



Batten Disease Newsletter

Bringing light to Batten's

Issue 19 Autumn 2009

from the UK's only dedicated Batten Disease support and research charity

Our London Marathon Star

Batten Disease Family Association

Registered Charity No. 1084908

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Dear Members

I hope you have all managed to find some enjoyment over the Summer despite the British weather! Summer holidays for us as a family over the last few years have been an opportunity to enjoy what we have, reflect on what we have got and can do in the positive sense and not what we haven't got. It does make a real difference to us in being able to cope with the rest of the year.

'Over the next 5 years, the BDFA will manage over £1 million of funds'

It is also an opportunity to reflect on what has been happening at the BDFA. You will see in this edition of the newsletter what the BDFA has been up to in the last 6 months. Some members raising the Batten Disease profile by attending the Rare Disease Day at the House of Commons in February as well as lobbying them again in June on our 1st Batten Disease awareness day. We held our 1st I/LINCL training workshop for parents and professionals in March at Guy's and St Thomas' NHS Hospital and Jan and Heather Band represented the BDFA at the International NCL congress in Germany. In our Batten Bulletin, you will see our new bright-orange T-shirts and Banners that have been a tremendous success and they have been used, most noticeably, at the London Marathon and the annual Batten Sponsored Walk. Many of you, your family & friends and our professional advisors have supported us at these and many other events. Thank you so much to all who have been involved in any of the above as it does make a great difference.

Over the next 5 years, the BDFA will manage over £1 million of funds, broken down as raising funds over £500,000 and allocating these funds wisely both in providing better support and facilitate more research. Much foundation work has been done to manage this effectively and we continue to build on our foundation stones with the right ICT structure and continuing to increase our paid support base with the introduction of our Family Support Officer, Sam Simpson, from September, generously funded for 2 years by the Roald Dahl Foundation. Sadly, however, we will also be losing Jan Sablitzky, our part-time Development Officer and first paid employee. Jan has been the backbone over the last 3 years in setting up a lot of the key foundation stones needed for the charity to grow and achieve our Vision and Strategy. She will be greatly missed and we wish her well in the future and thank her for her outstanding contribution to the BDFA and Batten Disease UK. So we have now brought forward our strategic objective of the recruitment of a part-time Charity Manager to start in Spring 2010 in order to continue the excellent work Jan established.

The next two years are critical in ensuring the charity, facilities for better support and research for Batten Disease in the UK, continue to happen. I hope to see many of you at our October Family conference and AGM, where I can update you further with our plans and exciting new initiatives. I hope you can also stay over on the Saturday night at the Apollo Hotel to enjoy a social evening together.

Finally, thank you for your continued support as a Member... it does make a real difference. Do keep in touch and if we can help you in any way, please contact us.

Julie

The Pickering family on the UK's first Batten Awareness Day 6th June 2009

Be a BDFA Volunteer

Anne, who helps us with our grant opportunities research, has sent us a few lines to share with anyone else who would like to help the BDFA in this way.

'It is a privilege to help raise funds and awareness for Batten disease. Smaller charities need all the enthusiasm and drive of volunteers in these challenging times. It is also great to work with fun-focused people for a good cause.'

We are looking for volunteers for particular tasks ideally close to our paid staff BDFA offices near Norwich and Ashford. If you would like to help, just get in touch with Georgette at the BDFA Fundraising Office on 01603 760111.

What being a BDFA Member is about

Ten years on since the BDFA was first set up in 1998, we are the only patient support and research organisation for Batten disease in the UK. Membership really matters. Being a member of the BDFA is about showing your support for our work and getting support through fully accessing our services. Membership helps create a wider visible network of families and professionals across the UK. It gives us a louder voice for Batters, both in the rare disease arena and to support our scientists in their research. Whether you are a family member or close friend, carer or professional, please fill out a membership form today and ask everyone around you to do the same and help make a difference. The work we do is for you. We can't do this without you. **It's just £10 a year.** This helps cover the costs of keeping in touch. Paying your £10 annual membership subscription by standing order really helps too! **Thank you.**

[Cover photo: A special cuddle. Lewis Crouch and little daughter, Tia, at the end of the marathon day].

