

MATERNITY GUIDELINES

Haemoglobinopathies; Sickle cell and thalassaemia

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1. Haemoglobinopathies

Haemoglobinopathies are common in people who have family origins from the malarial parts of the world; and in the UK are seen particularly amongst minority ethnic groups from Africa, the Caribbean, the Mediterranean, South East Asia, the Middle East, and the Far East but can be found (less frequently) in all ethnic groups. There are about 1000 recognised haemoglobin gene variants with varying degrees of pathology. Some people can have more than one gene variant.

2. Statement

Antenatal screening identifies pregnant women who are genetic carriers of sickle cell, thalassaemia or other haemoglobin disorders. People who are carriers are healthy and are unaware of their carrier status unless they have a specific blood test. Where both parents are carriers, there is a one in four (25%) chance that their baby could inherit a condition that requires treatment. For this reason, screening needs to be offered early in pregnancy.

The aim of antenatal screening is therefore to:

- Identify genetic carriers so parents have information about the risks of having a baby with a major haemoglobin disorder. Support parents to make informed choices about their pregnancy including tests for the fetus; carrying on with an affected pregnancy and preparing for the care of their infant; or termination of the pregnancy as an alternative option

This is a linked programme to **Newborn Bloodspot Screening (heel prick)** test that identifies babies who have suspected sickle cell disease. It may also detect babies who are genetic carriers of some haemoglobin variants. The key reason for offering newborn screening is that babies with sickle cell disease are vulnerable to life-threatening infections. By identifying them promptly after birth, they can be offered potentially life-saving penicillin, and be referred for specialist care.

Plymouth and the surrounding area is a low prevalence area, selective testing is offered based on family origin. This means that all women need to be asked what they consider their and the baby's fathers family origin to be.

3. Local policy

- To use the national family origin question (FOQ) to identify women at risk of a haemoglobin variant. A locally produced specific form based on the UK National Screening Programme recommendations is used. This will ensure that every pregnant woman has an FOQ with her booking FBC sample. If the woman accepts screening and the questionnaire shows that there is a risk of either parent being a carrier, a haemoglobin Electrophoresis blood test will be performed.
- **All** pregnant women booking with Plymouth Maternity services are eligible for Sickle cell and Thalassaemia screening, including women moving to the area at any gestation. In this case evidence of any previous screening should be documented if booking bloods are not taken at the booking. If blood for sickle cell and thalassaemia screening are not taken at booking, the screening team should be informed of any decline or omission.

4. The Process

This begins at the first point of contact usually the Community midwife who is responsible for:

- The provision of the information leaflet: *Screening tests for you & your baby* should precede the offer of screening.
- The discussion of family origin with every antenatal woman using the Family Origin Questionnaire
- Informing all women that routine analysis of blood will identify the risk of being a thalassaemia carrier.
- Documenting that the information has been received, understood and consent has been given to proceed.
- If the offer is declined the form must still be completed. Documenting the reason for declining is not obligatory but any comments may be documented and helpful.
- If this is a subsequent pregnancy and the laboratory has issued a haemoglobinopathy card further screening is not necessary but the family origin questionnaire must still be completed stating that the result is available.
- If the woman has already been tested please identify that the father of the baby is the same.

The family origin questionnaire must be completed with both parent's information and appropriate documentation about paternity in order that the sample can be processed by the laboratory. The laboratory will identify any family origin questionnaires found to be incomplete and the Screening team will follow it up in order to complete the screening.

All women identified as high risk on FOQ are entered into the Sickle cell and Thalassaemia database

All yellow copies of the FOQ, for high & low risk are kept alphabetically filed until confirmation that a copy has been received in the laboratory.

Evidence of this happening is provided by the retrospective monthly matching report compiled by the laboratory. Any omissions are followed up by the screening team which acts as the current failsafe along with the monthly booking spread sheet which has the results for every woman booked documented.

5. The Test

If the woman has been identified as high risk on the FOQ, booking bloods should be taken and sent as early in pregnancy as possible. The standard is for the **bloods to be taken by 10 weeks gestation**. This is to allow choice for the parents if their baby is at risk of a major haemoglobin disorder as they need time to allow for further tests, counselling and may wish to choose to end the pregnancy in line with religious or cultural beliefs.

All women including late bookers or transfers in must have an FOQ completed with a FBC sample.

Send an EDTA purple top bottle for haemoglobinopathy screening; identify this request on the form.

Women may also request to be tested pre conceptually. This can be arranged by the screening team or GP.

6. Testing the Father

The midwife should discuss the offer of screening for the father of the baby in the eventuality that the woman is screened positive.

