# Sickle Cell & Thalassaemia - (Haemoglobinopathy) Screening in Pregnancy

University Hospitals of Leicester NHS Trust

**Trust ref: C58/2006**

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## 1. Introduction and Who Guideline applies to

This guideline is intended for the use of all Medical, Midwifery, Nursing, Primary Care and Laboratory staff involved in the care of pregnant women in both Primary and Secondary care settings.

**Background:**

These guidelines aim to provide all staff within the Community, Maternity Units and the Special Haematology Laboratory with guidance about the UK National Screening Committee – NHS Antenatal and Newborn Screening Programmes for Sickle Cell and Thalassaemia (2006). With reference to the guidance provided by the NICE Antenatal Care Guidelines (2003) and the NSF for Children, Young People and Maternity services (2004).

## 2. Screening;

- Leicestershire is a high prevalence area and thus all pregnant people booking with University Hospitals of Leicester NHS Trust will be offered screening for sickle cell and thalassaemia. This should be at the first point of contact with the pregnant person, whenever and wherever that is.

- All pregnant people and partners will be given appropriate verbal and written information to enable an informed choice to be made including the use of the information from the NSC "Screening tests for you and your baby".

- Pregnant people must be aware that identification of carrier status will enable the delivery of appropriate care to both mother and baby – this will usually include father testing and referral to appropriate health professionals.

- Screening must be undertaken as early as possible, ideally at 8-10 weeks gestation, in each pregnancy. For specific conditions to be screened for in this programme, see appendix 4.

- Further samples for sickle cell and thalassaemia screening must be sent in subsequent pregnancies to ensure appropriate partner testing.

- Screening may be initiated by any health professional, although it is the Community Midwife's responsibility to offer screening at the booking appointment.

- Documentation in the Maternity health records will be made as to whether the offer of the screening test is accepted or declined. The combined request form / Family Origin Questionnaire must be completed and submitted to the Lab in all cases.

- Interpreting services must be used where appropriate reference can be made to the "Accessing Interpreters policy". Pregnant people should also be encouraged to access the information provided about screening in different languages at - https://www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief

- All pregnant people will be informed that they will be contacted by the SCAT service if they are a found to be a carrier of a clinically significant haemoglobinopathy and an appointment will be offered to discuss the result and initiate testing of the baby's father.

### 2.1 Universal screening

- Universal antenatal screening for sickle cell and thalassaemia will be carried out in a consistent manner.

- All antenatal screening samples for Haemoglobinopathy screening must include a fresh FBC. This includes any repeat samples. Blood will be taken for FBC (1 x 4.9ml EDTA bottle – red top).

- Complete the FBC request form.

- Blood will be taken for Hb Electrophoresis (1 x 2.7ml EDTA bottles – purple top).

- The ideal mode of requesting an antenatal haemoglobinopathy screening test is by using the EFOQ within the Sunquest ICE system.

- If electronic access to the FOQ is not possible - complete the Leicestershire combined request form / Family Origin Questionnaire. Inclusion of the mother's NHS / Hospital number is mandatory but in exceptional circumstances, if no NHS / Hospital number is available address can be used.

- Ensure all information required is documented on the request forms.

- If the pregnant person is a known carrier document details on the request forms. A direct referral must be made to the SCAT team at booking or as soon as the carrier status is identified by emailing the pregnant person's details and relevant history to SCAT@uhl-tr.nhs.uk

- If there is a donor egg pregnancy this must be documented on the Family origin questionnaire

- If there is a donor egg pregnancy and the biological father is a known Haemoglobinopathy carrier a direct referral to the SCAT team should be made.

- Establish if current father has been screened and record their details on the form if consent given.

- If a pregnant person has already been screened for haemoglobinopathies in this pregnancy and is transferring their care to Leicestershire. Offer re-screening locally, if re-screening is declined – submit FOQ/EFOQ marked as declined "screened elsewhere" in the reason. Ideally include a hard copy of this result with the form.
    - Please note if donor egg or donor sperm has been used
    - Please note if adoption / unknown ancestry
    - Please note if bone marrow transplant (BMT)

- Ask for the ancestral origin when verifying the ethnic origin of both the parent's. This information is important when interpreting the result of the haemoglobinopathy screen. All family origins must be documented, it is expected that multiple answers will be supplied on the form in these cases.

