declined | | |
| HP5 | Not tested - previous screen 'positive' | | |
| HP6 | Not tested - previous screen 'negative' | | |
| HP9 | MCH < 25pg | | |
| HP10 | High risk alpha0 | | |
| HP11 | Screen 'positive' | | |
| HP12 | Screen 'negative' | | |
| HP13 | Pending/Inconclusive results | | |
| HP14 | Father specimens requested/results previously known | | |
| HP15 | Father specimens available | | |
| HP16 | High risk couples (dark orange boxes in part 2) | | |

| | | |
|---|---|---|
| HP20 | Breakdown of all screen 'positive' women | Complete in part 2 |

| | | |
|---|---|---|
| M1 | **What is your first line screening method?** | |
| | *Select from list:* | |
| | *Other:* | |
| M2 | **Method used for confirmation onsite:** | |
| | *Click to highlight all applicable methods from the list:*<br>(You may highlight as many methods as needed) | Acid electrophoresis eg Cirate agar or Acid gel<br>Alkaline electrophoresis eg Cellulose acetate or alkaline gel<br>Capillary electrophoresis<br>DNA<br>IEF<br>Mass spectrometry<br>Sickle solublity test<br>Sent away<br>Other, please give details |
| | *Other:* | |
| M3 | **If you send confirmations away, where do they go?** | |

| | | |
|---|---|---|
| A2Ref | **What is your Hb A2 reference range?** | |
| A2Action Value | **When MCH <27pg, what is the lowest level of HbA2 at which you would consider a diagnosis of a beta thal carrier?** | |

Please fill in the yellow sections

Click here for more information on report formats

v 26/03/2014

## Data for Antenatal Sickle Cell and Thalassaemia Screening Programme

**High Prevalence Laboratories**

**Annual collection - 1 April 2013 to 31 March 2014**

NHS Screening Programmes
Sickle Cell & Thalassaemia

| Laboratory details | |
|---|---|
| **Trust name:** | |
| **Laboratory name** (Dept. and Hospital): | |
| **Maternity Unit:** | |

| Laboratory contact details | | | | |
|---|---|---|---|---|
| **Contact person:** | *Name:* | | | |
| | *Telephone:* | | *Email:* | |
| **Haematologist responsible (incl. tel. no.):** | | | | |
| **Lab lead for SCT screening:** | *Name:* | | | |
| | *Telephone:* | | *Email:* | |

*Please state if this is the same person as specified under "Contact person"*

| Sample referrals | |
|---|---|
| **Samples sent away for initial analysis:** | |
| **Samples for initial analysis received:** | |

*If received, complete a separate form for all referring labs - See guidance notes*

| | |
|---|---|
| **Data collection period if different from 1st April 2013 - 31st March 2014:** | |

***Please click on the data items to view the appropriate guidance for each***

| | Data item | Value | Comments |
|---|---|---|---|
| HP1 | Booking bloods received | | |
| HP2 | Booking bloods tested by 10 wks (KPI ST2) | | |
| HP3 | Booking bloods with FOQ (KPI ST3) | | |
| HP4 | Not tested - declined | | |
| HP5 | Not tested - previous screen 'positive' | | |
| HP6 | Not tested - previous screen 'negative' | | |
| HP9 | MCH < 25pg | | |
| HP10 | High risk alpha0 | | |
| HP11 | Screen 'positive' | | |
| HP12 | Screen 'negative' | | |
| HP13 | Pending/Inconclusive results | | |
| HP14 | Father specimens requested/results previously known | | |
| HP15 | Father specimens available | | |
| HP16 | High risk couples (dark orange boxes in part 2) | | |

| | | |
|---|---|---|
| HP20 | Breakdown of all screen 'positive' women | Complete in part 2 |

| | | |
|---|---|---|
| M1 | **What is your first line screening method?** | |
| | *Select from list:* | |
| | *Other:* | |
| M2 | **Method used for confirmation onsite:** | |
| | *Click to highlight all applicable methods from the list:*<br>(You may highlight as many methods as needed) | Acid electrophoresis eg Cirate agar or Acid gel<br>Alkaline electrophoresis eg Cellulose acetate or alkaline gel<br>Capillary electrophoresis<br>DNA<br>IEF<br>Mass spectrometry<br>Sickle solublity test<br>Sent away<br>Other, please give details |
| | *Other:* | |
| M3 | **If you send confirmations away, where do they go?** | |

| | | |
|---|---|---|
| A2Ref | **What is your Hb A2 reference range?** | |
| A2Action Value | **When MCH <27pg, what is the lowest level of HbA2 at which you would consider a diagnosis of a beta thal carrier?** | |

