

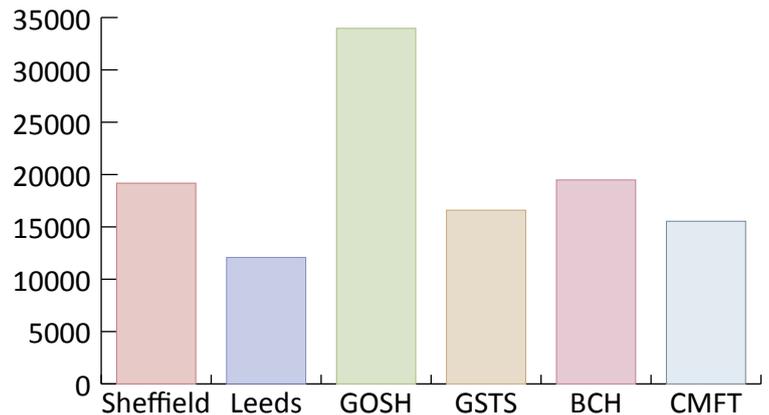


**W**ELCOME to the 2nd edition of the Expanded Newborn Screening newsletter. October 17th marked three months into the pilot study! A big well done to everyone who has made the pilot such a success so far. This newsletter will focus on the review of the results so far and compares with our projections for this point. We hope you enjoy it!

## The Number of Births:

During the 1st quarter 116,847 births have been reported. Over the year, we were anticipating a total of around 430,000 births. Based on the numbers for the first quarter, the total number of births over the year may be up by around 37,000.

The graph to the right depicts the number of births by centre during the 1st quarter. (Abbreviations: GOSH: Great Ormond Street Hospital, GSTS: Guy's St Thomas', BCH = Birmingham Children's Hospital, CMFT = Central Manchester Foundation Trust)



## The Number of Declines:

The total number of declines over the 3 month period is recorded as 122. This is just 0.1% of the total number of births which in general shows good acceptance of the expanded programme by mothers and great achievement by the midwives in encouraging mothers to participate. The number of declines reported each month is slowly falling. Only 26 were recorded in September-October, a small decrease from the 32 in August-September, and less than half those in the first month when a total of 64 declines were recorded. The number of declines still remains a little higher in Sheffield compared to other sites. Leeds & GSTS have lowest decline rates.

Site	Month 1	Month 2	Month 3	Total Declines at sites	% based on number of births at site
BCH	12	10	1	23	0.12
GOSH	14	13	9	36	0.11
GSTS	3	0	1	4	0.02
Leeds	1	1	1	3	0.02
CMFT	12	1	5	18	0.12
Sheffield	22	7	9	38	0.20
<b>Total Declines</b>	<b>64</b>	<b>32</b>	<b>26</b>	<b>122</b>	

## LCHADD Film

The LCHADD film featuring Will and his story of living with the condition, is now up online. Click [here](#) to view it. The team have entered the film into a NIHR new media competition. Fingers crossed!!

## GA1 Case Identified

The Expanded Newborn Screening Programme has successfully identified the first positive case of GA1. Screening identified the baby before they had suffered any ill-effect of the condition. The Specialist Metabolic Team are overseeing regular follow-up visits.

## The Number of screen positive, true positive and false positive cases:

Identifying babies with one of the five rare conditions is a two stage process. During the newborn screening, the tests identify babies with a raised metabolite which may be indicative of having one of the conditions. These cases are called *screen positive*. Further diagnostic testing is then completed; those who are determined to have the condition are called *true positive* cases and those who do not have the condition are called *false positive*. Other illnesses and certain medications can cause a screen positive result and therefore we predicted that there would be more screen positive cases over the course of the year than true positive cases.

It is difficult to make inferences from the numbers as each of the conditions are so rare and through chance it is unlikely that the cases will be evenly distributed throughout the year. However, in general, the numbers seem acceptable compared with our predictions. The number of screen positive cases of IVA are a little higher than anticipated, and the true positive cases identified through screening so far are mild. It seems sensible to reflect on the IVA situation in collaboration with colleagues overseas where screening has been undertaken for several years and we are planning a workshop.

Over the year pilot it is predicted that the following number of cases will be seen:

Condition	Screen positives	True positives	False positives
GA1	10	4	6
HCU	8	3	5
IVA	10	3	7
LCHADD	5	2	3
MSUD	8	4	4
TOTAL	41	16	25

The total numbers reported so far during the pilot are as follows:

Condition	Screen positives	True positives	False positives
GA1	1	1	0
HCU	0	0	0
IVA	8	3	5
LCHADD	0	0	0
MSUD	2	1	1
TOTAL	11	5	6

## Leaflet Translations:

Translations of the Advice for Parents & Health Professionals leaflet are available on the website in written format in 16 different languages (see right). Based on feedback from CLIMB, we have commissioned audio recordings of the six of these. Audio translations into Bengali, Gujarati, Somali, Hindi, Lithuanian, and Urdu should be available on the website from Mid-November.

### Written Translations:

- French (as spoken in Africa)
- Spanish (South American)
- Portuguese (African)
- Turkish
- Romanian
- Polish
- Russian
- Chinese Simplified
- Chinese Traditional
- Bengali
- Kurdish
- Hindi
- Gujarati
- Arabic
- Urdu
- Somali

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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