- Put each sample and form in separate specimen bags or attach form to adhesive strip if grip bags used.

## 2.2 Negative results

Negative results will be sent by the laboratory to the requester

The presence of results will be checked at the next routine antenatal appointment after booking. (at 14-18 weeks gestation if possible).

Pregnant people will be informed that their screening test for sickle cell and thalassaemia is negative and that no further investigations are needed.

Negative screening results will be documented in the Maternity health records.

Variants of normal haemoglobin or 'possible alpha thalassaemia' may also be present, but not clinically significant. Pregnant people in these circumstances will be reassured that no further action is needed.

## 2.3 Clinically significant results

- All results that are clinically significant, including unidentified haemoglobin variants that are potentially clinically significant, (see appendix 4) will be emailed by the laboratory to the Sickle Cell and Thalassaemia (SCAT) Service and a report sent to the requester advising father screening.

- The SCAT Service will then invite the pregnant person to discuss the result. The appointment letter to the pregnant person will advise that they attend the appointment with the baby's father so that they can be offered a haemoglobinopathy screen.

- The SCAT Service will process and triage referrals offering an appointment to the pregnant person to attend for counselling within 5 working days if the gestation is ≥12/40, if the gestation is below 12/40 offer an appointment within 7 working days of receiving the emailed result from the laboratory

- Counselling and written or digital information about the implications of an abnormal result will be provided by the SCAT Specialist Nurse. The pregnant person will also be issued with a haemoglobinopathy card.

- The Midwife or Obstetrician who is giving the pregnant person her results will check that the pregnant person has been contacted by the SCAT Service, and if not a referral will be made.

- Consider referral to the East Midlands Maternal Medicine Network

- Where results are inconclusive the laboratory will notify the requesting clinician and where appropriate biological father screening will be advised. Follow up of the pregnant person' s blood test may be advised.

- The laboratory will update their record of antenatal carriers.

### 2.4 Specialist SCAT service

The Specialist SCAT Specialist Nurse will counsel the pregnant person about the implications of an abnormal result. The baby's father will be offered testing and if accepted should ideally be done by 10+6 weeks of pregnancy).

All carriers will be invited by letter and/or phone for counselling at the SCAT Service. The pregnant person will be advised to attend the appointment with the baby's father.

Fathers will be offered screening for haemoglobin disorders at this consultation or at a time convenient to the couple. (Ideally by 10+6 weeks gestation)

If the baby's father is unavailable for or declines a haemoglobinopathy screen, the SCAT Specialist Nurse will discuss with the pregnant person the risks to the fetus and the options which are available to them including prenatal diagnosis and capillary blood sampling at birth. If the pregnant person requests prenatal diagnosis the SCAT Specialist Nurse will make the arrangements as per appendix 2b.

If baby's father screening for sickle cell and thalassaemia is normal, the SCAT Specialist Nurse will advise the couple of a 50% risk the baby will be a carrier. The Newborn Screening Blood Spot Test will also be discussed.

For pregnant people failing to attend 2 clinic appointments with the SCAT Service a letter explaining the risks, a haemoglobinopathy card and an information leaflet are sent to the GP with a copy to the midwife to follow up. The Antenatal Screening Co-ordinator is also informed.

The SCAT Specialist Nurse will complete an Antenatal Summary for all antenatal pregnant people found to carry a significant haemoglobinopathy and send it to the Antenatal and Newborn Screening Coordinator (a copy is sent to the pregnant person's GP). This will be filed within the hospital maternity notes

### 2.5 Father of baby screening that suggests clinical risk

- If baby's father screening for sickle cell and thalassaemia suggests clinical risk to the baby the SCAT Specialist Nurse will contact the couple as soon as possible (usually within 3 working days). An appointment will be made to discuss the risk to their baby of having a major haemoglobin disorder and the options that are available to the couple including prenatal diagnosis (PND). (Ideally by 11+6 weeks gestation).

