



WELCOME to the fifth Expanded Newborn Screening Newsletter. We hope you have had an enjoyable Easter. Please accept our apologies for being unable to send this newsletter out before the Bank Holiday weekend. We had feedback from the fourth edition of the newsletter that the readers wanted full information on the data and therefore we have revised the format of the newsletter to include this. Information which we had hoped to provide you in March on data collection for the research study is also included in this newsletter. We hope you enjoy reading it!

The number of births:

The study has now been active for 8 months. The number of births is collected from sites on a quarterly basis and therefore we will be able to report the numbers of births for the three quarters on the next newsletter. The number of births was a little higher than expected in the first quarter and a little lower than expected in the second quarter. The results of the third quarter will be interesting to see whether our figures are going up or down!

The number of declines:

The number of declines is still slowly falling with each passing month showing that the expanded screening programme is well accepted by both professionals and mothers. Well done! The number of declines is still a little bit higher in Sheffield but given the total number of declines across all 6 sites in the February to March period was only 8, we feel that this is pretty good going!

Updated leaflets now available

With the extension of the testing period for expanded screening now running until 31st March 2014, new midwife leaflets have been produced and are now available. These leaflets should now be used and any old stock discarded. New leaflets can be obtained from Harlow Printers. Details of how to order can be found at: <http://tinyurl.com/cpucyx7>

We are in the process of updating the written and verbal translations, we will let you know when these are ready for download from the website.

Site	Month 1	Month 2	Month 3	Month 4	Month 5	Month 6	Month 7	Month 8	Declines by site
BCH	12	10	1	2	3	4	0	1	33
GOSH	14	13	9	4	6	2	5	1	54
GSTS	3	0	1	2	0	1	0	1	8
Leeds	1	1	1	2	0	0	0	0	5
CMFT	12	1	5	4	5	4	3	2	36
Sheffield	22	7	9	5	4	3	4	3	57
Declines by month	64	32	26	19	18	14	12	8	193

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 www.expandedscreening.org/site/home/contact.asp



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

The number of screen positive, true positive and false positive cases:

As we have reported previously, it is difficult to make inferences from the numbers as each of the conditions are so rare. By chance it is unlikely that the cases will be evenly distributed throughout the year and therefore it is possible that everything will change during the remaining four months of the study. However, in general, the numbers seem acceptable compared with our predictions. The number of true positive cases is about on target. For all conditions except IVA, the numbers of false positive cases are lower than expected.

As reported previously, the number of screen positive cases of IVA are a little higher than anticipated; another false positive for IVA was identified in this past month. So far, all of the true positive IVA cases identified through screening have been mild. We have previously reported that we plan to run an international workshop to reflect on our cases with overseas colleagues where screening for IVA has been undertaken for several years. Plans for the workshop are progressing well; Jim Bonham and James Leonard have jointly produced a questionnaire to find out more from international colleagues about their experiences of screening for IVA. This background information will inform the discussion of the workshops.

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	3	3	0
HCU	1	1	0
IVA	11	3	8
LCHADD	0	0	0
MSUD	2	1	1
Total	17	8	9

The research study

As part of the research study a health economics analysis will be completed to look at the potential benefits of screening. In order for this to be completed data will need to be collected from each of the sites where a screen positive case is identified. Parents provide consent for the data collection which covers information about the number and length of clinical appointments, members of staff seen, tests ordered etc. Ethics and research governance approvals are now in place for all sites which allows the data collection to begin. Clare Gibson will begin collecting data for this in April starting in Sheffield before arranging to visit other sites.

Other news in brief:

- The six new films were aired at our Project Meeting and a positive response was received. The group felt that there would benefit in producing additional films about HCU and IVA and we are currently investigating the possibility of this.
- Information about the Expanded Newborn Screening study was presented at the Newborn Screening Laboratory Network Annual General Meeting and was well received by the audience.
- CLAHRC for South Yorkshire have published a Health Inequalities Project Case Book. Within this document information is provided on a study looking at consanguineous marriage and the likelihood of inheriting a rare condition. Take a look here: <http://tinyurl.com/blehaom>

The Expanded Newborn Screening 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.

© Copyright Sheffield Children's Foundation Trust 2013, a member of NIHR CLAHRC for South Yorkshire. This document can be distributed freely within NIHR CLAHRC for South Yorkshire's partner organisations.