Brighton and Sussex University Hospitals NHS Trust

Antenatal screening for Sickle Cell and Thalassemia

Maternity Protocol MP003

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	MP079 Newborn Bloodspot Screening
	MP036 Care in Labour of women with Haemoglobinopathies

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Key Principles

A protocol is a set of measurable, objective standards to determine a course of action. Professional judgement may be used in the application of a protocol.

Scope

This protocol applies to:

- Women who do not attend appointments

Responsibilities

Midwives & Obstetricians

- To access, read, understand and follow this guidance
- To use their professional judgement in application of this protocol

Management

- To ensure the protocol is reviewed as required in line with Trust and National recommendations
- To ensure the protocol is accessible to all relevant staff

Objective Standards

1 Antenatal Screening Sickle Cell and Thalassaemia

Background to the screening programme is given in Appendix A.

The antenatal screening programme is linked to the newborn blood spot screening programme.

1.1 Aims

The NHS Sickle Cell & Thalassaemia (SC&T) Screening Programme aims to:

- Ensure a high quality, accessible screening programme throughout England
- Support people to make informed choices during pregnancy and ensure timely transition into appropriate follow up and treatment
- Improve infant health though prompt identification of affected babies and timely transition into clinical care
- Promote greater understanding and awareness of the conditions and the value of screening (NHS England 2018)

1.2 Antenatal Sickle cell and thalassaemia screening programme: Objectives and Outcomes

To offer timely antenatal sickle cell and thalassaemia screening to all women, to Identify women and then couples, who are at increased risk of an affected pregnancy.

To facilitate informed decision making to those women [and couples] who are at increased risk, with the option of prenatal diagnosis and the option of termination or continuation of an affected pregnancy (NHS England 2018
For those women accepting prenatal diagnosis, 50% of prenatal diagnoses to be performed before 12weeks 6 days (NHS England 2018).

1.3 Newborn Sickle Cell Screening programme: Objectives and outcomes

To identify babies born with conditions where early intervention is likely to be beneficial (NHS England 2018)

To achieve the lowest possible childhood death rate and to minimise childhood morbidity from sickle cell diseases (NHS England 2018.

Please refer to the protocol for newborn screening for further information about these aims and objective: MP079 Newborn bloodspot screening.

1.4 Linked Antenatal and Newborn Sickle Cell and Thalassaemia Screening Programme

Aim: to link results from antenatal screening tests taken by parents-to-be with their baby's test result (NHS England 2016)

Objectives and Outcomes [NHS England 2016]:

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

1.5 For designated programme leads for maternity services and link speciality staff. Please see Appendix B

2 Pre-Screening information

2.1 All women should be given verbal and written information about screening for sickle cell and thalassaemia in pregnancy during the first contact with the midwife.

2.2 The trust uses the UK National Screening Committee [UK NSC] national patient information leaflet 'Screening tests for you and your baby'. This includes the section 'Sickle cell and thalassaemia.' This leaflet may be sent to the woman by post prior to the first appointment or given out at the first appointment. The midwife must document in the hand held notes that the leaflet has been received by the woman.

2.3 Copies of 'Screening tests for you and your baby' are available in other languages and can be downloaded from: https://www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief . If the leaflet is not available in the language required from the national website, it is possible to ask for the leaflet to be translated via the trust's Equality and Diversity Team.

2.4 Where there are specific communication requirements [e.g. English is not the woman's first language, visual/ hearing impairment] appropriate interpretation services should be used at booking and all subsequent stages in the screening pathway.

3 Offering Screening In Pregnancy

3.1 The Eligible Population

All pregnant women are eligible for screening

Screening for sickle cell, other haemoglobin variants and thalassaemia should be offered to all women as early as possible in pregnancy (NICE 2008; section 1.6.3.3, NHS England 2018).

The aim is to screen women before 10 weeks (i.e. by 9+6 weeks at the latest). Offering screening later may limit the choices available to parents should they be diagnosed as being at-risk of having a baby with a major haemoglobin disorder as screening decisions are often gestation dependent (Streetly 2001). However, screening can be offered at any point during the pregnancy. See section 3.7 for women booking late in pregnancy.

Screening should be offered in each pregnancy, regardless of past results. If a woman has been tested previously and haemoglobin carrier status is known, the test should still be repeated in a new pregnancy.

Screening should be offered even if information about the baby's father or second biological parent is unavailable

3.2 Known At-Risk Couples

Couples identified at booking as already knowing their carrier status (at –risk couples) should be referred immediately to the antenatal screening midwives for assessment and offer of prenatal diagnosis. Referral by the community midwife should be made by phone or email.

3.3 The type of screening offered depends upon the prevalence of sickle cell in the local population [NICE 2008]: currently BSUH NHS TRUST is designated as a low prevalence trust [NHS England 2017
As a low prevalence trust, all women are offered screening for thalassaemia using routine red blood cell indices.

As a low prevalence trust, screening for sickle cell and haemoglobin variants takes place initially using the Family Origin Questionnaire (FOQ). The FOQ assesses the risk of either the woman or the baby's father or second biological parent, being a carrier for sickle cell or other haemoglobin variants, or severe alpha thalassaemia. The laboratory uses this information as a basis for determining which women to test for the haemoglobin variants and helps in the interpretation of laboratory results.

At BSUH the FOQ has been incorporated in the antenatal booking blood form.

4 Limitations to Screening

1 in 1000 Caucasian people are carriers for a haemoglobin disorder, it is recognised by the National Programme Centre that selective screening based on the Family Origin Questionnaire (FOQ), will not identify all carriers of a haemoglobin disorder. Women should be informed of this in the pre-test discussion with the midwife.