- The SCAT Specialist Nurse will discuss the results with the Consultant Haematologist, their deputy or Laboratory Lead (Haemoglobinopathy) to find out if samples need to be repeated and whether DNA samples need to be sent to Oxford regardless of PND decision.

- Counsel the pregnant person/couple about their haemoglobinopathy trait(s) and the calculated risk that the baby will inherit a major haemoglobinopathy.

- Give written information to reinforce this (e.g. Public health England Information and choices for women and couples at risk of having a baby with Sickle cell disease and APoGI) in the appropriate language. An interpreter should be present if appropriate

- The major haemoglobinopathy that the baby is at risk of inheriting (e.g. Sickle cell anaemia) should be outlined; including symptoms, management, and life expectancy

- Ensure that written information is also available

- Discuss in detail the pregnant person's options in relation to PND (chorionic villus sampling (CVS) or amniocentesis), termination (TOP) and capillary blood sampling.

**OPTIONS ARE:**

- Have prenatal diagnosis (CVS or amniocentesis) to find out the baby's haemoglobin status and to continue with the pregnancy even if the baby has a major haemoglobin disorder.

- Have prenatal diagnosis (CVS or amniocentesis) to find out the baby's haemoglobin status and consider termination if the baby has a major haemoglobin disorder

- If prenatal diagnosis is accepted, arrangements for this will be made by the SCAT Specialist Nurse as per appendix 2b. (Ideally prior to offer PND by 12+ 0 weeks gestation). The details of how to send the samples is in appendix 1

- If lady is being referred to fetal medicine for PND for Haemoglobinopathies from outside of UHL – refer to appendix 2a for the correct process.

- The SCAT Specialist Nurse will notify the pregnant person /couple of the prenatal diagnosis results within 4 days of receiving the result (providing parental genotype is not pending).

- Decline PND and have a capillary blood sample taken at delivery (in addition to Newborn screening) to find out the baby's haemoglobin status.

- If capillary blood sampling at birth is accepted, the SCAT Specialist Nurse will liaise with the appropriate health professionals as per appendix 3.

- Decline PND and capillary blood sampling and wait for the baby to be tested by the Newborn screening programme to find out the baby's haemoglobin status.

- The Newborn Screening Blood Spot Test will be taken as usual by the midwife in accordance with NSC guidance on Newborn Blood Spot Screening, with parental results documented on the form.

- It should be made very clear to the pregnant person/couple that it is their choice whether to proceed to PND or not.

- If a "hard copy" of the baby's father result is available, it will be filed with the pregnant person's records in the SCAT department.

- The SCAT Specialist Nurse will inform the Newborn Screening Laboratory in Sheffield and the Antenatal & Newborn Screening Co-ordinator of any baby which is at risk of being born with a major haemoglobin disorder.

- The SCAT Specialist Nurse will continue to support the couple.

### 2.6 Reporting screen positive samples

- Processes will be in place for the reporting of screen positive capillary samples; this process is in addition to the process of reporting and follow up of samples identified through the Newborn Screening Programme which is outside the scope of this document.

- The laboratory staff will inform the SCAT Service of the capillary blood result where performed.

- The SCAT Specialist Nurse will ensure the parents are informed of the baby's capillary blood sample results.

- Any babies who have a major haemoglobin disorder detected on the newborn screening blood spot test will be reported to the named paediatrician, SCAT Service and General practitioner directly by the screening lab at Sheffield in line with the National Screening programme reporting process (Newborn outcome System).

- The SCAT Specialist Nurse will facilitate paediatric referral and provide support to the parents of the affected infant.

- Where the mother's status antenatally was unknown this will be fed back to the antenatal screening co-ordinator for investigation.

- A Confirmation of the baby's diagnosis will be carried out in the paediatric clinic.