What a star! Lewis Crouch, our first Silver Bonded Place London Marathon Runner, successfully completed the London Marathon in 4 hours and 45 minutes, and has raised a staggering £12,000 to help the work of the BDFA. Thank you to Lewis and his family, to everyone who generously sponsored Lewis and to everyone who came up to London to cheer Lewis on.

The BDFA's Vision is to bring light to Batten Disease by being the central point of excellence in the UK for supporting affected families and to facilitate research into the disease

TRUSTEES NEEDED

Do you or a close friend, relative or supportive professional have time, skills and commitment to support our charity's work as a volunteer Trustee and help us achieve our Vision?

The BDFA is developing rapidly and to help govern the charity effectively whilst the paid support 'do the doing', we are looking for more Trustees. We would really appreciate another Late Infantile or Infantile parent getting involved. Also, if you have any marketing/PR/grant application skills to help with our fundraising activities as part of the Fundraising Team, or any scientific background and expertise to help with our research efforts as part of the Research Team, we can put them to very good use!

If you feel you might be able to help as part of the BDFA Team, at this very exciting time for our small charity, please find out more by contacting: Julie Pickering via: info@bdfa-uk.org.uk or telephone: 01603 760111 or writing to: BDFA c/o Heather House, Heather Drive, Tadley, Hants RG26 4QR before 10th September 2009 for our Trustee's information pack and for an informal discussion.

To read more about our activities and plans, have a look at our current Trustees' Annual Report – the next report will be out at AGM (see page 12).



Some of our Trustees and Paid Support at our last AGM 2008

From left to right, back row: Marion Bayliss, Pauline Muncey, Heather Band, Sarah Kenrick, Julie Pickering, Liz Gatrell. Front row: Jan Sablitzky, Heather Sickelmore, Alison Jenkins, Philip Glazebrook, Georgette Ward

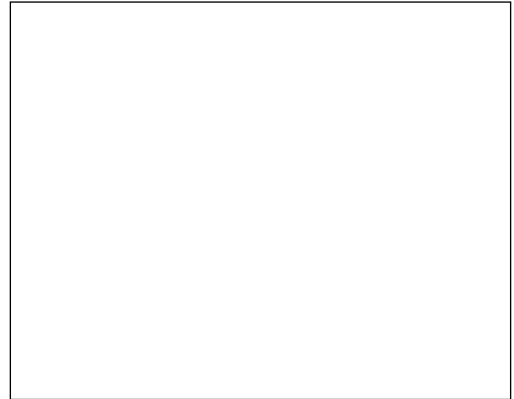
A special thank you

The BDFA would like to thank those Trustees who stepped down at our last AGM in October 2008:-

Rachel Griffith who, as Chair 2005-07, helped us manage the transition when our first paid support was introduced as a key milestone for the BDFA. Rachel continues as a BDFA LINCL family Befriender.

Steve Boxall, who in his time as a Trustee helped us get our current website set up and had the idea of having a BDFA Sponsored Walk which has raised over £65k in the past four years since the first posse did Bodmin Moor... Steve continues as a BDFA JNCL family Befriender.

Our new Family Support Officer



In September, the BDFA makes another key step forward in the resources it can offer families and professionals with our new part-time Family Support Officer, Sam Simpson, starting work for the BDFA. This coming strategy year, Sam will be working directly with families and professionals as well as behind-the-scenes developing family-friendly information and helping to plan future services. Do come and meet Sam at our Family Conference in October...

Hello, I'm Sam, the BDFA's new Family Support Officer. I'm delighted to have been given this opportunity to work with the BDFA and with families, carers and professionals involved with children and young people who live with Batten Disease.

I'm a qualified Social Worker with experience of supporting families and carers, working with children, young people, vulnerable adults, and older people. I went to university straight from school and studied for my degree in English and American Literature. After this, I began working for Kent County Council Social Services Department where my interest in Social Work grew. I was fortunate enough to be sponsored by KCC to do my Social Work training and qualified in 2005. I worked full time for KCC up until July 2008 when I left to go on maternity leave.