5 Pre-Test Discussion

At booking the midwife should discuss the reasons why screening is recommended. Pre-test discussion should ideally include the following according to individual client need:
The nature and effects of haemoglobin disorders
The test (noting that an extra sample is NOT required)
The chance of being a carrier according to family origin
The limitations of screening (as per 3.4)
These are screening not diagnostic tests and they will not identify all haemoglobinopathies
The results process
Options for pre-natal diagnosis and continuing the pregnancy or termination should a baby be diagnosed with a major haemoglobinopathy.

The midwife should document in the hand held notes that discussion has taken place and the woman's decision to accept (consent to screen) or decline screening. Screening should only be performed after documented informed consent; this does not require a signature from the woman.

The midwife can refer to the antenatal screening midwives by phone or email if parents require more detailed information or discussion at any stage in the screening pathway. The phone number for the antenatal screening midwives can also be given direct to the parents (see contacts Appendix B).

6 Using The Family Origin Questionnaire (FOQ) / Booking Blood Request Form

6.1 The FOQ is incorporated into the booking blood request form which has been approved for use by the National Programme Team.

6.2 All sections of the form must be completed in full in legible handwriting. The request form is in triplicate. Therefore when completing the form, ensure that the three sheets are aligned and that the writing has transferred to all three sheets. Where a sticky patient identification label is used, this must be affixed to all three sheets.

6.3 Ask the woman and baby's father or second biological parent, if they or their blood relatives originating from any of the areas listed on the form, for as many generations back as they are aware of. It may be necessary to tick more than one box for each parent if they have origins from more than one country.

6.4 Remember the partner may not be the father or second biological parent of the baby and so it is important to ask about the baby's biological father or second biological parent rather than partner.

6.5 In the case of pregnancies conceived by sperm and / or egg donation, the family origins of the donor/s are required and included on the form if known.

6.6 If the family origins of either biological parent is not known [for example if adopted or if the origins of a donor egg/ sperm are not known], then please tick the 'don't know' adoption/unknown ancestry box. It is important to clarify whether the pregnancy was conceived by sperm and/or egg donor if known.

6.7 If the woman or pregnant person has had a bone marrow transplant please tick the box. The baby's biological father or second biological parent must be tested as the results obtained will reflect the bone marrow transplant donor and not represent the genetic status of the fetus.

6.8 If either parent has been previously tested and the result is known, then please write this on the FOQ.

6.9 Any tick in a yellow box indicates to the laboratory staff that a full haemoglobinopathy screen is required.

6.10 Insert expected date of delivery, gestation and parity.

6.11 If screening is declined, please tick appropriate box.

6.12 The section on blood transfusion history must be completed as this information is essential for interpreting results of sickle cell and thalassaemia screening. Results may be misleading if there has been a blood transfusion in the last four months.

7 Women who book late or arrived unbooked in labour

7.1 Women booking late must be offered screening at the first appointment with the midwife.

7.2 Note that when screening takes place in the mid or third trimester, and the couple are found to be at-risk, the options available to the couple may be limited. The consequences of late screening should be discussed prior to screening.

7.3 For women arriving unbooked in labour, whilst it may be too late to detect an at-risk couple prior to the birth, it is still important to know if a woman is a sickle cell carrier, as this may impact medically (for example with anaesthesia), and is important to know for future pregnancies.

8 Women who transfer in

8.1 Women who booked elsewhere prior to transferring care to BSUH NHS TRUST should be offered repeat screening. This ensures a result is available in-house for all staff to access electronically if the hand held notes are not available.

8.2 If a woman declines repeat testing having been screened elsewhere, then a copy of the original result from the unit of first booking must be inserted in the hand held notes and hospital notes.

8.3 Women who have had screening outside of the UK must be advised that a repeat screen is recommended as their original screening test may not meet UK standards and may omit screening for certain conditions.

9 Declining screening

9.1 Screening is optional. All women have the right to decline screening.

9.2 Where screening is declined, the midwife at booking should continue to complete the FOQ section of the antenatal booking blood form and tick the box which states 'screening test declined'. If known, the reason for declining should be entered onto the form. The form should be forwarded to the laboratory either with the other blood samples (if screening for other tests accepted) OR via internal post if all blood tests were declined.

9.3 The decision to decline screening must be clearly documented by the midwife in the hand held notes.

9.4 Where women decline screening, ensure they are aware that they can opt to accept screening at a later date. Screening can be arranged through their community midwife at subsequent appointments. In such cases the antenatal booking blood form must be used for screening as it incorporates the FOQ which is an essential component of the screening test.

10 Taking the Sample (From the Woman)

10.1 The sample may be taken at booking by the midwife, practice nurse or phlebotomist according to local arrangements:

- A 4ml sample of blood is required in an EDTA tube [purple top with black ring]. The test can be performed on the same sample as used for the full blood count
- The Antenatal Booking Blood Request Form, must be completed fully prior to taking blood as per section 3.6 and accompany the sample to the laboratory

10.2 Samples are sent to BSUH Pathology for processing and should arrive within one working day of sample collection.

11 Unacceptable Samples

Where the sample is deemed to be unacceptable by the laboratory because of insufficient blood, incomplete data on the request form, mislabeling of sample bottle or for any other reason, the laboratory will inform the antenatal screening midwives by email. The screening midwives will inform the relevant community midwife team leader in order that they may action a repeat sample or a repeat FOQ.

Samples/FOQs should ideally be repeated within 10 working days of the request being received by the maternity unit.

