NHS Sickle Cell and Thalassaemia Programme News

Gateway Number 2013522

sct.screening.nhs.uk

March 2014

Failsafe Mechanism for Transfused Babies

In a bid to ensure that transfused babies are offered appropriate screening, the Sickle cell and Thalassaemia screening programme team introduced a failsafe measure which offers DNA testing for babies who have not had a pre-transfusion blood spot sample taken.

Babies who are not screened prior to having a blood transfusion are at considerable risk of being missed for sickle cell screening.

As a failsafe measure to ensure that transfused babies are offered appropriate screening, the programme commissioned a service of DNA testing for babies who have not had a pre-transfusion blood spot sample taken.

The process identifies babies with the sickle cell gene, regardless of transfusion state or gestational age, which require follow up and possible treatment. It is not aimed at replacing the pre-transfusion sample. NHS England is to pick up the costs for this service as of October 2014 (**Service specification No.18 NHS Sickle Cell and Thalassaemia Screening Programme**).

The Most Reverend and Right Honourable Dr John Sentamu with members of the SCT Steering Group

A Fond Farewell to our Chairman, The Archbishop of York

Archbishop of York, John Sentamu, played host to the Sickle Cell and Thalassaemia Steering Group for one final time at Bishopthorpe Palace on 19 February 2014.

The Archbishop was thanked officially by deputy Chief Medical Officer, Professor David Walker who, during his speech, made mention of the Archbishop's extraordinary contribution to the programme during his 12 years in post and highlighted some of the key successes he had helped to bring about.

The Archbishop thanked the group for their support over the years and gave a personal account of his experiences with the programme. He pledged to follow the programme's progress with interest.

Changes to Family Origin Questionnaire

The Family Origin Questionnaire (FOQ), a tool used by health professionals to assist in informing the level of risk that an individual has of being a carrier of sickle cell or thalassaemia, has undergone some small changes following requests by stakeholders. Changes include the addition of Sri Lanka under family origins and more tick boxes to explain unknown ancestry, eg bone marrow transplant and donor egg or sperm.

The new form will be version 3.0 and dated February 2014. We will announce it in the news section of our website when it is ready, and the revised form will be accessible in PDF format on the programme website as well as available to order in hard copy. The changes are small and it is expected that old stocks of FOQ forms will be used up before moving onto the revised version.

sct.screening.nhs.uk/foq

F-Only Project

Beta thalassaemia major may be detected as a by-product of newborn screening for sickle cell disease, as these babies have little or no HbA production. The UK NSC recommends that clinically significant findings of conditions which are not part of the screening programme, but are detected by current methods, should be referred for clinical follow up.

Laboratory guidance recommends that babies with an action value of 1.5% or less Hb A should be referred. Not all cases will be detected using this action value.

The F-Only project aims to provide evidence relating to the 1.5% action value by correlating DNA findings with the Hb A% on newborn blood spots.

Data on confirmatory DNA results on babies with Hb A results of 1.5% or less are being collected. A second call for data has recently been sent to laboratories.

Newborn Screening Using Mass Spectrometry

Pilots for this project are now underway at laboratories in Leeds, Oxford, Guys and St Thomas's and Birmingham and have highlighted a number of issues which need to be considered before the use of TMS is implemented more broadly.

Careful consideration needs to be paid to the action values implemented as these differ between instruments and sites. The pilot project has also demonstrated that lack of sensitivity on some instruments compromises results and leads to high repeat rates. There will therefore, be limitations on the instruments recommended for use as this directly impacts action values.

At this time not all newborn laboratories have the appropriate instrument capacity/sensitivity.

Implementation will need to be carried out in close co-operation with the laboratory advisors to ensure consistency and ensure that appropriate action values are set. The programme's laboratory advisors will write up the project this summer.

MBE for Dr Lorna Bennett

The programme is delighted to announce that Dr Lorna Bennett has been awarded an MBE for services to people with blood disorders and her contribution to haemoglobinopathy services.

Dr Bennett received her award as part of The Queen's Birthday honours, a system that recognises people who have committed themselves to serving and helping Britain. Joining her at the celebration event in June of last year were some high profile celebrities including Tony Robinson, Clare Balding and Aled Jones.

Lorna was responsible for the Camden and Islington Sickle Cell and Thalassaemia centre for many years at the same time as leading high quality work streams for the screening programme, including running the Professional Eduction for Genetic Assessment and Screening (PEGASUS) course for counsellors. She was also a member of the programme's training and education sub group. We wish her well in her well deserved retirement.

Dr Lorna Bennett after receiving her MBE

New carrier leaflets coming soon

Two new leaflets for parents who are expecting a baby and blood tests have shown that both parents-to-be carry a gene for unusual haemoglobin, are close to completion. The leaflets give information about: Sickle cell and thalassaemia Explain the likelihood of inheritance The tests that can be done during pregnancy to find out if an unborn baby has sickle cell disease or thalassaemia major Options and choices for parents to be We will let you know when the leaflets are available via the programme's website.

QA in the new structure

The new regional Quality Assurance teams were established on 1 April 2013 and are now taking shape. Their role is to provide a comprehensive and consistent approach to the quality assurance of antenatal, newborn and adult screening programmes.