## 3 Education and Training:

Antenatal and new-born screening is part of a mandatory training day for all midwives

## 4. Monitoring Compliance

| What will be measured to monitor compliance | How will compliance be monitored | Monitoring Lead | Frequency | Reporting arrangements |
|---|---|---|---|---|
| Key performance indicators for ANNB screening | NHS England and PHE QA teams will monitor through the ANNB screening programme boards | | Quarterly | |
| Monitoring of Trisomy screening standards | NHS England and PHE QA teams will monitor through the ANNB screening programme boards | | Annually | |

## 5. Supporting references:


1. UK National Screening Committee – Sickle Cell and Thalassaemia Programme (2013).
2. NICE Antenatal Care Guidelines (2019).


## 6. Key Words

Antenatal, Electrophoresis, Sickle Cell and Thalassaemia Service (SCAT Service)

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**The Trust recognises the diversity of the local community it serves. Our aim therefore is to provide a safe environment free from discrimination and treat all individuals fairly with dignity and appropriately according to their needs.**
**As part of its development, this policy and its impact on equality have been reviewed and no detriment was identified.**

### EDI Statement

We are fully committed to being an inclusive employer and oppose all forms of unlawful or unfair discrimination, bullying, harassment and victimisation.

It is our legal and moral duty to provide equity in employment and service delivery to all and to prevent and act upon any forms of discrimination to all people of protected characteristic: Age, Disability (physical, mental and long-term health conditions), Sex,

Gender reassignment, Marriage and Civil Partnership, Sexual orientation, Pregnancy and Maternity, Race (including nationality, ethnicity and colour), Religion or Belief, and beyond.

We are also committed to the principles in respect of social deprivation and health inequalities.

Our aim is to create an environment where all staff are able to contribute, develop and progress based on their ability, competence and performance. We recognise that some staff may require specific initiatives and/or assistance to progress and develop within the organisation.

We are also committed to delivering services that ensure our patients are cared for, comfortable and as far as possible meet their individual needs.

| CONTACT AND REVIEW DETAILS | | | |
|---|---|---|---|
| **Guideline Lead (Name and Title)**<br>Haemoglobinopathies working party | | | **Executive Lead**<br>Chief Nurse |
| **Details of Changes made during review:** | | | |
| **Date** | **Issue Number** | **Reviewed By** | **Description Of Changes (If Any)** |
| September 2021 | 3.1 | **Maternity services governance group** | **Updated diagnosis pathway chart appendix 1** |
| October – December 2023 | 4 | **SCAT team, screening midwife, fetal medicine** | **Change of process for the management and requesting of PND's for women booked at UHL and externally.**<br>**Added consider referral to EMMMN** |
| March 2025 | 5 | **Helen Ulyett – Antenatal & Newborn Screening Co-ordinator** | Added<br>A direct referral must be made to the SCAT team at booking or as soon as the carrier status is identified by emailing the pregnant person's details and relevant history to SCAT@uhl-tr.nhs.uk<br><br>If there is a donor egg pregnancy this must be documented on the Family origin questionnaire<br><br>If there is a donor egg pregnancy and the biological father is a known Haemoglobinopathy carrier a direct referral to the SCAT team should be made. |

## Appendix 1 – Sending PND sample to Manchester.

# Supporting Information for The Sickle cell & Thalassaemia Prenatal Diagnostic Testing Pathway

All SCT Prenatal diagnostic testing is completed in Manchester Genomics Lab. Samples from procedures in UHL need to be taken to Cytogenetics Lab at LRI as they have an established direct PND pathway for urgent obstetric SCT samples from Leicester to Manchester Genomics Lab. (This bypasses Cambridge regional Genomics Hub route that non obstetric samples must follow).

All forms will be prepared by local team requesting PND – In Leicester this is Leicester SCT CNS Team outside of Leicester referrals accepted by Fetal Medicine teams will come from Local Antenatal Screening Midwives/ Coordinating teams.

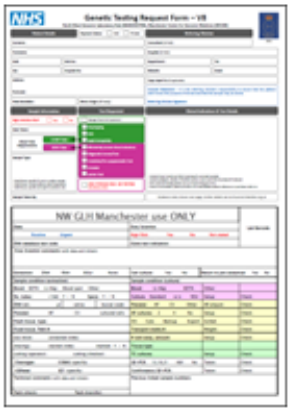
All paperwork referral forms and blood results required will be emailed by local team to the FM shared email and will require printing by FMM.