11.1 Where repeat samples take longer than 10 working days to arrange (for example the woman is away or declines to attend for repeat sampling within 10 days), the midwife should document reasons why there has been a delay.

11.2 The midwife responsible for actioning the repeat must always follow up the results and arrange a plan to deliver the results to the woman being mindful that there has already been a delay in obtaining a result because of the need to repeat.

11.3 The laboratory team enters the details of every rejected sample or FOQ onto a database and updates the entry when the repeated sample has been received in the laboratory. The database is accessible by the screening midwives who review every Wednesday and action any outstanding repeats.

12 Results Processes

12.1 Accessing results (Including fail-safes to ensure all women who accept screening receive a result)

12.1.1 Results will be available to staff within 3 working days of sample receipt in the laboratory. Results will be available on ICE (the electronic pathology results reporting programme).

12.1.2 While the majority of results will be available within 5 working days, in the case of some rare haemoglobin disorders, final diagnosis may take some weeks. In these cases the laboratory team will inform the antenatal screening midwives in order to advise the woman and CMW of the reason for the delay.

13 Failsafe

10 day check

It is the responsibility of the sample requestor to follow up results within 10 working days of the sample being taken. The sample requestor should ensure carrier results have been acted upon and follow up missing results or laboratory requests for a repeat sample. The sample requestor must document that results were followed up and acted on. An example proforma that can be used for recording that results were followed up is given in appendix F.
If any results are missing at the 10 day check the laboratory should be contacted to confirm a result is being processed. If a result is found to be missing during a weekend clinic the community midwifery leads should be emailed to action the missing result on the next working day. A confirmation email will be sent back to the midwife who found the result to be missing, once the community leads have the result.
Whilst every attempt should be made to check results after 10 working days it is recognised that some community clinics do not have facilities for venepuncture and therefore women attend hospital phlebotomy to have bloods taken. In such cases, the sample requestor will not

know when the bloods were actually taken and so cannot follow up results at 10 days. In such cases it is essential that the requestor follows up results at the next AN appointment.

Next AN appointment
It is the responsibility of the health professional (midwife or doctor) providing care at the next antenatal appointment (usually at the 16 week appointment) to check the results, document results (with informedconsent) in the hand held notes and inform the woman of the results during the appointment.

Results check at time of dating scan
As an additional failsafe, the Antenatal Screening Support Worker (ASSW)will ensure a full set of booking blood results are available at the time women attend dating scan. Where the woman has not yet had screening, this can be offered at the time of scan.

Women who miscarry or terminate following screening
All women should be notified of their results following testing and this includes women who terminate or miscarry following screening. This is especially important with women who are found to be carriers, as this affords the opportunity for partner screening in advance of a future pregnancy. Carrier women will be informed by the screening midwives in order to discuss implications for a future pregnancy. Non-carrier women should be informed by the midwife who requested the sample.

14 Screening Results: Interpreting Results and Follow Up

Results will fall into one of four possible categories:

- Nothing abnormal detected: Non-carrier result
- Inconclusive result
- Clinically significant haemoglobin disorder
- Genetic carrier

If there is any uncertainty about a result, contact either the screening midwives in the first instance or the laboratory staff for further advice.

14.1 Nothing abnormal detected: Non-carrier results

These will be either one of the following:
Not a carrier of thalassaemia, sickle cell or haemoglobin variant (women assessed as high risk by FOQ). The woman was screened for sickle cell, Hb variants and thalassaemia and nothing abnormal was detected.

Not a carrier of thalassaemia (for women assessed as low risk by FOQ)

The woman was screened for thalassaemia only, because neither she nor her partner had family origins from an at risk region. Nothing abnormal was detected.
However as she werewas only assessed for thalassaemia, there is still a small chance that she could be an undiagnosed carrier of sickle cell or Hb variant (see section 3.4).

In both cases the woman should be informed that nothing abnormal has been detected and there is no indication to test the baby's father or second biological parent. However screening is not diagnostic and will not detect all carriers. Therefore we cannot exclude the small chance of the woman being an undiagnosed carrier.

There is no further action in these cases as testing of the baby's father or second biological parent is NOT indicated.
It is appropriate for the community midwife to discuss these results with women.

14.2 **Inconclusive Result**

These results must be actioned by the screening midwife team. When community midwives discover an inconclusive result whilst following up booking screening results, they should contact the screening midwife team by email or phone to ensure they are aware of the result.

Some haemoglobin variants are difficult to identify and analysis of the original sample may take longer. Occasionally further tests on the woman may be indicated.
In such cases the laboratory will email the antenatal screening midwives to advise that the results is inconclusive and whether further tests should be offered. Testing the baby's father or second biological parent is advised f nothing abnormal is detected, the risk to the baby can be excluded.
The screening midwives will inform the woman of the inconclusive result and possible implications. The screening midwives will discuss and offer testing of the father or second biological parent and arrange this where accepted.

14.3 **Clinically significant haemoglobin disorder**

These results must be actioned by the screening midwife team. When community midwives discover a significant haemoglobin result whilst following up booking screening results, they should contact the screening midwife team by email or phone to ensure they are aware of the result.

While most people will know if they have a significant haemoglobin disorder, in rare cases this is only detected for the first time at antenatal screening. In such cases the laboratory will email the antenatal screening midwives to advise that the woman has a significant haemoglobin disorder.

The screening midwives will inform the woman of the result and possible implications for both the woman and unborn baby. The screening midwives will discuss and offer testing of the baby's father or second biological parent and arrange this where accepted.

In addition the antenatal screening midwives will refer to the consultant obstetrician and haematology services to ensure appropriate care is offered with regard the woman and theirher pregnancy (refer to protocol: MP036 Care in Labour of women with Haemoglobinopathies).

