



**Save Babies Through
Screening Foundation, UK**



Report of the 2011 LSD World Symposium 15th to 18th February 2011

Introduction

This is the report of the 2011 LSD World Symposium. Save Babies Through Screening Foundation in the USA kindly sponsored my attendance at this event. It was a significant learning opportunity for me. Although I have a working knowledge of Krabbe Leukodystrophy and of course newborn screening, nevertheless I had a significant amount of pre conference studying to do around other Leukodystrophies, genetics and other subject areas to enable me to keep up with the many presentations. The pre conference studying proved extremely successful.

Also I was fortunate to be invited to deputize for Micki Gartzke at the LSD Newborn Screening Transitional Research Network Meeting (NBSTRN) Working Group on 15th February which again was a significant learning opportunity. I was also invited to share with the group the current situation on newborn screening in the United Kingdom. I was pleased to meet Lori Wise, a patient advocate for Fabry disease who was also invited to attend the meeting. It was interesting to hear of her family experiences and for us both to have the opportunity to give the patient perspective.

The UK is lagging behind many other countries in terms of newborn screening and there are many barriers and difficulties still to overcome in the UK. We are running about 10 years behind the USA in terms of screening which is disappointing. However being a small country with a National Health Service in place we do actually have much of the infrastructure in place and some of the issues being addressed by the NBSTRN in terms of standardization and harmonization are not relevant to the UK. On the other hand we have no competition to invigorate our Department of Health to move quickly forward in terms of NBS.

Strangely it took me all the way to the Symposium in the USA to sit down with some nurses and doctors from hospitals in the UK to speak about work that was progressing in both newborn screening and on Krabbes disease and to drive out some more people that I needed to connect with in terms of both areas of work.

These are the main headliners from the 2011 World Symposium:

Newborn Screening Transitional Research Network 15th February 2011

Group members gave a number of excellent and informative presentations in terms of their work to improve technologies and diagnosis in different State laboratories, the new platforms, assays. DNA products for testing, the outcomes from testing for LSD's to date, follow up and some of the difficulties encountered around diagnosis. I was surprised to learn how rapidly newborn screening for LSD's is being introduced in some States and there was significant discussion on the relative merits of different testing methodologies, outcomes, the LSD's that were scheduled for NBS rollout and future work. I was very much impressed on how information was being shared to attain the best outcomes for patients as NBS for LSD's was moved forward.

I was naturally a little lost with some of the scientific information being discussed, however a number of the presenters have kindly agreed to share their slide presentations with me to assist my future understanding and learning. Sincere thanks to Michael Watson, Joe Orsini, Dieter Matern, Patti Duffner and Amy Brower for this. Thanks to Rodney Howell for his directions and assistance with my work on NBS for Krabbe in the UK. Thanks also for the generosity of other Group members in sharing information with me as we proceeded through the World Symposium Agenda and presentations.

The many aspects around the goal to develop a state of the art technology system for the gathering, storage, retrieval and access of newborn screening data critical to the work of laboratories, to research and to support clinicians across the USA was on the agenda and discussed in detail. This included discussion on many of the challenges e.g. resources required, development of the system, the many sources of data, driving down the levels of data, protocols, data standardization, adaptability of the system, crossing State lines in terms of data sharing, parental consent to enable data sharing, legal and ethical issues, approvals processes and the commitment to maintain the data over a number of years. Also securing sufficient numbers of patients to actually test the data and building a resource of scientists to determine exactly how the data can best be used. It was agreed that IT expertise needed to be secured to advise on many of these challenges. Discussions were in progress on this as well as the fact that there were lessons to be learned from some of the databases established by the pharmaceutical companies for certain LSD's.

I learned a lot about the economic and resource factors in respect of newborn screening and the challenges that had to be overcome. Importantly the cost of testing and particularly DNA testing. This information was of great importance to me personally as without doubt, one of the big challenges for us in the UK is ensuring that we have full and complete cost benefit analysis to support the extension of our newborn screening programme. This cost benefit analysis has perhaps not been done as well as it might in the past and in the current economic climate is critical for the UK.

Following the meeting I was able to share and discuss the many action points and outcomes with Micki Gartzke (who I had deputized for), including the decision to set up disease specific sub groups Each having clinical, molecular, bio chemist and newborn screening experts within each group.

2011 LSD World Symposium Report - 16th to 18th February 2011

The presentations were over the 3 days in 3 categories, **basic research**, **transitional research** and **clinical research**. I was originally disappointed that the initial session on 15th February on the fundamentals of Lysosomal Storage disorders had to be postponed to next year. It would have been great for people like me, i.e. with no or little medical or scientific background. However I have to say that it was more than compensated by what I learned from the professionals at the LSD NBSTRN meeting.

We had an inspiring keynote address by Dr Steven Walkley, who received the **Award for Innovation and Accomplishment** in the field of lysosomal disease, research and therapy.