Communication and the provision of information should precede the offer of screening.

The Screening Midwife will liaise with the Community midwife to make arrangements for the test to be performed.

The father can have his blood taken by the midwife, the GP, or at the hospital by the screening team. The father's sample should be clearly labelled; the combined laboratory form completed stating who he is the partner of, marked HBE in the other tests box and for the attention of the named lead laboratory Biochemist Des Rogers.

7. The Results

When the woman attends for the 1st Trimester Scan the complete results of her booking bloods will, if available, be given to her and filed in her notes.

The Screening team will follow up any missing, inconclusive or incomplete results on one occasion and send them to the woman to be filed in her notes. Any missing results need to be followed up or repeated at the 16 week appointment by the Community Midwife or at her next Community Midwife appointment

All results will be available to the requestor.

8. Negative Results

Those women who are found to have no evidence of a haemoglobinopathy will be reported as normal.

An appropriate letter and Green card issued from the lead Haematologist Dr Mike Hamon will be sent to the General Practitioner (GP) for issuing to the woman and/or the father of the baby.

9. Carrier results

Because the woman has been found to be carrier of a haemoglobinopathy, the Community midwife will be notified by the Screening team that the father should be offered screening.

The GP will be sent information specific to the inherited trait and an amber haemoglobinopathy card to confirm the result

This information is given to the woman at an appointment with the GP.

If the father declines the test or is absent please inform the Screening team.

The standard is now to test the same partner in a subsequent pregnancy as confirmation of the result.

The woman will be identified as "at risk" in this instance and offered the same choices as a couple at risk.

10. Identification of a high-risk couple

The couple should promptly be offered counselling from a professional who has attended a recognised haemoglobinopathy counselling course. This may be one of the screening team, a haematologist or both dependant on the needs of the woman and her family. Any consultation should be documented and placed in the woman's notes.

Arrangements prior to the appointment will include translator if available or use of the Big Word service.

The couple will be referred to a Fetal Medicine Consultant at the next available appointment for further counselling if prenatal diagnosis is appropriate. Diagnosis will include DNA sampling using chorionic villus sampling or amniocentesis.

Some couples will already have been identified as carriers from a previous pregnancy or from having requested a carrier test. It is important to offer these couples the option of PND early in their pregnancy if this is their wish. Primary care is likely to play an important role in this process and can make a direct referral to a specialist counsellor or fetal medicine specialist, rather than waiting for the routine booking process.

11. Linkage with Newborn screening

All significant antenatal carriers need to be alerted to the Newborn screening laboratory in Bristol, the screening team co-ordinates the communication using the regional alert form. This will be documented on the SC&T Carrier log spread sheet.

A neonatology alert will also be generated by the Screening team for at risk couples continuing the pregnancy.

Any at risk couples will be included within the MDT spreadsheet and care planned within the MDT environment as there is a neonatologist present.