14.4 Genetic carrier: Action following identification of maternal haemoglobinopathy carriers

These results must be actioned by the screening midwife team. When community midwives discover a carrier result whilst following up booking screening results, they should contact the screening midwife team by email or phone to ensure they are aware of the result.

In such cases the laboratory will email the antenatal screening midwives to advise that the woman is a carrier or suspected of being a carrier for a haemoglobin disorder.

The screening midwives will inform the woman of the result and possible implications. The screening midwives will discuss and offer testing of the baby's father and arrange this where accepted.

Informing women of carrier results and offering testing of baby's father

If the woman has an inconclusive result, is a carrier of a significant haemoglobinopathy or diagnosed with a haemoglobin disorder, then both the woman and the father of her baby should be offered counselling and paternal screening without delay.

It is the responsibility of the antenatal screening midwives to contact the mother and explain the significance of the result and offer screening for the baby's father for sickle cell, other haemoglobin variants and thalassaemia.

Women should be contacted within 5 working days of the result being generated by the laboratory.

The antenatal screening midwives contact the woman by phone to inform her of the result and offer an appointment for further discussion.

If it is not possible to contact the woman by phone, a letter should be sent the same working day asking the woman to contact the screening midwives to discuss the carrier status. An example template letter is given in Appendix D.
A letter inviting the father for screening can also be enclosed with the mother's letter that she can give to him. An example letter template is given in Appendix E.

Further counselling may take place in person face-to-face or over the phone according to client's preference.

Discussion of the result should include the following:

- Haemoglobin disorders and the specific Hb type found in the mother
- The screening test for dads
- Inheritance patterns and possible risks of the baby inheriting a condition if the father is found to be a carrier
- Choices available to the baby's parents should they be found to be at risk of having a baby with a major haemoglobin disorder [to include the option of prenatal diagnosis, postnatal diagnosis, continuing or ending a pregnancy should the baby be affected]

Verbal information should be backed up with written information. National patient information leaflets explaining the commonly diagnosed carrier traits can be found at https://www.gov.uk/government/collections/adult-carriers-sickle-cell-thalassaemia-unusual-haemoglobin

Additional resources to help explain carrier status and inheritance to parents can be found at the APoGI website (see contacts Appendix).

The National Information Leaflet: 'Sickle cell and thalassaemia screening: information for fathers' [a non-gendered version is not currently available] should be offered to all women to give to their baby's father (and be enclosed in the letter Appendix E). The leaflet is accessible online at: https://www.gov.uk/government/publications/tests-for-dads-sickle-cell-and-thalassaemia-screening

The baby's father should be counselled and paternal blood samples taken [where accepted] ideally by the end of 11 weeks of pregnancy (NHS England 2016).

All discussions must be documented in the hand held or hospital notes by the midwife counselling the couple.

15 Taking the sample from the baby's father or second biological parent

15.1 Where a woman is identified as being a carrier and baby's father or second biological parent has accepted screening:

- A 4ml sample of blood in an EDTA tube should be sent to the laboratory
- A routine blood sciences form should be used requesting 'Hb electrophoresis'. This should be completed in full by the midwife arranging the test.
- The form must include written details of who the partner of (give mother or birthing parent's name and date of birth). This ensures samples can be tracked and matched up with the correct woman.

Where available the father or second biological parent's NHS number and hospital ID number should be used. Where there is no NHS / Hospital ID number the sample requestor must write the following on the form: Acceptance of sample without NHS/ Hospital ID number has been agreed by haemoglobinopathy laboratory and AN screening team.
The paternal or second biological parent's blood sample may be taken at the GP surgery, antenatal clinic or phlebotomy department by the midwife / practice nurse / phlebotomist: whichever allows the earliest opportunity for sampling. The father or second biological parent should be given the correct form to take along, completed as detailed in section

15.2 Taking the sample from the baby's father or second biological parent where indicated.

If the baby's father or second biological parent consents to testing, their permission must be sought to share information about their haemoglobin type with the baby's mother or birthin parent. The baby's father or second biological parent's results cannot be released to the woman or birthing parent without their consent.

A plan should be made by the antenatal screening midwife to communicate the father or second biological parent's result back to the parents in the hand held notes. Results will normally take no more than 5 working days.

The laboratory email the father or second biological parent's result to the screening midwife team as soon as it is known to facilitate prompt follow up.

Record of care, information given, partner consent to testing and results process should be documented in the hand held or hospital notes.

15.3 Where the baby's father or second biological parent has been screened previously

If the baby's father or second biological parent has been tested previously, a repeat screening test should be performed for all subsequent **pregnancies "For every pregnancy we need to test both parents to see if there is any risk for your baby". (PHE 2018)**

If the father or second biological parent declines a repeat test, a copy of the first result should ideally be obtained.

In the absence of a written result or if obtaining a copy of the result is likely to take more than a week, then a repeat screening test should be recommended.

Note that if the father or second biological parent has only ever been screened outside of the UK then a repeat screen should always be recommended. This is because their original screening test may not meet UK standards and may omit screening for certain conditions.

15.4 Where the baby's father or second biological parent declines screening or is not available

If the baby's father or second biological parent is not available for screening or declines screening and haemoglobinopathy status is unknown, further advice should be given to the mother as to the likelihood of him being a carrier based on his family origins (if known).

In such cases pre-natal diagnosis should be offered to the woman. Discussion should include information about the risk of miscarriage from prenatal diagnosis. The risk of miscarriage should be balanced against the likelihood of the father or second biological parent being a carrier.