Please fill in the yellow sections

Click here for more information on report formats

v 26/03/2014

# National Data Collection

## Breakdown of risk status for all screen 'positive' women (carrier or affected) with the conditions listed below

Link to guidance

NHS Screening Programmes
Sickle Cell & Thalassaemia

**Please fill in the risk status breakdown matrix for all screen positive cases with the conditions below, even for cases where the father results are unknown.**

Scroll right for guidance on completing the risk status breakdown matrix

| Mother's test result | Father's test result: Hb S | βThal | δβ thal | Hb Lepore | Hb D | Hb C | Hb E | Hb O-Arab | HPFH | High risk alpha0 | Compound Hetero-zygous** | *Other | Not a carrier | Father result not available |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Hb S | | | | | | | | | | | | | | |
| βThal | | | | | | | | | | | | | | |
| δβ thal | | | | | | | | | | | | | | |
| Hb Lepore | | | | | | | | | | | | | | |
| Hb D | | | | | | | | | | | | | | |
| Hb C | | | | | | | | | | | | | | |
| Hb E | | | | | | | | | | | | | | |
| Hb O-Arab | | | | | | | | | | | | | | |
| HPFH | | | | | | | | | | | | | | |
| High risk alpha0 | | | | | | | | | | | | | | |
| Compound Heterozygous** | | | | | | | | | | | | | | |
| Egg donor/bone marrow transplant | | | | | | | | | | | | | | |

*** If available please specify any "other" results for father:**

**** Please specify any compound heterozygous results:** *e.g, 2 Mothers SC/Father not carrier; 1 Mother SβThal/Father S, etc.*

**If possible, please identify any homozygous cases shown above:** *e.g. 1 Mother SS/Father not carrier; 2 Mothers EE/Fathers EE, etc.*

**If this breakdown information is unavailable at your lab or is incomplete, please give a brief explanation below:**
*Include any women who are not categorised above where a test for the baby's father has been requested (include father result where available).*

| Risk: | | Totals: | |
|---|---|---|---|
| High Risk | 0 | Sickle Variants | 0 |
| Low Risk | 0 | Total Thal | 0 |
| Minimal Risk | 0 | High Risk Alpha0 | 0 |
| Not a carrier | 0 | Total HPFH | 0 |
| PNA | 0 | Total screen+ | 0 |

*Please note: The "Risk" and "Total" fields will populate automatically and do not need to be filled in as part of the return*

### Guidance on completing this matrix:

**What to include in this matrix:**
Fill in the matrix for **all screen positive mothers with the specified haemoglobinopathy results.**

**Mother/father results:**
Mother results are respresented by the rows while father results are represented by the columns. For example, if there were 2 mothers with an HbS result where the fathers in both cases also had an HbS result, a 2 would go in the cell D10. If there were 3 cases where the mother had an HbS result but the fathers were not carriers, a 3 would go in cell P10, and so on.

**Homozygous cases:**
For cases where either the mother or the father is affected, for the purposes of this matrix these cases would still be put under the Hb variant that they are affected with. For example, both Hb AS and Hb SS cases would be counted under Hb S on this matrix.

**Father result not available:**
This column allows you to account for any screen 'positive' mother results where the father was not available for testing, or where father results cannot be linked to the mother's result.