The Paper work will include

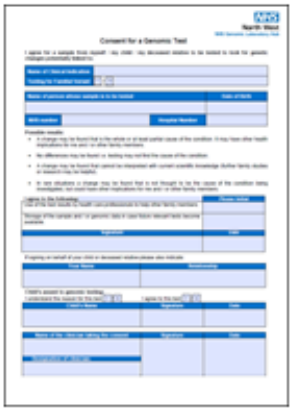
1. Manchester Genetic Testing V8 Form which will be completed by local team bar the sample type, sample taken by and fetal gestation which will require completing at time of prenatal procedure by FMM.
2. FMM to complete the usual form for cytogenetics as per any CVB sample.
3. The local team will complete a Rare and Inherited Disease Referral Request form for each Parent as this will accompany the fresh parental blood samples that are required to be taken on the day of the procedure by FMM which will be sent with the fetal sample.
4. The local team will complete a Consent for Genomic blood test form for each parent as they take consent for the parental blood samples (each parent should sign the genomic blood test consent forms locally in advance of booking PND and completed forms sent via email with the rest of the paperwork)
5. The local team will prepare a copy of each parent's screening results which would include the locally reported Haemoglobin Screen (HPLC) and Full blood count.
6. The FM Consultant will take consent for the procedure on the day of the procedure.

Paperwork 1-5 will need to be included with fetal sample and minimum of 5mls EDTA blood sample from each parent and taken to Cytogenetics Lab via porter – same as all other PND CVB samples.

Results will go back to Local requesting team to deliver to the Mother/couple.







Key to abbreviations : SCT – Sickle Cell & Thalassaemia, CNS – Clinical Nurse Specialist
FMM - Fetal Medicine Midwife, FMC - Fetal Medicine Consultant

Supporting Information Produced September 2023 Sarah Barnes SCT CNS Leicester 0116 2586081