These cases should be managed on an individual basis in conjunction with the fetal medicine team. Where prenatal diagnosis is requested, these cases MUST be discussed with the appropriate prenatal diagnostic laboratory (see contacts) prior to testing to ascertain whether additional blood samples are required from the mother or birthing parent and to clarify to likelihood of limitations to testing with the absence of paternal or second biological parent DNA diagnosis.

15.5 **Follow up of Paternal Blood Results**

15.5.1 Nothing abnormal detected on screening of baby's father:
The parents should be informed by the screening midwife that they are not at risk of having a baby with a major haemoglobin disorder. However there is a 50% chance that their baby will inherit the same heamoglobin type as found in the mother. The implications of this [according to haemoglobin type] should be discussed with parents.

- If the mother is a carrier of sickle cell or other unusual heamoglobin variant, the newborn blood spot screening programme will usually detect whether the baby has inherited this. Parents will be informed by their health visitor.

- If the mother is a carrier of thalassaemia, then this will not be detected on newborn blood spot screening and parents will not know if their baby is a carrier unless tested for other reasons.

In all cases, the screening midwife should send a letter to the parents confirming the results. A copy should be sent to the woman's GP, community midwife. A copy should be filed in the hospital notes.

15.5.2 Where the father is found to be a carrier at-risk couple identified

Ideally, at-risk couples (where both mother and father are identified as carriers of a significant haemoglobin disorder) should be identified by 12weeks 6 days of pregnancy (NHS England 2016).

16 Care Following Diagnosis Of At-Risk Couple

A couple may be diagnosed at-risk of having a baby with a major haemoglobin disorder either pre-conceptually or through the antenatal screening programme.

Known at-risk couples should be referred direct to the antenatal screening midwives at booking to expedite discussions about prenatal diagnosis.

Couples identified as being at-risk on the AN screening pathway should be offered an appointment in antenatal clinic at the earliest opportunity (ideally within 5 working days of partner result). This appointment may be with either a consultant obstetrician or midwife who has undergone additional training (antenatal screening midwife who has completed a genetic risk assessment and counselling module).

Discussion should include the possible outcomes for their baby regarding haemoglobin type (inheritance patterns) and implications for an affected baby, methods and risks of prenatal diagnosis and the options available should a major haemoglobin disorder be identified (including continuing or terminating the pregnancy).

Where unusual haemoglobin types are identified, specialist counselling can be accessed by referring the couple in writing to the haematology department (prenatal diagnosis team) at Kings College Hospital (contact details given in Appendix G).

The option of discussing care for a baby with a major heamoglobinpathy with a paediatrician should also be given. See contacts for referral details.

Written information should be provided along with written confirmation of carrier status. The following leaflets are available:

Haemoglobin blood test results: information for a couple at risk of having a baby with sickle cell disease. Available at www.gov.uk/government/publications/baby-at-risk-of-having-sickle-cell-disease-description-in-brief

Haemoglobin blood test results: information for a couple at risk of having a baby with thalassaemia major. www.gov.uk/government/publications/baby-at-risk-of-having-thalassaemia-description-in-brief

Further support: For all at-risk couples, extra support surrounding testing, diagnosis and either continuing or terminating an affected pregnancy is available through the charity ARC – Antenatal, Results and Choices. Both the Sickle Cell Society and the UK Thalassaemia Society also provide useful information and support. Contact details are given at the end of this document.

All discussions and subsequent decisions made by the parents should be documented in hand heldmaternal notes by the screening midwife or doctor. The woman's GP, community midwife and health visitor should be informed of the outcome by letter.

16.1 Couples declining prenatal diagnosis

For parents declining pre-natal diagnosis, a plan for post natal diagnosis should be made in advance of the birth (see section 3.9).

The screening midwife will inform the paediatricians, GP, CMW and HV that the couple has declined prenatal diagnosis in writing.

16.2 Couples Accepting Prenatal Diagnosis

If pre-natal diagnosis (PND) is requested, referral is made for CVS or amniocentesis (choice of test is gestation dependent) within 5 working days. CVS is preferable to amniocentesis as it provides more DNA. If amniocentesis is performed and parents request screening for aneuploidy, a 20ml sample is required.

Cases must be discussed with the PND lab at the Kings Red Cell Centre lab before the invasive test is performed as they may also require extra blood samples from the parents to be sent with the invasive sample. The lab will advise regarding turn-around time for results.

Testing for aneuploidy is also available to parents and should be offered as part of the pre-test counselling. If parents requested combined screening, the NT measurement should be recorded prior to invasive testing, and if 3.5mm or more, CGH array is offered for chromosomal analysis. For further information see protocol MP002 Fetal Anomaly Screening Programme

The sample should be sent to the cytogenetics lab at Guys hospital with both a Guys request form (for aneuploidy testing) and the Kings Red Cell Centre lab request form for prenatal diagnosis of haemoglobinopathy.

The Guys lab will extract DNA from the sample and forward it to the Kings red Cell Centre lab for the Haemoglobinopathy test.

PND test results should be available within 4 working days of the procedure.
NHS Sickle Cell and Thalassaemia Screening Programme 2006: objective P3.
Results are emailed to the antenatal screening midwife team.

Parents are informed of the results and may be given over the phone or in person at a hospital appointment according to couple choice, by either a consultant or midwife with PEGASUS training or equivalent i.e Genetic Risk Assessment & Counselling Module..
Written confirmation should always be sent to the parents.

17 Screening Results

17.1 Testing babies born to known at-risk couples.

It is recognised that couples known to be at high risk (1 in 4) of having a baby with sickle cell disease or thalassaemia major, who declined prenatal diagnosis, might wish to know the result for their child before the normal time for reporting the result from the newborn bloodspot screening programme.

