



**W**ELCOME to 7th edition of the Expanded Newborn Screening newsletter. We are now nearing the end of the study with 10 months completed. This newsletter will review activity during previous month and provide you with some information about what will happen once the study ends. We are continuing our series of Meet the Team and this month are featuring Bradford and Leeds. As always, we hope you enjoy the newsletter!!

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

In the last month we have seen two false positive cases of IVA and one true positive case of GA1. The numbers of cases to date during the Expanded Screening pilot are shown below:

Condition	Screen positives	True positives	False positives
GA1	4	4	0
HCU	1	1	0
IVA	15	4	11
LCHADD	0	0	0
MSUD	2	1	1
<b>Total</b>	<b>22</b>	<b>10</b>	<b>12</b>

## IVA Questionnaire:

We have previously reported that there have been more than expected false positive cases of IVA, and that the true positive cases of IVA have all been mild. It was agreed that this needs to be investigated further. Jim Bonham is arranging an international workshop to find out how common mild cases of IVA are around the world, and how different countries treat mild cases of IVA. In preparation for this, Jim in collaboration with James Leonard has developed a questionnaire which has been circulated to world experts and inquires about their definition of "mild" and the treatment protocol followed for mild cases. Watch this space for an update

## Remember the new leaflets:

The information leaflet for parents has been revised with the extension of the testing period. Old leaflets should be destroyed and new leaflets obtained from Harlow Printers as soon as possible. Any queries please contact Jason Sowter ([Jason.Sowter@sch.nhs.uk](mailto:Jason.Sowter@sch.nhs.uk))

## The number of declines:

The number of declines per month remains very low at around 0.02%. The low number of declines that has persisted throughout the pilot shows really good acceptance of the expanded screening programme by both professionals and parents. See the table below for a monthly breakdown of declines at each site:

Site	16/07 17/08	18/08 17/09	18/09 17/10	18/10 17/11	18/11 17/12	18/12 17/01	18/01 17/02	18/02 17/03	18/03 17/04	18/04 17/05	Declines by site
BCH	12	10	1	2	3	4	0	1	0	1	34
GOSH	14	13	9	4	6	2	5	1	2	3	59
GSTS	3	0	1	2	0	1	0	0	0	1	8
Leeds	1	1	1	2	0	0	0	0	0	0	5
CMFT	12	1	5	4	5	4	3	2	1	1	38
Sheffield	22	7	9	5	4	3	4	3	3	2	62
<b>Total Declines</b>	<b>64</b>	<b>32</b>	<b>26</b>	<b>19</b>	<b>18</b>	<b>14</b>	<b>12</b>	<b>7</b>	<b>6</b>	<b>8</b>	<b>206</b>



We want your feedback and comments! We want this newsletter to be useful and interesting to you. Please provide feedback and any information that you would like including in the newsletter via the website: <http://tinyurl.com/cjwg8nh> .

## As the pilot nears the end, what happens next?

Whilst screening will continue until 31st March 2014, only babies screened up to and including 19th July 2013 will be eligible to be involved in the research study. The study involves data collection about the care that a baby receives. Data collected (for up to 21 weeks in true positive cases) will be used for the health economics model.

Jim Chilcott and his team at Health Economics and Decision Science within the School of Health and Related Research at the University of Sheffield is currently building the health economics model. This process involves producing a model of the care received along with associated costs and outcomes for clinically identified cases compared with cases identified through screening. Information is drawn from the academic literature and key clinicians are helping Jim with this task.

The first model will be ready in November. The findings from the model along with important findings about running the pilot (such as appropriateness of screening cut-offs etc.) will be presented in a report to the National Screening Committee in November. The report will also include information on interim findings from the Communication study which was introduced in the May 2013 edition of the newsletter. The National Screening Committee will review the data supplied and make decisions on whether to support continuation of expanded screening in the long term.



### The Clinical Team

Top (L to R): Beth Jameson, Collette Stainforth, Hazel Rogozinski  
Bottom (L to R): Indy Hunjan, Jo Wildgoose, Kirsten Foster



### The Lab Team

(L to R) Mick Henderson, Tim Williams, Daniel Herrera, Janet Mitchell.

## Meet the Team at Bradford and Leeds:

The Bradford clinical team comprises of 2 consultants (based in St Mary's Hospital, Manchester), 4 specialist metabolic dietitians, a specialist dietetic assistant practitioner, 2 specialist nurses. In addition to this we have close links with the psychologist and the child development centre. We provide a metabolic service across most of Yorkshire in addition to Bradford. This ensures a wide spectrum of metabolic disorders are seen making the Bradford clinic one of the most varied and interesting in the country.

We work with the metabolic laboratory based at St James University Hospital, Leeds. The laboratory has lead on a number of innovations including being the pilot site for electronic messaging of results to CHR D through the CFH IT spine. This is now working well across Yorkshire. We all work very well together despite being based on different sites. The challenges of cross-site working have been overcome by formal quarterly multi-disciplinary team meetings, bi-annual network meetings and informally by regular phone contact.

Expanded newborn screening is important to allow early detection of treatable disorders. This is highlighted by a case that the Leeds laboratory and Bradford clinical team were involved in.

We recently had a positive case. The Leeds laboratory phoned the result through to the one of the Bradford Metabolic consultants who initiated liaison with local services. When the infant was seen in the Bradford Metabolic clinic, the result was confirmed and diagnosis and treatment were explained to the parents. Parental preference was for care to be provided locally. This was possible due to the comprehensive guidelines from the expanded newborn screening website and our experience in managing patients who live in a wide geographical area. We anticipate that this early detection will allow this child to grow up without the complications of this potentially serious condition.

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