I live in Kent with my husband Richard (an estate agent) and our baby boy, Elijah, who will be one in August. Our dog 'Snoop' is a very important member of the family and spends his time keeping our four cats and four rescue rabbits in check!

I believe that I am joining the BDFA at a very exciting time. I am committed to working to maintain and develop our support services so that families and professionals can feel that there is always somewhere to turn at each step of the Batten's journey. I am looking forward to meeting and speaking to you all in the coming months. If you have any questions or suggestions for developments in the BDFA's support services, then please do not hesitate to get in touch.

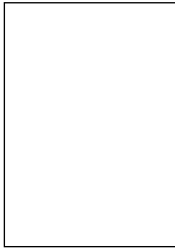
Sam

Sam Simpson, Family Support Office email: support@bdfa-uk.org.uk

We are delighted to report that this post is funded for 2 years by the Roald Dahl Foundation – read more about this exciting partnership in the Spring issue of our Batten Bulletin.



Development Officer Update



Jan doing her Batten's Awareness Day thing in bright orange at the NCL2009 event (see page 11) [blurry shot as Jan is always dashing about!]

Hello, As you may know, back in September 2006, the BDFA recruited me as their first-ever employee, to be responsible for: developing, maintaining and promoting the services offered to families, carers and professionals involved with children and young people who have Batten disease.

My job has naturally morphed and evolved to meet changing demands as the BDFA has grown rapidly over the past 3 years – an intended outcome of my 'Development Officer' role! In that time, I have done practically everything from licking the stamps on thank-you letters to donors, manning our support helpline and attending research and networking events. Funds – both donations, registered membership, support resources, events and grants for Battens Research have all increased significantly. It has been wonderful to have this visible support and widening participation in our charity's activities to help our beneficiaries.

Last year, it was great to have Georgette, our Fundraising Officer, join the team to take care of that side of things. This enabled me to make more excursions out of the BDFA office to 'take Batten's out there' and take on the role of coordinating the Support Team's work to build on existing activities and introduce new initiatives in response to the findings of the BDFA Family Support Project. I am therefore delighted to welcome Sam Simpson, as the BDFA's new Family Support Officer to focus on providing and further developing our support services portfolio to families and professionals, both in-house and in liaison with statutory care, health and education authorities.

Working with the dedicated team at the BDFA has always been very rewarding. Working within a small organisation during a period of rapid change has proven invaluable experience and insight into the day-to-day activities needed to be a proactive charity and its potential strategy for future growth and development over the next few years and beyond.

I am so pleased to have made my visible contribution to the BDFA's development and there are exciting times ahead for improvement in services and progress in research. However, I am now returning to the Higher Education Sector where I worked for many years before moving from London to bring up my young family and then helping the BDFA for three years.

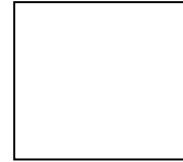
It has been a very great pleasure and privilege to have met such wonderful families and professionals and the charity will always be special to me.

Jan

Jan Sablitzky
BDFA Development Officer 2006-09

Living Life with Battens

A chance for parents, carers and professionals of children and young adults with Juvenile Batten Disease to come together from across the UK for support and get key information.



Here at the BDFA we are always looking at ways to develop our support services to families and professionals caring for children and young adults with Batten's. Building on the success of our new 'training' opportunity for parents of INCL/LINCL children in March 2009, we are now developing, in liaison with our professionals' group advisors and other families, two more events designed for JNCL families with children and young adults at different stages of Batten Disease. We hope to run the first one in March 2010 at Guy's Hospital. Through these unique events, we hope families will feel encouraged and supported in managing the disease for their child and their family's quality of life now and in the future as they plan the next steps together. Please contact Sam, our Family Support Officer, for further information.

Did you know?

Aiming High Participation Fund

Aiming High for Disabled Children is a three year project to transform services for disabled children and young people. A key aspect of this is increasing the amount of short break activities that are available which disabled children and young people can get involved in.