## Supporting Information for The Sickle cell & Thalassaemia Prenatal Diagnostic Testing Pathway

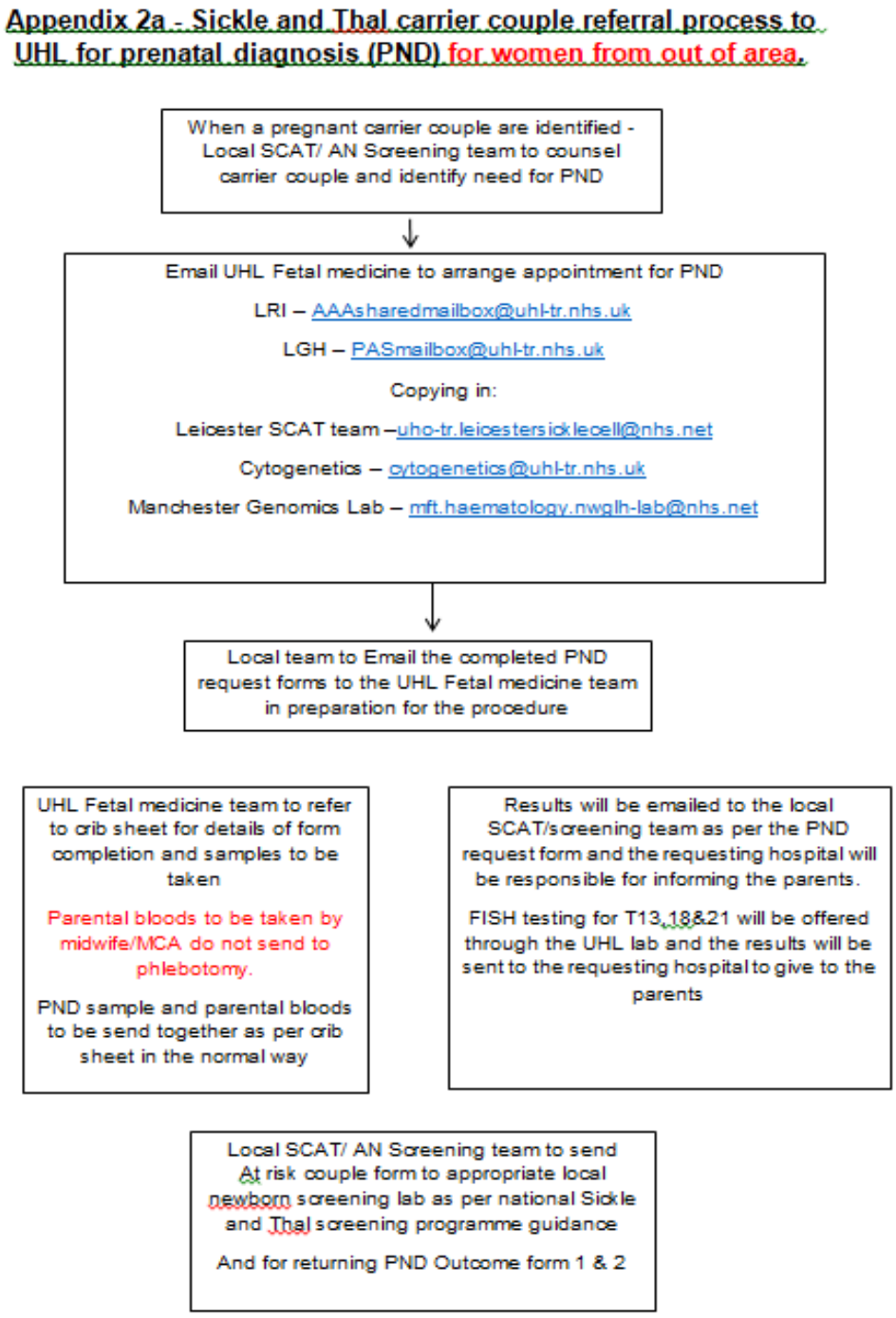
### Visual Check List before sending to Cytogenetics Lab at UHL