**Egg donor/hard bone marrow cases:**
Please give details in the comments box below the table.

**Report formats 4a (β thal carriers) and 4b (possible β thal carriers):**
Include both under "βThal".

**'High Risk'** should reflect **HP16: 'High Risk'** in part one

**'Total Screen+'** should reflect **HP11: 'Screen Positive'** in part one

**Data quality check (Programme use only):**

| All high risk couples accounted for? | All screen+ mothers accounted for? |
|---|---|
| Yes | Yes |

v 26/03/2014

# PND Data Returns

PND RETURN
Data fields for Sickle Cell and Thalassaemia Screening Programme
Annual collection - April 1st to March 31st

NHS
Antenatal and Newborn Screening Programmes

| | | DATA ITEM | DATA VALUES |
|---|---|---|---|
| SAMPLE | PND 01 | Date sample taken | Provide date |
| | PND 02 | Date sample received | Provide date |
| | PND 03 | Sample type | Amnio |
| | | | CVS |
| | PND 04 | Valid sample | Yes |
| | | | No |
| | PND 05 | Repeat sample | Yes |
| | | | No |
| | PND 06 | Internal or External sample<br>Internal sample is sample taken in same hospital as laboratory | Internal |
| | | | External |
| PARENTS | PND 07 | EDD | Esitmated Delivery Date |
| | PND 08<br>PND 09 | Purported paternal genotype<br>Purported maternal genotype<br>Allele 1 and Allele 2 status for both partners<br>eg Normal / Hb S (two data fields each) | Normal |
| | | | Hb S |
| | | | Hb C |
| | | | Hb D Punjab |
| | | | Hb E |
| | | | Hb O-Arab |
| | | | Lepore |
| | | | β Thal |
| | | | α Thal |
| | | | Sample not available |
| | | | Other |
| | PND 10 | Mother's PCT (or postcode)<br>by GP postcode if available<br>or by mothers residence<br>or by referring hospital | There are currently 152 PCTs in England |
| | | | Unknown |
| | PND 11<br>PND 12 | Mother's FO<br>Father's FO<br>Plese note that this is different to ethnic category | African |
| | | | Afro Caribbean |
| | | | Cypriot |
| | | | Other Mediterranean |
| | | | Indian |
| | | | Pakistani |
| | | | Bangladeshi |
| | | | Middle Eastern |
| | | | South East Asian |
| | | | Other |
| | | | Not stated |

NHS Sickle Cell & Thalassaemia Screening Programme

Antenatal and Newbo Screening Programm

# NEWHAM 'AT RISK PREGNANCY' ALERT FORM

| Maternal Surname | First Name | DOB | NHS No | Hb'pathy screen result | Place of test | Date |
|---|---|---|---|---|---|---|
| | | | | | | |
| **Paternal Surname** | **First Name** | **DOB** | **NHS No** | **Hb'pathy screen result** | **Place of test** | **Date** |
| | | | | | | |

| Maternal Address including Post code | Tel No (home) | Tel No (mobile) |
|---|---|---|
| | | |

| GP | GP Address | GP Tel No | Named Obstetrician |
|---|---|---|---|
| | | | |

| Gravida/Parity | EDD | Gestation | Maternity Unit | Referrer's Name | Referrer's Tel No | Date of referral |
|---|---|---|---|---|---|---|
| | | | | | | |

| Hb'pathy CNS accepting referral | Want to consider PND in this pregnancy? (tick for yes) | Appropriate to defer contact by Genetics Unit until after outcome of pregnancy? (tick for yes) | Clinical Genetics notified (date of notification) |
|---|---|---|---|
| | ☐ | ☐ | |

| Other relevant information/comments, language problems, previous affected family member, late booker, social problems (lone worker issues) | NNS Lab notified (date) |
|---|---|
| | |

**REPORT FROM BARTS NEWBORN SCREENING LABORATORY**

| RECORD FOUND: | Baby's Name | M/F | Baby's DOB | Baby's NHS No | Baby's Address |
|---|---|---|---|---|---|
| | | | | | |

| Date of Specimen | Date of Test | Hb Pattern | NNS Lab Signature | Date |
|---|---|---|---|---|
| | | | | |

| Enquirer's Name | Enquirer's Tel No | Enquirer's Postal Address |
|---|---|---|
| | | |

