

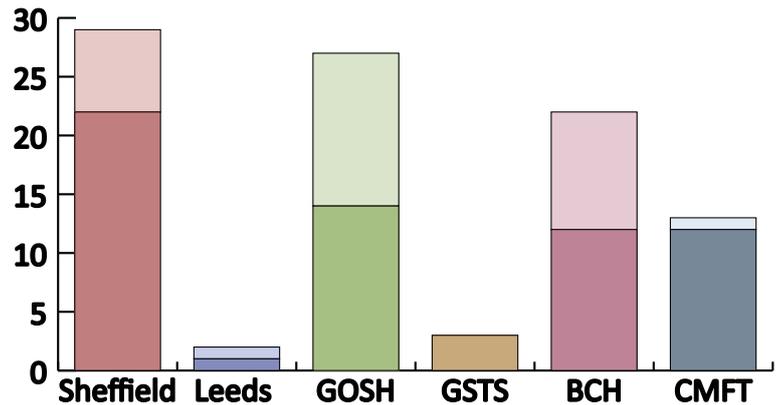


**W**ELCOME to the first Expanded Newborn Screening Newsletter! It has been an exciting couple of months, as finally after months of preparation the Expanded Newborn Screening Project went live on 16<sup>th</sup> July 2012. This was a result of dedicated hard work from all around the country for which we are truly grateful. We would like to keep you up-to-date with progress in the pilot and will be issuing a monthly newsletter to do this. We hope you enjoy!

## The Numbers:

From the 16th July to 17th September, it is estimated that around 70,000 babies will have been screened. During the 1st month, screening identified 2 true positive cases and 1 false positive case, all for IVA. During the 2nd month, screening identified 1 true positive case for MSUD (see below) and a further screen positive case for IVA.

During the first two months a total of 96 (estimated to 0.13% of the total offered screening) parents declined the expanded screening. The 2nd month saw a total of 32 declines, half of that received during the first month. Some geographical areas are reporting higher numbers of declines (see right) which the team are working hard to address.



The graph above depicts the number of declines in the first and second months of expanded newborn screening pilot. The darker colour within each stack depicts the 1st month. The number of declines in the 2nd month has decreased across all sites.

(Abbreviations: GOSH: Great Ormond Street Hospital, GSTS: Guys's St Thomas', BCH = Birmingham Children's Hospital, CMFT = Central Manchester Foundation Trust)

## A Case of Maple Syrup Urine Disease (MSUD) Identified Just in Time

The first positive case of MSUD has been identified through the Expanded Newborn Screening Programme. The infant, who was 9 days old when the screen positive result was identified, was already beginning to experience difficulties as a result of the rare condition. A Specialist Metabolic Nurse made an urgent home visit to discover a lethargic baby who was not responding well to stimuli. Mum had realised the baby was a little under the weather but had not known how serious the situation was. The infant was blue-lighted to hospital where treatment for MSUD was immediately commenced. Confirmatory testing was completed using blood

and urine analysis. The baby responded well to treatment given in Intensive Care and is now doing well at home with Mum. The Specialist Metabolic Team are overseeing regular follow-up visits and they hope that treatment will allow the baby to grow and develop normally. Mum is reported to be very grateful for the screening and feels very lucky that she is living in an area where her baby was screened for this rare condition as would not be the case elsewhere. This case highlights the positive impact that newborn screening can have on individual lives.

# Taking Screening Forwards

## IVA Screening & Diagnosis

Both true positive cases of IVA have been identified as mild variants. It is felt that there is a need to monitor the process used to diagnose IVA to ensure that the cut-offs set in screening are appropriate and to gain knowledge on how appropriately identify mild cases. There is discussion around using another organic acid, isovaleryl glycine (IVG), as a marker of severity by detecting it in urine. Some work has already been completed on correlating IVG excretion with severity in a limited number of genotypes. We are considering expanding this work. Repeated measurement of IVG in individual

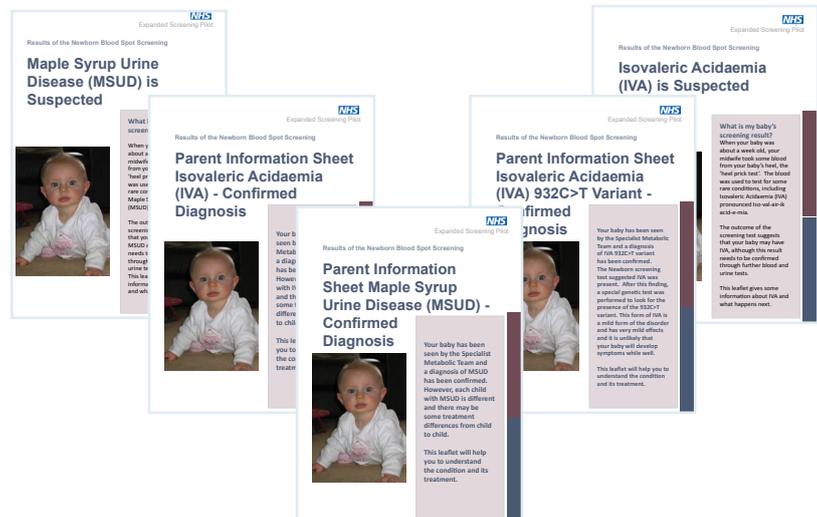
patients over a period of time to build up a picture of IVG levels in times of illness may be a means of stratifying risk. We feel that a meeting with experts from The Netherlands and Germany to discuss their experience with mild mutation (known and unknown) cases would be beneficial.

## Developing International Links

Dr Jim Bonham is spreading the word about the benefits of newborn screening in places far afield such as India and South Africa. These countries do not currently have a newborn screening programme in place and are keen to use the expertise within the NHS to begin to develop their programmes.

## LCHADD Film

The Expanded Newborn Screening Project has commissioned Optical Jukebox to produce a film. The film provides information about expanded screening programme and focuses on the case of William, now 22 years old who was identified with LCHADD whilst seriously ill as a baby. The film has been well received at high profile events and will shortly be available on the website. There are plans to make more short films in the future.



## The Expanded Screening Website

Information about the pilot and five conditions can be found on the expanded screening website. The website contains information for both members of the public and professionals. Information on screening protocols, diagnostic protocols, recommended clinical and dietary management can be downloaded after by obtaining a password from the core team to enter the restricted area. Parent

leaflets (including translation of the midwifery and parent information into 15 languages) can easily be downloaded without need for the password.

The usage of the newborn website is being monitored. Discussions internally and with CLIMB suggest that there may be more effective ways of delivering information to members of the public. A review is underway and hopefully the website will soon have a new look!

We want your feedback and comments! We want this newsletter to be useful and interesting to you. Please provide feedback on any information that you would like to be included in the next newsletter, any ideas you may have for the website etc. etc. Please contact via the website at <http://www.expandedscreening.org/home/contact.asp>

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