To be a known '1 in 4' high risk pregnancy, the haemoglobin results must be known on both parents.

This service should be seen as a special 'on-demand' service to alleviate parental anxiety and not as a fail-safe or substitute for the newborn bloodspot screening programme, nor to gain a more rapid entry into clinical care.

Where parents request postnatal testing, a liquid capillary blood specimen **(not cord blood)** should be taken from the baby for analysis soon after birth.

The specimen should be taken in a clinical setting by the neonatal team.

This blood specimen should be sent to a specialist laboratory, which has expertise in analysing and interpreting results on newborn babies. Therefore samples taken at BSUH CANNOT be analysed in-house and must be sent to the Red Cell Centre at Kings College Hospital, London (See contacts).

The neonatal team have responsibility for following up the results and arranging to communicate these with the parents.

The fact that such a specimen has been taken should be noted by the midwife on the newborn screening bloodspot card. The screening laboratory will undertake the routine screen as usual and the test will act as a 'failsafe' and quality check. It is imperative that the test result is given to the parents as soon as possible to decrease their natural anxiety about the status of their child. Testing of the liquid specimen should be seen as a parallel test to the screening specimen and not a substitute.

Further information and best practice guidance on screening is available in the Laboratory Handbook (Oct 2017) developed by the NHS Sickle Cell & Thalassaemia Screening Programme Centre.

18 Failsafe arrangements

The antenatal screening midwives maintain a shared database of all carrier women recording offer, acceptance or decline of partner test, partner test results and outcomes for at-risk couples.

On a weekly basis (every Wednesday) the antenatal screening midwives cross check their database of carrier women with the list of carriers held by the laboratory. This ensures that the screening midwives have been informed of all carrier women.

This is reviewed on a weekly basis to ensure all the results are followed up in a timely manner. Data and audit findings will be included in the Annual Screening Report compiled by the Antenatal Screening Co-ordinator.

19 Governance

All incidents relating to antenatal screening for sickle cell and thalassaemia screening are reported via the trusts internal reporting system known as DATIX.

All incidents related to screening for sickle cell and thalassaemia should also be reported to the Antenatal screening co-ordinator who will liaise with the and where necessary complete a SIAF (Screening incident assessment form) in order to notify the Regional QA screening team and the Screening and Immunisations Lead.

All incidents related to screening for sickle cell and thalassaemia should also be reported to the laboratory and the Pathology Quality Team.

For further information relating to management of incidents please refer to the protocol: MP085 Maternity & Gynaecology Risk Management Strategy.

The Antenatal Screening board for sickle cell and thalassaemia meets quarterly and will address all issues pertaining to the programme at BSUH.

20 Training

All midwives must attend a yearly update on antenatal screening for sickle cell and thalassaemia as part of their mandatory education update sessions.

All midwives new to the trust should complete the Antenatal & Newborn Screening eLearning Module.

All band 5 midwives must complete the Antenatal & Newborn Screening eLearning Module. as part of their competencies before they can apply for a band 6.

All screening midwives must complete a course in genetic risk assessment and haemoglobinopathy screening in order to counsel at-risk couples.

21 Data and Monitoring

Minimum auditable standards: the following data is collected

- number of bookings
- number of women tested
- number of women tested by 10 weeks
- number of women identified as carriers for haemoglobinopathies
- number of partners tested
- number of partners tested by 13 weeks
- number of at-risk couples
- outcome of at-risk pregnancies

The above data is reported on an annual basis to the following organisations (See Contact list in Appendix F & G):

- Regional Screening and Immunisation Lead
- Regional Quality Assurance Screening Lead

Data for the NSC Key Performance Indicators for SC&T screening collected and submitted on a quarterly basis. Current KPI data requirements for SC &T screening are available at http://www.screening.nhs.uk/kpi

An annual audit of women's notes will take place to review documentation processes.

Data and audit findings will be included in the BSUH Trust Annual Antenatal and Neonatal Screening Report compiled by the Antenatal Screening Co-ordinator.

22 References

Department of Health [2000] The NHS plan: A plan for investment, a plan for reform. London.

NHS England [2016] NHS public health functions agreement 2016-17. Service specification no. 18 NHS Sickle Cell and Thalassaemia Screening Programme. Publication date: 5th February 2016.

National Institute for Clinical Excellence – NICE [2008] Clinical guideline 62 – Antenatal care for uncomplicated pregnancies. March
2008: updated Feb 2019
https://www.nice.org.uk/guidance/cg62

Streetly, A [2001] A survey of Haemoglobinopathy Screening Policy and Practice in England. NHS Screening Programme.

NHS Sickle Cell & Thalassaemia Screening Programme [2011 a] Standards for the linked Antenatal and Newborn Screening Programme. 2nd Edition.
https://www.gov.uk/government/publications/standards-for-sickle-cell-and-thalassaemia-screening

NHS Sickle Cell & Thalassaemia Screening Programme [2011 b] Summary of changes :Standards for the linked Antenatal and Newborn Screening Programme
https://www.gov.uk/government/publications/standards-for-sickle-cell-and-thalassaemia-screening

NHS 2017 NHS trusts: area prevalence for sickle cell and thalassaemia
https://www.gov.uk/government/publications/nhs-trusts-area-prevalence-for-sickle-cell-and-thalassaemia

NHS 2018 Guidance Antenatal screening Updated 6 July 2018
https://www.gov.uk/government/publications/handbook-for-sickle-cell-and-thalassaemia-screening/antenatal-screening