## Additional References

[1] NHS Sickle Cell & Thalassaemia Screening Programme *Standards for the Linked Antenatal and Newborn Screening Programme 2nd Edition October 2011* http://sct.screening.nhs.uk/standardsandguidelines [Accessed February 2015]

[2] National Institute for Health and Clinical Excellence *CG-62 Antenatal – NICE Guideline* (2008, modified December 2014) http://guidance.nice.org.uk/CG62/NICEGuidance/pdf/English. [Accessed February 2015]

[3] National Institute for Health and Clinical Excellence Quality standard for antenatal care. September 2012. Nice quality standard 22 http://guidance.nice.org.uk/qs22 [Accessed February 2015]

## Reading List


British Committee for Standards in Haematology Guidelines (2010) *Significant haemoglobinopathies: guidelines for screening and diagnosis.* British Journal of Haematology 149, 35-49

East London NHS Foundation Trust (June 2012) *Policy for service users who fail to attend appointments in children, young people, sexual and reproductive health services and CAMHS.* Trust Intranet

Healthcare staff incident reporting http://www.nrls.npsa.nhs.uk/report-a-patient-safety-incident/healthcare-staff-reporting/

East London NHS Foundation Trust (July 2012) *Screening Proceedures Policy.* Trust Intranet

Map of Medicine (2010 revised 2013) *Linked Sickle Cell and Thalassaemia Screening - Antenatal Screening Pathway* http://eng.mapofmedicine.com/evidence/map/linked_sickle_cell_and_thalassaemia_screening1.html

NHS Sickle Cell & Thalassaemia Programme *Laboratory Handbook 3rd Edition October 2012* http://sct.screening.nhs.uk/standardsandguidelines [Accessed February 2015]

UK National Screening Committee (2014) *Key Performance Indicators for Screening 2014/15 Version 1.1.14*

NHS Sickle Cell & Thalassaemia Screening Programme (2012) *Guidelines for the referral of sickle cell and thalassaemia prenatal diagnosis samples to molecular haemoglobinopathy laboratories. Version2.4* http://www.sct.screening.nhs.uk/guidance [Accessed March 2015]

NHS Sickle Cell & Thalassaemia Screening Programme (2015) *Data Report 2013-14.*

NHS Sickle Cell & Thalassaemia Screening Programme (2012) *Policy Framework for Antenatal Sickle Cell & Thalassaemia Screening Programme* http://www.sct.screening.nhs.uk/policy-antenatal [Accessed March 2015]

NHS Sickle Cell & Thalassaemia Screening Programme (2013) *Counselling Competencies* *http://sct.screening.nhs.uk/standardsandguidelines*

NHS England Serious Incidents Framework http://www.england.nhs.uk/wp-content/uploads/2015/03/serious-incident-framwrk-15-16-faqs-fin.pdf

NHS England http://www.england.nhs.uk/ourwork/patientsafety/serious-incident/

Patient Safety Serious Incident Reporting & Learning Framework http://www.nrls.npsa.nhs.uk/report-a-patient-safety-incident/serious-incident-reporting-and-learning-framework-sirl/

Royal College of Obstetricians and Gynaecologists (2010 - Reviewed December 2014) Amniocentesis and Chorionic Villus Sampling (Green top Guideline 8) https://www.rcog.org.uk/globalassets/documents/guidelines/gtg_8.pdf

University of York (2014) *Involving Fathers in Antenatal Screening for Sickle Cell Disorders: Improving Informed Decision Making* http://www.sct.screening.nhs.uk

## Useful Websites

Brent Sickle Cell & Thalassaemia Centre http://sickle-thal.nwlh.nhs.uk/

Healthtalkonline (DIPEx) http://www.healthtalk.org/

NHS Choices http://www.nhs.uk/Pages/HomePage.aspx

Sickle Cell Society http://sicklecellsociety.org/

UK Thalassaemia Society http://ukts.org/

Unit for the Social Study of Thalassaemia & Sickle Cell http://www.tascunit.com/