



**WELCOME** to the 12th Expanded Newborn Screening newsletter. The National Screening Committee (NSC) held a public consultation from Dec 2013 – March 2014 in relation to the five conditions that formed part of the pilot to help determine whether they should become part of the national programme. Thank you to everyone who contributed; I understand that the responses will be summarised and published. The outcome is that four of the conditions that have been evaluated (glutaric aciduria type 1, homocystinuria (pyridoxine unresponsive), isovaleric acidemia and maple syrup urine disease) will be screened nationally, with all sites in England offering the screening tests by early 2015. Wales, Northern Ireland and Scotland have expressed their interest but need to make independent policy decisions based on the NSC's advice.

This is an excellent outcome, so well done to everyone involved in the project, and thank you for your hard work and commitment, it really was a team effort.

## What will happen now?

Areas involved in the Expanded Newborn Screening pilot project will continue to screen babies for all five conditions until 31 August 2014, and will then need to screen for four beyond that date.

Areas that have not previously screened for these conditions will begin to do so in the coming months, such that screening is underway across all sites early in 2015. This national roll-out will be co-ordinated by the UKNSPC. The policy details on pre-screening advice and reporting of results, together with the laboratory aspects, are currently under consideration. Guidance will emerge during the summer.

If you have any queries in the next few weeks please contact [jason.sowter@sch.nhs.uk](mailto:jason.sowter@sch.nhs.uk) or your regional co-ordinator.

## *Expanded Newborn Screening*

### **Advice for parents and health professionals**



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## Which activities are currently underway?

### Roll out

While the pilot study worked effectively it is important that the UKNSPC details policy in relation to the national roll-out. In particular the future of the website, the nature of pre-screening information and midwifery training, along with a route to report testing to Child Health Record Departments, needs to be agreed. NEQAS have kindly undertaken to provide an EQA scheme to support the new disorders.

### Publications under preparation

We are now preparing academic publications relating to the ENBS project. Anticipated publications include the health economics information, the review conducted by the PHG Foundation, a paper around the whole project. Several papers regarding detailed aspects of the project are also under discussion.

### Communication study

Louise Moody and her team at Coventry University are running the Communication Study, which will help to understand the most effective way in which information can be given and parents can be supported. Louise and her team have already interviewed seven clinicians and one parent, but still need your help in arranging further clinician and patient interviews. R&D approvals are still being sought at some sites.

### Films

Optical Jukebox have completed filming of six new films. An updated MSUD film showing Habul and her mum's progress since her diagnosis (Habul at One Year) is now available on our website, and we hope to see finished versions of the other films in the next couple of weeks. Many thanks to all our volunteers!

## Screening results update

We now have screening data for 21 months since we began the ENBS project. In that time 753,632 babies have been screened, with only 250 parents declining screening (0.03% of parents). We have identified 27 true positives, and 33 false positives:

	MSUD	HCU	IVA	GA1	LCHADD	Total
True positives	2	3	9	8	5	27
False positives	2	1	26	1	3	33

As screening is now extended until 31 August 2014 we anticipate approximately 6 further true positive results. By that date we will have screened approximately 930,000 children.

### Contact details

Dr Kathy Jeays-Ward has taken over the role of Project Manager, and can be contacted by email ([k.jeays-ward@sheffield.ac.uk](mailto:k.jeays-ward@sheffield.ac.uk)) or phone (0750 7638983). Jason Sowter is available by email ([jason.sowter@sch.nhs.uk](mailto:jason.sowter@sch.nhs.uk)) or phone (0114 226 7980)

This project is supported by the National Institute for Health Research Collaboration for Leadership in Applied Health Research and Care for South Yorkshire (NIHR CLAHRC SY). The views and opinions expressed are those of the authors, and not necessarily those of the NHS, the NIHR or the Department of Health. CLAHRC SY would also like to acknowledge the participation and resources of our partner organisations. Further details can be found at [www.clahrc-sy.nihr.ac.uk](http://www.clahrc-sy.nihr.ac.uk).  
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