NHS

Screening Programmes
Sickle Cell and Thalassaemia

Protocol for Reporting newborn screening results for sickle cell disease to parents

April 2012

Introduction

This protocol aims to clarify reporting arrangements for three types of newborn sickle cell screening results: Condition not suspected results, carrier results and screen positive results for sickle cell. The protocol has been developed with the Newborn Bloodspot screening programme (NBSP) and the Sickle Cell and Thalassaemia (SCT) Screening programme and is informed by a recent HTA report[1]; the SCT programme response to the HTA report (http://sct.screening.nhs.uk/cms.php?folder=2420) and the Map of Medicine (MoM) pathways describing the linked antenatal and newborn screening pathway[2]. It should be noted that it is important not to consider a sickle cell carrier result as a screen positive result.

Principles for reporting newborn screening results

- National protocols developed about communication of newborn genetic carriers should be broadly consistent with the protocols for communicating adult carrier results and in particular communicating sickle cell or thalassaemia carrier results to pregnant women.
- People prefer to be informed about results by someone who is well informed and can answer questions about the conditions rather than by someone they know but who is not well informed about the conditions[1].
- Parents should be informed of all newborn bloodspot screening results, with results recorded in the Personal Child Health Record when a carrier result is given.
- Information on the mother's SCT carrier status should be linked with the system of communication to parents about all newborn screening results.
- There needs to be sensitivity over the possibility of screening results revealing non-paternity. The screening programme has issued guidance on raising issues of non-paternity at http://sct.screening.nhs.uk/labupdates
- Newborn laboratories should use newborn status codes and linked text when reporting screening results
- The use of nationally produced parent information leaflets should be the norm to ensure standard messages.
- The Human Genetics Commission report on preconception genetic testing and screening (April 2011) recommends that carrier status information obtained incidentally through tests or investigations carried out for other purposes (for example after prenatal diagnosis or through newborn screening) should be provided to GPs and stored in a secure and accessible format [3]

Issuing laboratory reports of screening results.

The SCT Laboratory Handbook provides detailed guidance on reporting analytical data to facilitate reporting results in a standardised manner [4].

All "condition not suspected" and carrier Sickle Cell results: Laboratories are responsible for sending all screening results to Child Health departments or their equivalent in a timely manner using appropriate status codes. Further details of laboratory reporting are found in the Laboratory handbook [4]

Screen positive results: The present situation of newborn screening laboratories reporting screen positive sickle cell results to counsellors, practitioners or clinicians and other local reporting routes works well in providing support and choice to parents. This system does not, however, give a robust failsafe system to ensure these babies enter the care pathway. The report of the Paediatric Peer Review identified that some centres do not have processes to check that all babies enter the care pathway (West Midlands Quality Review Service, 2011) [5]
http://www.wmqi.westmidlands.nhs.uk/wmqrs/review-programmes/view/sickle-cell-and-thalassaemia-children-

The report recommended "The NHS SCT programme should ensure that newborn screening laboratories communicate details of affected baby to the named lead of the specialist team as well as to community nursing teams. Close liaison between the screening programme and commissioners on precise configuration of clinical networks will be needed to achieve this"

The National Haemoglobinopathy Project produced a guide to effectively commissioning high quality sickle cell and thalassaemia services, published in March 2012. (http://sct.screening.nhs.uk/haemcommissioning)

This contains mandatory standards on the links between screening and care. To support this work the SCT programme is working with laboratories and clinical networks to develop an additional reporting loop so that screen positive sickle cell results are also reported by the newborn laboratory to the designated specialist network centre, as well as the local centre. This will act as a failsafe as the designated specialist centre will be responsible for ensuring screen positive babies enter the care pathway but still allows the present system of offering local care provision and choice to parents to continue. The likely networks of clinical care are listed in the care guidelines: Sickle cell disease in childhood: Standards and guidelines for clinical care, October 2010[6] and given in Appendix One.