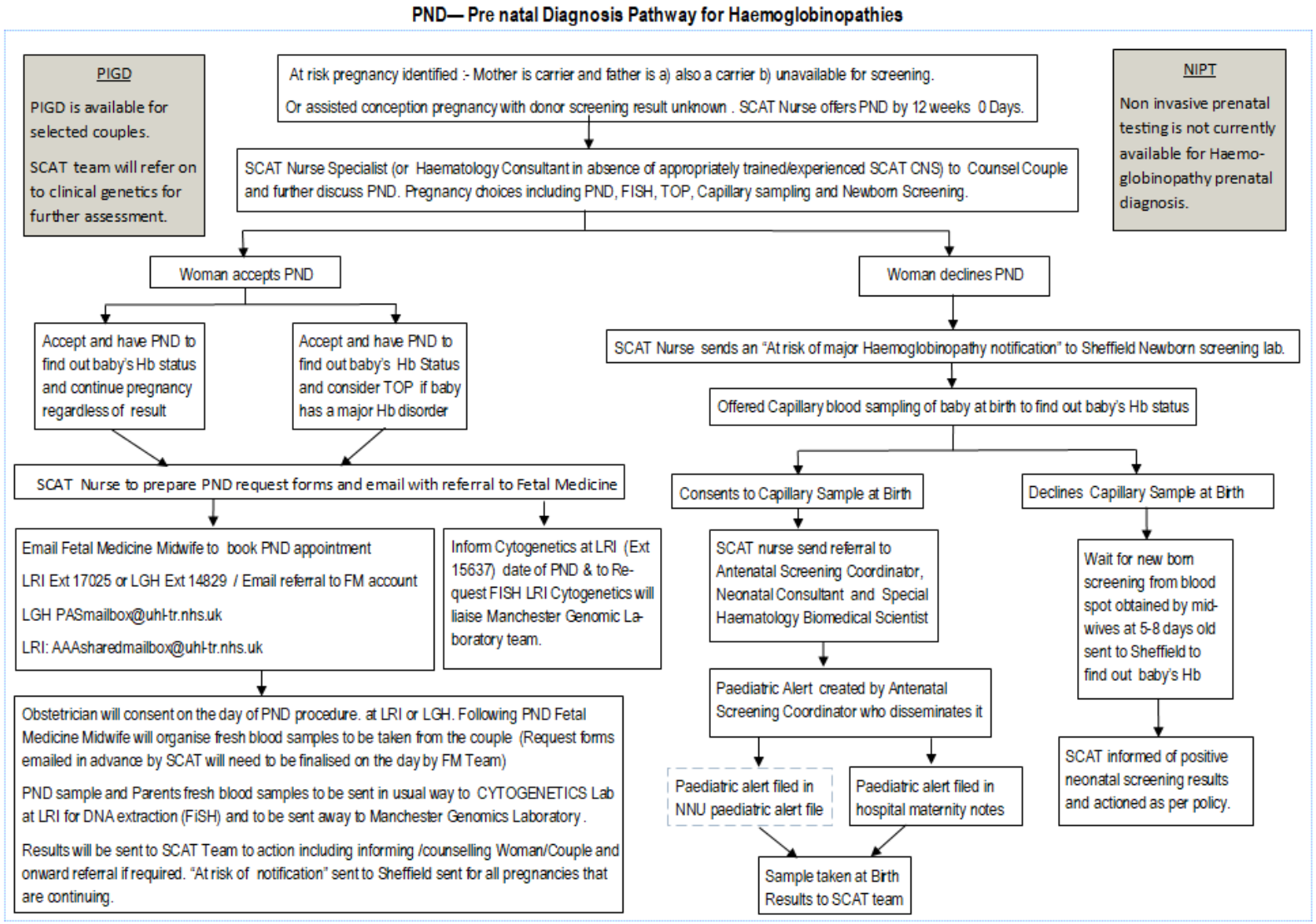
Key to abbreviations : SCT – Sickle Cell & Thalassaemia, CNS – Clinical Nurse Specialist
FMM - Fetal Medicine Midwife, FMC - Fetal Medicine Consultant

Supporting Information Produced September 2023, Sarah Barnes SCT CNS Leicester 0116 2586081