PHE 2018

NHS Sickle Cell and Thalassaemia Screening Programme
Information for fathers invited for a screening test for sickle cell disease and thalassaemia major

NHS Sickle Cell and Thalassaemia Screening Programme [20172] Sickle Cell and Thalassaemia: Handbook for Laboratories. October 2017
https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/656094/Antenatal_Laboratory_Handbook.pdf

23 Patient information

UK National Screening Committee [2008] 'Screening Tests for You and Your Baby'. Click on this link to view/ download a copy of the information leaflet 'Screening for sickle cell and thalassaemia in early pregnancy'
Patient information : http://infectiousdiseases.screening.nhs.uk/public

24 Appendix A - Background information: Antenatal screening for sickle cell and thalassaemia

Background

The NHS Sickle Cell & Thalassaemia (SC&T) Screening Programme was set up in England in 2001 following Government commitment in the NHS Plan (2000).

Sickle cell disease and Thalassaemia disorders, also known as haemoglobinopathies or haemoglobin disorders, are autosomal recessive conditions affecting the haemoglobin in the red blood cells. They are classified into 2 forms: qualitative or quantitative:

Sickle Cell disorders: These are qualitative disorders affecting the quality of haemoglobin produced. As the structure of the globin chain is abnormal, they are also termed structural variants. 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, 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.

Thalassaemias are quantitative disorders, including 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 [which are of normal structure] that form the structure of haemoglobin within the red blood cells. This 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.

There are over 1000 known variants of these 2 forms, but most do not give any clinical symptoms. It is possible to carry both conditions. An infant may inherit one form from each parent, which can give rise to a more serious clinical situation.

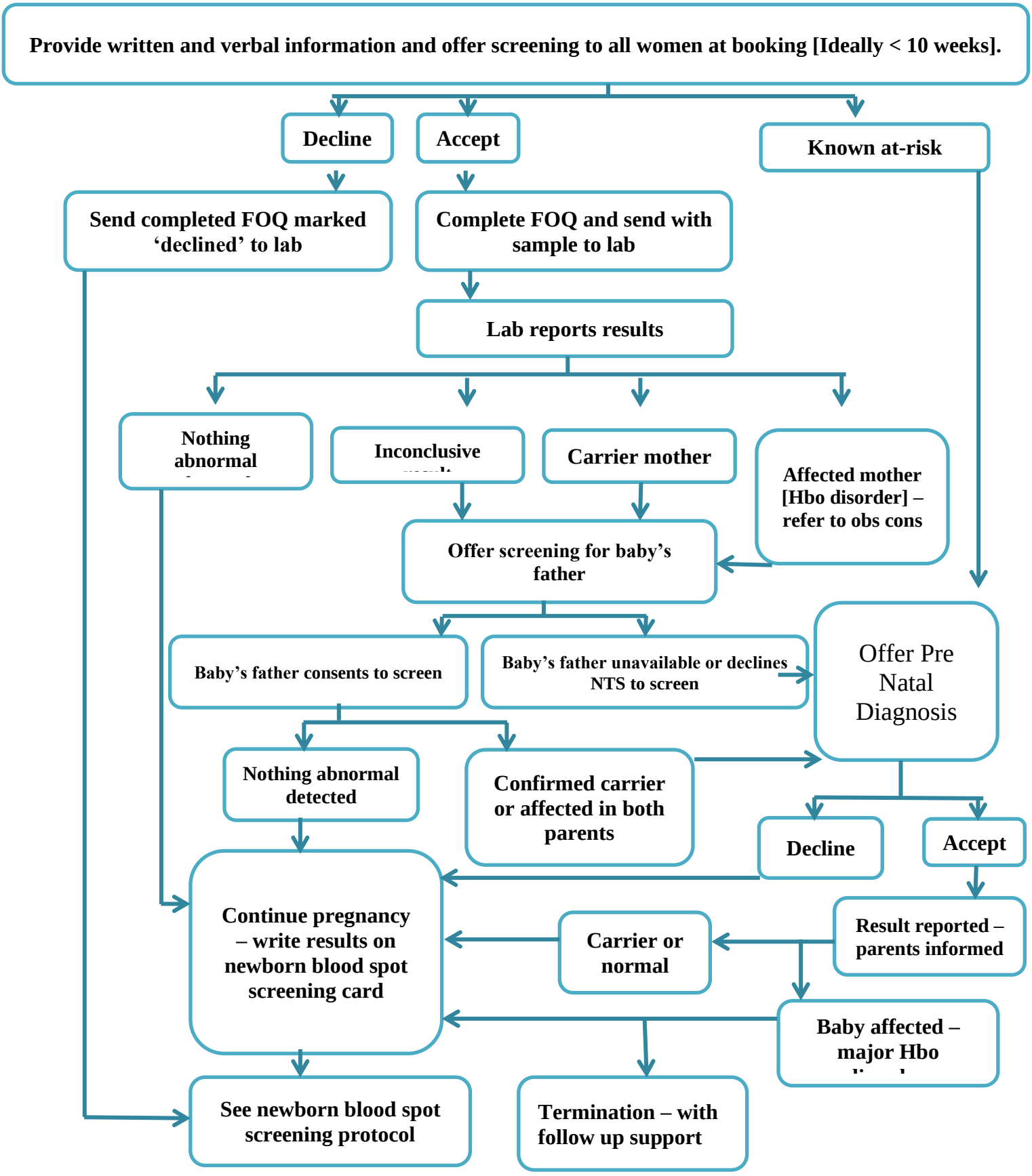
These disorders are mainly found in individuals who have originated from Africa, Asia, the Caribbean, the Middle East, and the Mediterranean, however because of migration and subsequent integration, they can affect any ethnic group.