Dr Walkley spoke about the definition of a rare disease, what rare disease teaches us and why we study it. He went on to outline some statistics on rare disease in the USA, in Europe and the world. He pointed out that if we brought together all rare diseases they would in total be as prevalent as diseases such as Cystic Fibrosis. He gave us 5 different reasons for studying LSD's and I hope that I have captured these correctly:

- They are experiments in nature that we can analyze. We can explore the medicine of them. We have learned what the lysosome is all about. Now we need to think about genes, the relationship of lysosomes and other areas that feed in, other storage functions and the importance of degradation compounds, ectopic dendritogenesis, ganglioside storage and other areas.
- It will give us critical insights into other more common diseases e.g. Parkinson's Disease, Alzheimer's Disease (e.g. where there are similarities to Niemann Pick)
- It could inform potential new therapies for more common diseases e.g. statins were originally designed to treat rare disease.
- It will open up new ways of doing research, highlighting some of the issues in terms of diseases being too rare to stimulate the scientist's interest, too few patients for effective clinical trials and sometimes in the past the lack of interest from pharmaceutical companies.
- It may say something about ourselves, i.e. we are none of us 'normal', we all carry different genes that either makes us healthy or unhealthy. We live in an imperfect world and we are trying to make it better.

I have not attempted to outline the context of all the presentations over the three days however just to say that many of the LSD's were covered in detail. Some of the information I could comprehend and absorb really well and some I found difficult to understand. One of the things that I did flag in my feedback form on the conference was the pace of the presentations. Presenters had 15 minutes a subject and often the presentation slides were progressed too quickly to assimilate the information and/or the presentations were hurried and garbled. I recognize that this may not have been as much of an issue for the clinicians and scientists who specialised in LSD's. Some presentations were excellent both in timing and message. I have more detailed notes if these will be useful however they will take some time to transcribe.

Activities and Networking Meetings around the Symposium

1. I met with Dr Maria Escolar and Dr Michelle Poe in respect of the position on Krabbes disease for patients and families in the UK. I had developed an outline business plan on a proposal to hold a scientific conference in the UK. The position is that Krabbes disease is generally diagnosed when symptoms present, unless a parent has already lost a child when there is pre natal testing available. There is no newborn screening programme in the UK for LSD's. Babies who are symptomatic shortly after birth are given palliative care only until they die.

Dr Escolar was pleased with my Business Plan and will speak to Micki Gartzke in greater detail on how they can support my objectives.

2. I attended the Council for Patient Advocate lunchtime meeting which was generated so much information, ideas and business generally that we had to adjourn and meet again in the evening. The meeting was chaired by Dr Elsa Shapiro. The Lysosomal Disease Network wanted to achieve specific things from the meeting in terms of patient group support for some of the research projects, however the patient representatives wanted to develop this much further. We were able to have a central contact point and exchange contact details. I was able to offer advice on how the UK LSD Collaborative Working group functioned and some of the things that could be achieved. There was an issue of scale of course. We are only a small country which assists good communications.
3. Prior to the meeting I had been in communication about newborn screening policies with Dr Bridget Wilcken from NSW, Australia. Dr Wilcken is trying to assist with a piece of work I am doing in trying to build models of different screening policies in other countries to try and influence the UK government on NBS programme expansion. She had asked John Forman who is Executive Director of the New Zealand Organization for Rare Disorders to look me up at Symposium. John also sits on the NBS Advisory Group for NZ and works closely with Dr Wilcken on NBS issues.

We had a useful lunchtime meeting with offers of help with my modelling work. Also the UK LSD collaborative has been awarded a grant to do some research on the transition from child to adult LSD services within our National Health Service. Our work on this is underway. John had also done some work on transition, albeit on a smaller scale and agreed to share this.

4. I met with Dr Joe Orsini to discuss the application process for submitting Krabbes Disease to the National Screening Committee (NSC) in the UK. Dr Orsini kindly had a look at all the necessary requirements. He advised that we are in a position to complete a lot of the submission form but had not all the necessary information available that is required. I spoke to Dr Howell who has now pointed me in the direction of information in the USA which we have now shared with Dr Orsini. I am trying to commence some work which will hopefully lead to a submission on newborn screening for Krabbes Disease to the NSC however I still require a clinician sponsor in the UK. It will without doubt be rejected however the earlier we start the work the earlier we get it on the agenda.

General observations

LSD World Symposium is an ideal learning opportunity. The amount of information is overwhelming and it is great to learn what exactly is going on across the world. It can be really inspirational coming from a country that on one hand are leaders in research, however on the other hand seem to be really well behind on other fronts e.g. NBS.

One of the really beneficial things is the opportunity to network and have dedicated meetings outside the formal sessions. I got so much work done there.

Circumstances prevented me having a 'buddy' there at Symposium. I would have found that so useful. I found out later that there were people present who I would really have liked to meet and speak with however I did not know who they were.

Pat Roberts
Executive Director

March 2011