



**WELCOME** to the 3rd edition of the Expanded Newborn Screening newsletter. It has been a quiet month for the pilot, but we have lots of good news to share with you. We would like to thank you for all your hard work in 2012, and wish you a Merry Christmas and a Happy New Year!

## The Numbers

Only 2 cases have been identified this month both of which were screen positive for IVA. One case has been identified as true positive for IVA. The other case has involved quite a lot of detective work to finally identify it as a false positive. The diagnosis is now thought to be another metabolic disorder called SCBADD. Babies are quite often identified with this condition through screening. Often the condition does not have any clinical impact though delayed neurological development has been reported in some cases. The number of declines seen each month continues to decline. There were just 17 families who declined to participate in the expanded screening this month, this has fallen substantially from the 64 declines which we saw in the first month.

## Extension of the Pilot Study

We are pleased to announce that the UK National Screening Committee has provided funds to extend the pilot until 31st March 2014. This will allow the UK NSC to receive the full evaluation of the pilot and take a view on whether a full national roll out is appropriate beyond this point. The letter from Dr Anne MacKie, Director of Programmes at the UK National Screening Committee confirming these arrangements can be downloaded from the expanded screening website.

## Funding for additional research

We are pleased to report that we have secured funding for a small qualitative study which will aim, through interviews, to understand more about how positive screening results are communicated and the impact it can have on a family. The information collected in the study will be used to develop guidelines for this. Dr Louise Moody will be leading this research which we hope will commence early Spring. Louise's contact details are: [L.Moody@coventry.ac.uk](mailto:L.Moody@coventry.ac.uk)



## Congratulations Professor Bonham!

We are pleased to announce that Jim's dedication to the area of newborn screening has been recognised and he will be made an honorary professor in the Department of Human Metabolism, University of Sheffield. Jim's title will be Honorary Professor of Inherited Metabolic Disease.

Scan this QR code with your smart phone to connect directly to the ENBS website.



## Website News!

Redevelopment of the website is underway. We hope it will be up and running in the early New Year! As part of the website redevelopment, we are developing condition-specific booklets which contain all the information (from screening protocols, to guidelines, to parent booklets) for the pilot on that condition. These will be available to download from the website soon.

## Film News!

During the Autumn the expanded screening team worked with Optical Jukebox to produce a film about Will and his experience of living with LCHADD. We are pleased with the positive response that the film has received and feel that it helps viewers understand more about the rare condition. As part of the website development, it has been agreed that more films will be commissioned and will focus on each of the conditions in expanded screening pilot. Filming for these is due to commence in January.