25 Appendix B – Proforma for recording screening results at 10 days

BOOKING SCREENING RESULTS TO BE CHECKED 2 WEEKS AFTER TAKEN:
Clinic _____[surgery / children's centre]

<table>
<tr><td colspan="4">Name, DOB, ID number</td><td rowspan="9"></td><td></td><td>Accepted /declined</td><td>Result</td><td>Date result</td><td>MW checking</td><td rowspan="9">Comments: results missing/ repeat required/ decline form sent/ referrals made?</td></tr>
<tr><td>Gest at booking</td><td></td><td>MW at booking</td><td></td><td>Group & Rhesus</td><td></td><td></td><td></td><td></td></tr>
<tr><td rowspan="2">Date booked</td><td rowspan="2"></td><td rowspan="2">Date booking bloods taken</td><td rowspan="2"></td><td>Hb</td><td></td><td></td><td></td><td></td></tr>
<tr><td>Sickle and thal</td><td></td><td></td><td></td><td></td></tr>
<tr><td></td><td></td><td></td><td></td><td>Rubella</td><td></td><td></td><td></td><td></td></tr>
<tr><td></td><td></td><td></td><td></td><td>Syphilis</td><td></td><td></td><td></td><td></td></tr>
<tr><td></td><td></td><td></td><td></td><td>HIV</td><td></td><td></td><td></td><td></td></tr>
<tr><td></td><td></td><td></td><td></td><td>Hep B</td><td></td><td></td><td></td><td></td></tr>
<tr><td></td><td></td><td></td><td></td><td></td><td colspan="4"></td></tr>
<tr><td></td><td></td><td></td><td></td><td>Downs screen</td><td colspan="2">Accepted / Declined</td><td colspan="2" rowspan="2">Tick if transfer booking:</td></tr>
<tr><td></td><td></td><td></td><td></td><td>Anomaly scan</td><td colspan="2">Accepted / Declined</td></tr>
</table>

26 Appendix C - CARE PATHWAY: Antenatal Screening for Sickle Cell and Thalassaemia



27 Appendix D – Letter for Carrier Mums Inviting Dad for a Screening Test

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 (delete as appropriate: biological father/second biological parent) also carries a gene to make unusual haemoglobin. For this reason we are inviting you and your baby's (delete as appropriate: biological father/second biological parent) to an appointment to discuss your test result. At that time, we will offer the baby's (delete as appropriate: father/second biological parent) 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 (delete as appropriate: father/second biological parent). 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,

https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/422529/Letter_for_carrier_mums_and_inviting_dad_for_screening_test_V1.0_2013_07_23.doc

28 Appendix E- letter inviting baby's father for screening

Dear……

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,

https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/408715/Letter_inviting_dad_for_screening_test_V1.0_2013_07.23.pdf

29 Appendix F - BSUH Contacts

BSUH designated programme leads for maternity services
Midwifery: Karen Gregory– Antenatal screening co-ordinator
Obstetrics: Jo Sinclair Consultant obstetrician

Leads for laboratory
Lab Leads Alison Laverick(Senior BMS) and Sue To (Manager).
Quality Officer Helen Frost.
Pathology Quality Manager John McBride.

Antenatal Screening midwives:
Antenatal Screening Co-ordinator at BSUH [cross site]
Karen Gregory karen.gregory12@nhs.netMobile: 07876 357 423
Office: 01273 696955 X67477

Antenatal Screening Support Midwives [BSUH]

Mel Sanders [part time]
Melanie.sanders1@nhs.net
Screening Office: PRH: 01444 441881 X 5404
Screening Office RSCH: 01273 696955 X 67477 Trust shared antenatal screening NHS net email account
bsu-tr.antenatalscreening@nhs.net

Fetal Medicine Consultants at BSUH
Win Khine: w.khine@nhs.net Sec: Benedetto Lorusso X 64388

30 Appendix G - External Contacts

Cytogenetics [used by PRH and RSCH]
Cytogenetics Laboratory, Genetics Centre, Guys Hospital, 5th Floor Tower Wing, Great Maze Pond, London, SE1 9RT Tel: 020 7188 1709
Fax: 020 7188 1697

DEPARTMENT OF HAEMATOLOGICAL MEDICINE
South Thames Regional Centre for Prenatal Diagnosis of Blood Disorders
Red Cell Centre
Kings College Hospital, Denmark Hill, London SE5 9RS Tel: 020 3299 4337 (office)
020 3299 2265 (lab)
Fax : 020 3299 1267
E-mail : pnd@kch.nhs.uk
Website: kingspath.co.uk/

Public Health England leads the NHS Screening Programmes:

Screening Quality Assurance Service (South Regional Screening QA Team)
Public Health England South East
York House, Massetts Road,
Horley, Surrey, RH6 7DE

DD: 01138 248045
Fax: 01293 778888

https://www.nhs.uk/conditions/pregnancy-and-baby/screening-tests-abnormality-pregnant/

31 Appendix H - Support groups and further information

Antenatal Results and Choices [ARC]
Helpline: 0845 077 2290
E-mail: info@arc-uk.org
Web site: www.arc-uk.org

UK Thalassaemia Society [UKTS]
Telephone: 020 8882 0011
Web site: www.ukts.org
E-mail: office@ukts.org

Sickle Cell Society
Telephone: 0208961 7795 / 8346
Web site: www.sicklecellsociety.org

APOGI (Accessible publishing of genetic information)
www.chime.ucl.ac.uk/APoGI