Method of informing parents of screening results

It is recommended that parental SCT results should be reviewed before reporting all newborn bloodspot result to assist in the effective communication of information. At present this is practically very difficult and time consuming without effective linkage across the antenatal interface. As a pragmatic response therefore parental results only need be reviewed before discussing screen positive or carrier results with parents.

All "Condition not suspected" results: The agreed method for reporting these screening results (when all five conditions screened for by the bloodspot programme are not suspected) is to send a letter to parents. A template for such a letter was developed as part of a pilot study in 2008 (available at http://newbornbloodspot.screening.nhs.uk/getdata.php?id=11006).

Sickle Cell Carrier results: Reporting carrier results has significant resource implications which have different challenges in high prevalence and low prevalence areas for sickle cell disease. The HTA report identifies that some parents prefer a face to face conversation whereas parents who know they are carriers are happy to receive a letter with an offer of an appointment to discuss the result.

As a pragmatic response the programme recommend the ideal method for reporting SCD carrier results is a face to face conversation but given the resource implications we suggest a letter or telephone call with a leaflet (translated if appropriate) and an offer of a follow up appointment is used as an interim solution until resources allow face to face reporting . A template letter is available in Appendix Two of this document. Two leaflets for parents have been developed by the programme:

- Information for mums and dads: your baby carries a gene for sickle cell
- Information for mums and dads: your baby carries a gene for unusual haemoglobin

These leaflets can be downloaded from http://sct.screening.nhs.uk/leaflets

While the Programme recommends that carrier results should be reported to parents, we do not specify who should do this. It does not need to be a specialist nurse. It is important that carrier result reporting does not delay reporting of major haemoglobinopathy results by specialist nurses. Specialist (or trained) health visitors are in post in some, but not all, parts of the country to report carrier results to parents. In all cases these discussions should make clear "up front" that the baby is well and does not have a "disease" and include informing parents of the screening results for the other conditions screened for.

Sickle cell screen positive results: The present situation of newborn screening laboratories reporting screen positive sickle cell results to counsellors, practitioners or clinicians and other local reporting routes works well in providing support and choice to parents. The SCT programme are developing an additional reporting loop so that screen positive sickle cell results are also reported by the newborn laboratory to the designated specialist network centre, as well as the local centre. This will act as a failsafe as the designated specialist centre will be responsible for ensuring screen positive babies enter the care pathway whilst allowing the present system of offering local care provision and choice to parents to continue.

The likely network centres are listed in the care guidelines: Sickle cell disease in childhood: Standards and guidelines for clinical care, October 2010[6] and given in Appendix One. The National Haemoglobinopathy Project (http://sct.screening.nhs.uk/cms.php?folder=2558) has developed a commissioning specification for haemoglobinopathy care. The commissioners will be required to designate network centres.

It is desirable that all screening results are recorded in primary care.

Written materials

Pre-screening information: pre-test information should be offered to parents. This can be given postnatally but it is optimal if is given antenatally (especially if either parent is known to be a carrier) emphasising the genetic and lifelong nature of the information. Pre-test information is available in the leaflet "Screening tests for you and your baby" and "Screening tests for your baby". These are also available in 18 languages on the NSC website http://www.screening.nhs.uk/languages.

All "Condition not suspected" results: A template for a results letter was developed as part of a pilot study in 2008 (when all five conditions screened for by the bloodspot programme are not suspected) and is available at http://newbornbloodspot.screening.nhs.uk/getdata.php?id=11006.

Sickle cell carrier results: The use of simplified carrier leaflets available in English and other languages is recommended when reporting carrier results. Two leaflets for parents have been developed by the programme:

- Information for mums and dads: your baby carries a gene for sickle cell
- Information for mums and dads: your baby carries a gene for unusual haemoglobin

These leaflets can be downloaded from http://sct.screening.nhs.uk/leaflets

These leaflets take account of recommendations in HTA report in terms of simplification of language, the need for accessible materials etc. These leaflets are available on the SCT website in Bengali, English, French and Urdu, and also available in hard copy in English.

