Expanded Newborn Screening Newsletter

Issue 8
July 2013

WELCOME to the 8th edition of the Expanded Newborn Screening newsletter. On the 19th July 2013, The Expanded Newborn Screening Programme turned one. We would like to say a huge well done and thank you to everyone who has contributed to make the pilot a success. As the 12 months results will be available very shortly, we will not be publishing any results in this edition but these will all be included in the bumper edition in August. In this newsletter we will remind you of the process now the study section has ended and ask for your help in evaluation. Our Meet the Team this month features Manchester. As always, we hope you enjoy the newsletter!

The study has finished. What happens now?

The 19th July marked the end date for babies screened to be included in the data collection for the cost-effectiveness study. However, screening is continuing and there are a number of activities going on.

Screening will continue until 31st March 2014. As such, we request that midwives and health visitors continue to distribute the leaflets on the Expanded Screening to new mothers. Don't forget that it is the new version of the leaflet which should be now handed out. This leaflet states that the screening continues until 31st March 2014. Should you have any queries about this please contact your Regional Co-ordinator or Jason Sowter (Jason.Sowter@sch.nhs.uk)

Data collection will be continuing in the centres for parents of babies who were screened up to and including 19th July and who had a screen positive result. This data collection will inform the cost-effectiveness model. Other work around the health economics model is currently going on including interviews with clinicians to gather information on the treatment and outcomes related to clinically identified (i.e. not through screening) cases of each condition. The first version of the model will be ready by late October to provide information on the cost-effectiveness of the expanded screening which will be included in the report to the National Screening Committee.

Now the pilot study has been completed, other data is also being gathered together to inform the National Screening Committee report. We would like your help with this by completing a short questionnaire (see right).

We will continue to collect data from labs on any new true or false positives of the conditions on a month by month basis. The newsletter will continue to keep you informed with any new cases which occur during the continued screening period and our findings as we prepare for the report to the National Screening Committee.

Last, but certainly not least, Louise Moody will shortly begin the interviews for the qualitative study which will explore communication of a screen positive result.

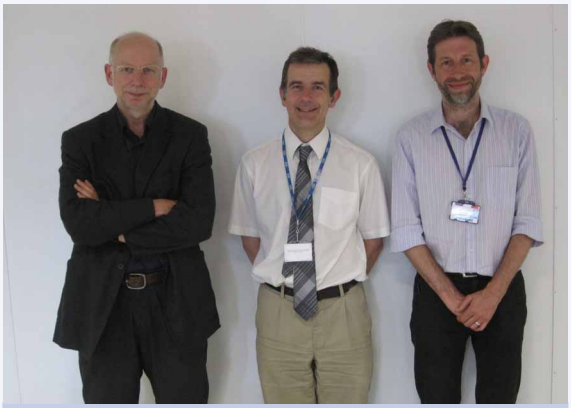
We want your input

We would like to gather views from doctors, nurses, dietitians, clinical biochemists, clinical scientists, regional co-ordinators, midwives and health visitors who have been involved in the pilot. You should recently have received a request to complete a questionnaire via email. If for any reason you have not received this and would like to help by completing a short questionnaire, please contact Jason Sowter. Different versions of the questionnaire are available for the different professionals who have been involved, therefore please can you also state your role in your email request.

We want your feedback and comments! We want this newsletter to be useful and interesting to you. Please provide feedback and any information that you would like including in the newsletter via the website: http://tinyurl.com/cjwg8nh .

NHS
National Institute for Health Research

Meet the Team in Manchester



The metabolic consultants:
Left to right: (John Walter, Andrew Morris, Simon Jones

Staff from the laboratory, dietetic and nursing services

The clinical team in the Willink Biochemical Genetics Unit at Central Manchester University Hospitals consists of 4 specialist metabolic consultants (two of whom also cover the Bradford and Leeds clinics), and a number of specialist metabolic dietitians and nurse specialists, who collectively look after patients with a wide variety of inherited metabolic diseases, some of whom have been diagnosed through newborn screening and some when presenting clinically. The laboratory team of clinical scientists and technologists provides both the newborn screening service for IMDs (PKU, MCADD and the expanded panel) and a diagnostic service for an extensive range of disorders. The clinical and laboratory teams are co-located, making referral to the clinical team and diagnostic follow up of positive screening cases very easy to organise! Here is what they have to say:

"*When I began my career in biochemical genetics I quickly became used to making the laboratory diagnosis of MCADD in seriously ill, sometimes even deceased, children several times a year. It was very frustrating, especially knowing that the treatment for MCADD is so simple once it is known that a child is affected. The introduction of newborn screening for MCADD in the UK has changed all that. My hope for expanded newborn screening is that we do the same for many more disorders, and not just those on the current expanded panel. The value of newborn screening was brought home to us recently when we made the diagnosis of Glutaric aciduria type 1 in a child with significant clinical problems who was born just a little too soon to benefit from the expanded screening pilot*." **Claire Hart, Consultant Clinical Scientist / Head of laboratory service**.

"*Standardisation of a biochemical test for simultaneous screening of the five extra metabolic diseases in the newborns has been a common vision of all 6 pilot sites from the outset. Testing strategies were developed through careful planning and close collaboration among the 6 laboratories before the pilot study began. They were then transformed and maintained to high standards to ensure a robust test at the operational level through sheer hard work of each local newborn screening teams, our lab team in the Willink Unit is no exception.*"
Teresa Wu, Principal Clinical Scientist, Laboratory

"*As a clinician I fully support the expansion of newborn screening and am very happy with the success of the ENBS pilot so far, a number of children have been identified and treated and, importantly, the false positive rate has been within expectations*". **Simon Jones, Consultant Metabolic Paediatrician**

"*As metabolic dietitians we are well aware of the benefits to many patients of early diagnosis in implementation of dietary treatment and long term outcomes. We are therefore encouraged to see the positive effect of the ENBS pilot project in picking up cases early so preventing or minimising clinical problems for these children. It was a positive experience to be involved with my dietetic colleagues from the other pilot sites to develop the standardised dietetic treatment guidelines for these 5 disorders and hope this collaborative working continues*"
Fiona White, Chief Metabolic Dietitian

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