Haemoglobinopathy Guidelines

Haemoglobinopathies are a group of inherited conditions that affect globin chain production. They fall into two main categories – haemoglobin variants, in which abnormal *forms* of globin chain are produced, eg sickle cell disease which causes anaemia and sickle cell crises; and thalassaemias, in which an abnormal *amount* of globin chain is produced resulting in severe anaemia.

NHS Sickle Cell and Thalassaemia (SCT) Screening Programme

The NHS SCT screening programme aims to identify people who are affected by or are carriers for sickle cell, thalassaemia, and other haemoglobinopathies. Screening is offered to all pregnant women and all newborn babies (as part of the newborn blood spot screening).

Antenatal screening

Antenatal screening occurs as early as possible, ideally by 10 weeks. Written information is provided and consent obtained. Scotland uses a low prevalence screening policy and if screening is accepted, all pregnant women will be screened for thalassaemia trait, using red cell indices. Additionally, a Family Origin Questionnaire is completed to assess risk of the mother or father being a carrier for sickle cell or other haemoglobin variant, in order to identify those who need further testing.

If the mother is found to be a carrier or affected by a significant haemoglobinopathy, the baby's father will be offered screening.

Couples identified as being high risk from antenatal screening will be offered counselling and diagnostic tests.

Postnatal screening

The Newborn Bloodspot Screening Programme in Scotland includes postnatal screening for phenylketonuria, congenital hypothyroidism, cystic fibrosis, medium chain acyl-CoA dehydrogenase deficiency, and sickle cell disorder. It will also identify carriers of sickle cell or other haemoglobin variants. Newborn screening for thalassaemia is not recommended by the UK National Screening Committee; however, the newborn blood spot test does detect most cases of beta thalassaemia major. It does not detect babies who are thalassaemia carriers; these patients do not require early diagnosis or treatment.

Screening will be offered and written information provided to parents of all newborn babies in Scotland. Written consent or refusal of each test should be obtained. If screening is accepted, a sample is obtained on day 5 in the form of a newborn bloodspot card and is sent to the Scottish Newborn Screening Laboratory (SNSL) in Glasgow.

Author: Kathleen Duffin ST Paeds, Susan Baird Cons Paed Haematology
Implementation date: 1 3 16 Review Date: 1 3 26

Actions

High risk patients (parents both carriers of sickle cell or other haemoglobinopathy, or mother carrier and partner status unknown)

- These patients will be identified antenatally by the obstetric/haematology team, and should have a "Special Features" entry on maternal Trak.
- Baby will be screened with day 5 newborn blood spot.
- They should NOT routinely have cord bloods or day 1 venous bloods for FBC or haemoglobinopathy screen. There is no indication for early testing, and diagnosis is more reliable if samples are processed centrally at the SNSL.
- Neonatal team to notify Dr Susan Baird* (paediatric haematology consultant, RHSC or in her absence Dr Angela Thomas) via email when these patients deliver. Email should include mother's name and DOB, baby's name, DOB, CHI and hospital number.
- Dr Baird will notify newborn screening lab of births, and results will be sent to her directly to allow timely communication of results to high risk couples.

One parent carrier of sickle cell or other haemoglobinopathy

- Baby will be screened with day 5 newborn blood spot.
- They should NOT routinely have cord bloods or day 1 venous bloods for FBC or haemoglobinopathy screen.
- Notification of the paediatric team is not required for these babies as they are not at high risk of a serious haemoglobin disorder. Carrier results from newborn blood spot will be communicated via the Health Visitor or GP.
- Neonatal team should advise family to contact the Health Visitor or GP in 6-8 weeks time for results. Discharge letter should be sent to inform GP.

One parent carrier of beta thalassaemia or alpha thalassaemia trait

- Carriership will not be detected on newborn blood spot screening and no specific testing is required.
- If parents request testing, advise them to visit their GP. This should not be undertaken before six months of age. Testing should preferentially occur when blood tests are being performed for another reason.

This guideline has been developed in collaboration with the Paediatric Haematology team, Obstetric Haematology Team, and Newborn Screening Coordinator. *Dr Baird – susan.baird@nhslothian.scot.nhs.uk

https://www.gov.uk/guidance/sickle-cell-and-thalassaemia-screening-programme-overview