## Appendix 2a: Sickle cell & thalassaemia carrier couple PND referral process

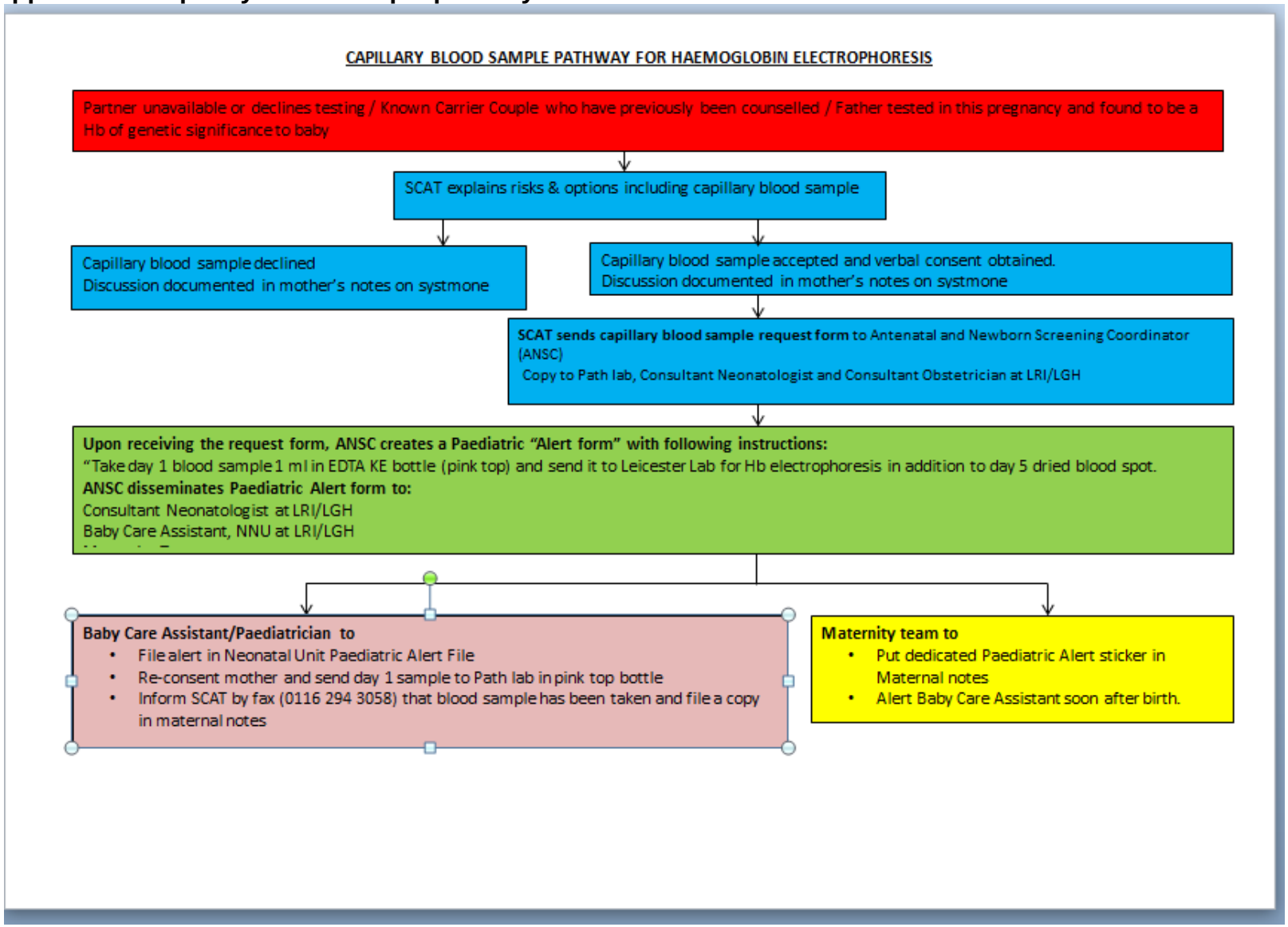


## Appendix 2b - PND pathway for Haemoglobinopathies for pregnant people booked at UHL




SCAT- Sickle Cell and Thalassaemia PND - Pre Natal Diagnosis. NIPT- Non invasive prenatal testing. Hb - Haemoglobinopathy TOP - Termination of pregnancy. FISH - Florescent in situ Hybridization PIGD - Pre Implantation Genetic diagnosis. Rev Aug 2023

## Appendix 3: Capillary blood sample pathway



## Appendix 4: Conditions to be detected by the Antenatal screening programme.

Conditions to be detected as part of the antenatal screening programme

i. Significant maternal haemoglobinopathies; these should be detected by antenatal screening and are important for maternal care
   - HbSS
   - β thalassaemia intermedia Hb H disease
   - (β thalassaemia major would
   - be clinically apparent)

ii. Maternal conditions requiring baby's father testing

a) Conditions in (i)

b) Carrier states in mother

| Potential significant disorders in the fetus |
|---|
| Hb-SS Hb-SC |
| Hb-SD Punjab |
| Hb-SE $_{Arab}$ |
| Hb-SO |
| Hb-S/Lepore |
| Hb-S/β thalassaemia Hb-S/δβ thalassaemia Hb-β/Lepore $^{o}$ $_{o}$ |
| Hb Bart's HF (α /α) |
| β thalassaemia major (except cases with silent or near silent maternal phenotype) |
| Hb E/β thalassaemia |

| Carrier states in mother |
|---|
| Hb – AS |
| Hb-AC |
| Hb-AD Punjab |
| Hb-AE $_{Arab}$ |
| Hb-AO |
| Hb-A Lepore |
| β thalassaemia trait |
| δβ-thalassaemia trait $^{o}$ |
| α thalassaemia trait |
| HPFH |

c) Any compound heterozygote state including one or more of the above conditions
d) Any homozygous state of the above conditions.