There are more details about the teams at **www.screening.nhs.uk/regionalteams** including how to contact them. In addition to the new QA teams, KPIs were produced for the screening programmes in December of last year. These give a high level overview of the quality of screening programmes at key points on the screening pathway. The key messages for the SCT indicators are:

- reported coverage for Sickle Cell and Thalassemia (ST1) is high at 98.2% for those trusts that state they can provide cohort data. 27 Trusts across England cannot provide cohort data as required for ST1
- ST2 measures whether the result is available before 10 weeks gestation. The acceptable level is 50%. The England average was 48.7% with London (33.9%), West Midlands (39.5%) and North West (47.8%) reporting below 50%
- ST3 assess the proportion of Family Origin Questionnaires (FOQ) that are correctly completed and returned to the laboratory with the sample. The FOQ in low prevalence areas is the SCT screening test; failure to complete the FOQ will result in women not being screened. In high prevalence areas failure to use the FOQ increases the false positive rate. Completion rates are not as high in high prevalence areas compared with low prevalence areas

Training

Short Counselling Course

The SCT programme is offering a study day for non-specialists (nurses, midwives, health visitors and others) at King's College London. Students will develop an understanding of the universal newborn screening programme in general and improve their evidence based knowledge and skills to provide information and counsel couples who are at risk of having a baby with a haemoglobinopathy in providing results to parents of babies identified as carriers.

The next course will be run on 24 April 2014. More information is available on the Kings College, London website.

Laboratory training days

The programme, in collaboration with London Metropolitan University, will be running CPD-accredited training days for antenatal and newborn screening laboratory staff on 9 April 2014 in London and on 1 May 2014 in Leeds. Bursary places are available and delegates will pay only £20 to attend. Non-NHS staff can attend for £120. The day is suitable for both antenatal and newborn screening laboratory staff and haematologists with an interest in screening.

UK NSC considers pulse oximetry

Following a review by the UK National Screening Committee (UK NSC) last year, the use of the non-invasive pulse oximetry technique in the detection of congenital heart disease in newborn babies went out to public consultation.

There were a large number of responses, which were considered by the UK NSC on 12 March and a recommendation will be made for ministerial approval. Further information about the review can be found on the **UK Screening Portal**.

Consultations are also open with the UK NSC for:

- increasing the number of diseases tested for in **newborn blood spot screening** (closes on 20/03/14)
- **type 2 diabetes** (closes on 25/03/14)
- **atrial fibrillation** (closes on 24/03/14)
- first trimester screening for **Trisomy 13 and 18** (closes on 16/06/14)

Screening incidents

One of the most important reasons for reporting and investigating screening incidents is to enable learning to occur and for this learning to be widely shared. As part of the UK NSC's commitment to safety and quality there will be at least one lesson learnt included in every edition of Screening Matters.

Screening Matters is the newsletter of the UK National Screening Committee, published three times a year. It covers evidence and policy development, training and education, quality assurance and IT and information, as well as providing updates on all the non-cancer NHS Screening Programmes in England. The newsletter is aimed at everyone involved in screening - policy makers, commissioners, providers and wider public health professionals. If you do not already receive this newsletter do **contact us** to subscribe.

The most recent review of screening incidents highlighted two areas are of particular concern for the SCT programme: offering screening to all eligible babies and timely referral of screen positive babies to care.

Screening Matters issue 17 can be downloaded from

www.screening.nhs.uk/screeningmatters

New service replaces NHS Numbers for Babies

From 1 July 2014 the Personal Demographics Service (PDS) will replace the NHS Numbers for Babies (NN4B) service.

Beyond this date, birth notifications will be sent or received by the PDS only. Maternity systems that have not been upgraded to use PDS will no longer be able to register births and receive NHS numbers for babies.

So far only a couple of maternity and child health systems have become PDS-compliant. Any maternity units with non-compliant systems will need to use the standalone web-based Birth Notification Application (BNA) to register births and receive NHS Numbers. This will lead to additional effort and the risks of manual data entry, though the birth notifications will still be sent automatically to the NHSP IT system (eSP).

Trusts are encouraged to check whether their maternity system will be compliant by 1 July and liaise with their supplier if not. For more details, see the latest **NN4B bulletin**

Other news in brief

- UK NSC Director of Programmes Dr Anne Mackie has written an article on Public Health England's **Public Health Matters blog** that introduces many of the current debates around screening.
- The Screening Tests for You and Your Bary information booklet given to all pregnant women is being reviewed. The booklet covers all the antenatal and newborn screening tests offered for mothers and babies, along with the additional retinal screening tests provided during pregnancy for women with diabetes.
- **The Antenatal and Newborn e-learning module** has been updated to reflect the latest developments in the NHS Screening Programmes and to incorporate lessons learnt from serious incidents.
- It remains the case that the UK NSC does not recommend screening of women in late pregnancy for the carriage of Group B Streptococcus (GBS). Work done within Public Health England determined that use of enriched culture medium (ECM) test for GBS would be outside current clinical guidance and therefore the test has not been introduced.
- Professionals are needed to join the Quality Assurance teams of peer reviewers. Please see the **advert on the UK Screening Portal**.