In addition the SCT programme has drafted a standard carrier letter which can be used, alongside existing local processes to communicate about all newborn haemoglobin variant carrier results (Hb S, C, D and E) (see Appendix Two)

Screen positive babies: A book for parents of babies with sickle cell disease is available at http://sct.screening.nhs.uk/cms.php?folder=2503. Fact sheets about sickle cell and thalassaemia (created using cartoon-imagery and very simple, yet informative language) are available in English, Bengali, French and Urdu, as well as on audio. These are also available on the website.

National guidance for counselling about issues of non-paternity, to support those who counsel and visit parents to assist in standardised information giving is available at http://sct.screening.nhs.uk/labupdates#fileid11468 .

Training

A one day accredited training programme has been developed for those who are involved in reporting newborn sickle cell screening carrier results. The course will be run by the School of Nursing and Midwifery, Kings College, London (*http://sct.screening.nhs.uk/externaltraining*) with the first course in April 2012. This course is aimed at any health professional giving newborn carrier results.

Issues not addressed in this protocol

- The role of primary care in both communication of results and receiving information for future use. Further work on coding and transfer of results is in development.
- Screening for thalassaemia, as well as unidentified haemoglobin variant carriers in the newborn period.
- Achieving effective linkage of antenatal results with newborn results remains elusive despite the evidence of beneficial effects in reporting newborn screening results.
- Guidance on cascade testing policy (testing should be offered and available if parents wish to be tested and accompanied with appropriate counselling). This is good practice in genetics but to date the programme has no formal policy in this area.
- How and when to offer -children who are carriers information and counselling about their future "reproductive choices".

References

1. HTA report into reporting carrier results: Kai J, Ulph F, Cullinan T, Qureshi N. Communication of carrier status information following universal newborn screening for sickle cell disorders and cystic fibrosis: qualitative study of experience and practice. *Health Technol Assess* 2009;**13**(57) available at http://www.hta.ac.uk/project/1510.asp
2. The overall screening pathway including reporting of results is described in the Map of medicine pathways at http://eng.mapofmedicine.com/evidence/map/linked_sickle_cell_and_thalassaemia_screening1.html and http://eng.mapofmedicine.com/evidence/map/newborn_blood_spot_screening1.html
3. Human Genetics Commission, Increasing options, informing choice: A report on preconception genetic testing and screening. 2011. http://www.hgc.gov.uk/UploadDocs/DocPub/Document/Increasing%20options,%20informing%20choice%20-%20final.pdf
4. NHS Sickle Cell and Thalassaemia Screening Programme, Handbook for Laboratories. 2009. http://sct.screening.nhs.uk/cms.php?folder=2493#fileid10756

5. UK Forum on Haemoglobin Disorders, Services For Children And Young People With Haemoglobin Disorders Peer Review Programme 2010-2011: Overview Report. 2011.

6. NHS Sickle Cell and Thalassaemia Screening Programme, Sickle Cell Disease in Childhood: Standards and Guidelines for Clinical Care. 2010. http://sct.screening.nhs.uk/cms.php?folder=2493#fileid10756