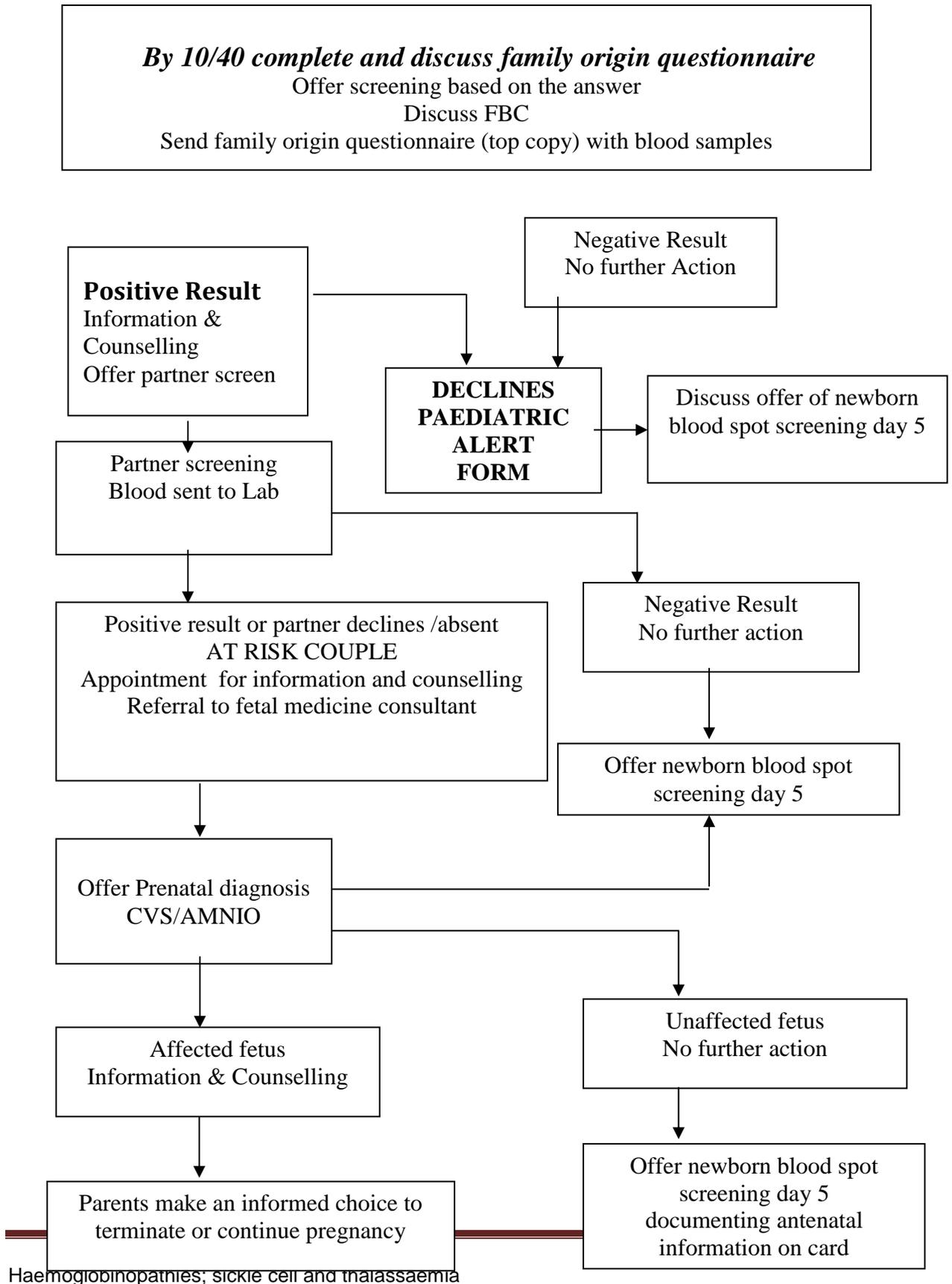
Postnatally when undertaking the newborn bloodspot the midwife should document on the bloodspot form any known carrier status for mother or father.

12. Failsafe arrangements

Each month the laboratory team provide a report which matches each pregnant woman with evidence of a completed FOQ.

The Screening team will follow up any missing FOQs each month with a request for completion going out to the community midwifery teams for completion. As part of the failsafe the yellow copy of the FOQ form is kept until a result is available on the hospital blood results system

ANTENATAL SCREENING PATHWAY



PROCESS FOR COMPLETING FAMILY ORIGIN QUESTIONNAIRE

The following Flow Chart is provided as guidance for using the family origin questionnaire for midwives and other health care professionals providing antenatal care for pregnant women.

Provide ALL women with information about Sickle Cell & Thalassaemia Screening Programme.
Are translation services **required**?

Midwife to complete Questionnaire
Ask ALL women about their family origins and their baby's father's family origins

If ticks in shaded boxes for mother and/ or baby's father explain to woman that they will be offered screening for haemoglobin variants

Consider & discuss whose baby will require Neonatal BCG
Document and identify in handheld notes & write on FOQ discussed and recommended.

Take blood sample or give woman forms for phlebotomist use purple top EDTA bottle
(**all** samples sent to lab **MUST** have a top copy of questionnaire enclosed with antenatal booking blood sample)

Laboratory **WILL NOT** test sample if not accompanied by the correct forms and **YOU** will have responsibility to get forms thus delaying screening test.

One duplicate/pink copy to go in 'hand held' maternity notes
Third copy /yellow copy sent in to Screening team level 6

Monitoring and Audit

See audit tool

Auditable standards:

Reports to:

CDS RM Committee
Clinical Governance & Risk Management Committee

Frequency of audit:

3 yearly

Responsible person:

Antenatal screening midwife

Cross references

Guidelines can now be found on the network share (drive)
G:/DocumentLibrary/UHPTClinicalGuidelines/Maternity

References

Information for Health care Professionals NHS Sickle Cell and Thalassaemia Screening Programme 2012

Standards for the linked Antenatal & Newborn Screening Programme. **NHS Sickle Cell & Thalassaemia Screening Programme** November 2011

Antenatal care for uncomplicated pregnancies (CG62) NICE 2013 updated March 2016

Department of Health (2000) **The NHS Plan: a Plan for Investment, A Plan for Reform.** London: The Stationary Office.

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