Appendix One: Draft Networks of clinical care

Region	Newborn Screening Lab	SHT Network Centres with associated local hospitals
East of England	Cambridge Great Ormond Street	East of England Trusts now linked into North Middlesex London network E&N Herts (Lister & QEII); West Essex (PAH, Harlow) (choice given); Norfolk, Suffolk, Peterborough & Cambridge units (satellite clinic held at Addenbrookes and discussions to set one up in Norwich)
East Midlands	Sheffield	**Nottingham** **Leicester** Associated local hospitals: Derby, Kettering and Northampton (adults)
London		
East London & Essex	Great Ormond Street	**The Royal London** Associated local hospitals: Barking Havering & Redbridge, Whipps Cross Hospital, Basildon, Newham, Homerton, SE Essex (Southend); NE Essex (Colchester); Mid-Essex (Chelmsford); West Essex (choice given)
North East London	Great Ormond Street	**North Middlesex Hospital/Great Ormond Street** Associated local hospitals: Chase Farm at Enfield, Princess Alexandra at Harlow, Addenbrookes at Cambridge, King's Lynn, Norfolk and Norwich, Ipswich
North Central London	Central Middlesex	**University College Hospital/ Whittington Hospital** Links to many trusts particularly for tertiary thalassaemia review
North West London	Great Ormond Street Central Middlesex	**Central Middlesex** **Imperial (St Mary's Hospital & Hammersmith)** Associated local hospitals: Ealing Hospital, West Middlesex at Isleworth, Hillingdon Hospital, Northwick Park, plus outside the Brent area looser links with Luton, Milton Keynes, Watford and Bedford
South London	King's/GSTT	**King's College Hospital, London** **Guy's and St Thomas's Hospital (GSTT)** Associated local hospitals: Mayday Hospital at Croydon (paeds), Queen Mary's Sidcup, Queen Elizabeth Hospital at Woolwich, Brighton and other SE Coast hospitals including Medway,

		Dartford and Farnborough Some patients from: SW Essex and SE Essex
South West London	St Helier	**St George's Hospita**l (?) Associated local hospitals: St Helier, Mayday (mainly adults), Royal Surrey at Guilford, East Surry at Redhill, St Peter's Hospital at Chertsey
Northeast and Yorkshire & Humberside	Newcastle Leeds Sheffield	**St. James' Hospital, Leeds** **Sheffield** Associated local hospitals: Bradford, South Tees (James Cook Hospital) plus Newcastle & NE
North West	Manchester Liverpool	**Manchester Children's Hospital** **Alder Hey Children's Hospital** Associated local hospitals : Blackburn (Queen's Park), Lancaster, Tameside
South Central	Oxford Portsmouth	**Configuration yet to be confirmed** Hospitals in Region: Milton Keynes, Royal Berkshire Hospital at Reading, The John Radcliffe Hospital at Oxford, Southampton (including Basingstoke and Portsmouth)
South East Coast	Various	All trusts now linked to South London via King's or GSTT
South West	Bristol	**Bristol Royal Infirmary** Local: Derriford Hospital, Plymouth (low prevalence area)
West Midlands	Birmingham	**Birmingham Children's** Associated local hospitals: Sandwell, Wolverhampton, Coventry, University Hospital of N. Staffs at Stoke, (Northampton and Kettering paeds)

Appendix Two: Draft Letter for parents of babies who are reported as Sickle cell carriers

Dear Parent / carer

Re: Newborn Screening

When your baby was about a week old, your midwife took some blood from your baby's heel. The blood was used to test for some rare conditions listed below: phenylketonuria, congenital hypothyroidism, sickle cell disease, cystic fibrosis, and MCADD.

The results show the baby is not thought to have any of these conditions but please remember that screening tests are not 100% accurate.

The test results show that your baby is healthy, and does not have sickle cell disease. However, your baby carries a gene for

_____

Your baby is healthy; there is no need to worry. Enclosed is a leaflet that explains your baby's result which you will be able to keep for future reference.

If you have any questions about your baby's screening result, you should discuss this with your GP or health visitor (insert Name) who can be contacted at (insert phone number)

They will be able to explain your baby's result in more detail.
Yours sincerely

Yours sincerely,

<INSERT signature >

<INSERT name >

For further information visit www.newbornbloodspot.screening.nhs.uk
For the NHS Sickle Cell & Thalassaemia Programme visit http://sct.screening.nhs.uk/
For general health advice and information you can call NHS Direct on 0845 4647 or visit www.nhsdirect.nhs.uk

If you would like this letter in another format please ring <INSERT CHRD contact telephone number>