The idea behind the fund is to enable disabled children to participate in activities that they would like to do, but where something is stopping them from joining in. Where a short break activity has been identified, but a barrier exists that is stopping a disabled child joining in, an application can be made to the fund to have that barrier removed. Common barriers are lack of transport, the need for an extra worker to support the child as they do the activity, training for the staff at the activity to meet the child's need, or a piece of equipment the child needs in order to join in.

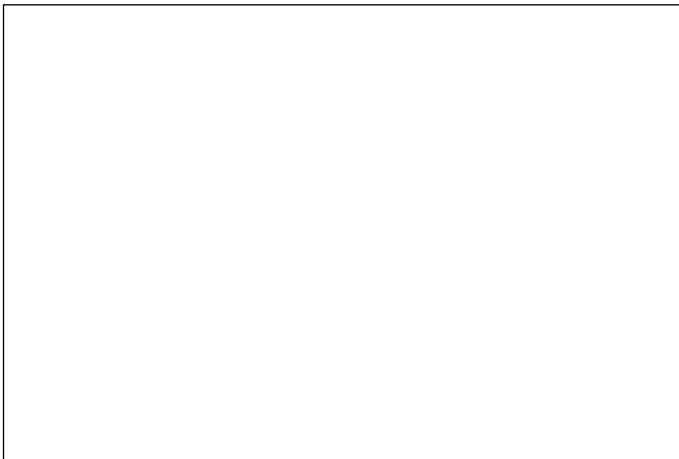
These are just examples. If you identify other barriers, an application can be made to the fund to have them removed. You can apply for funding if your child is disabled, you know what activity they would like to do, you are clear what the barrier is that is stopping them from doing it, the activity will give you a break from caring and one of the people who works with your child supports your application.

Applications to the fund can be made by downloading a simple application form from www.sncwd.org.uk or by contacting the Aim High Programme Manager, Karen Parry, on **0191 566 2190**.

Getting the support you need

At the BDFA, we know what a challenge it can be for many families to access the services and resources required to help care for their child's increasing and complex needs. Our Family Support Project findings demonstrated that access to and benefit from services is highly variable around the country and there is an element of luck in being referred to anyone with a knowledge of Batten disease as it is so rare. This is something that the BDFA is trying to do something about at NHS service commissioning level in the coming months to benefit all families across the UK, so please watch this space.

In the meantime, we wanted to share with you here one family's positive experience that demonstrates what a difference it can make when things happen as they should happen. Please remember, do contact us for support and information at the BDFA too as we will always try to help and if we can't, signpost you to someone who can. So, please give Sam, our Family Support Officer, a call or drop her an email...



Being fairly new to this community, Victoria and I took another step on our rollercoaster journey in March when we decided to attend the BDFA's new training day for Infantile/Late Infantile families and professionals in London. We are glad we did, if only to confront our demons and we came away knowing that we are not alone. Speaking to others whilst we were there, made us realise that the coordinated care that we have received in Lancashire appears to be better than others, so I promised to share our news with you so that you may be able to put pressure on your local authorities to improve the standard of care you receive from a clinical, educational and social care point of view.

The first point to make is the proportionally short time it took to receive a diagnosis. Epilepsy for Harry was first diagnosed in December 2007 following the usual indicators of slow development, with the full LINCL diagnosis 6 months later. This was in part due to the commitment of the clinicians at the Royal Preston Hospital especially Dr Basu, the Paediatric Neurological Consultant, and her broad approach to exploring all possible outcomes by securing early EEGs, MRIs, CT Scan, Lumbar Punctures and so on but also in part due to Victoria's dogged research on the internet; the result of which was to specifically ask if a test for LINCL could be done. Following a hastily arranged appointment with the Consultant Geneticist in Manchester, the tests sadly proved positive on Midsummer's Day 2008.

Simultaneously, our life plans were being turned upside down. We had bought a house into which we were going to move that summer, the children had been booked into a different school and our careers were taking us out of the county. All change. Dr Basu initiated the provision of wider care by linking with Sue Pigeon, the Specialist Health Visitor who became the central coordinator. The first priority was to secure school places, which was made easier due to our bad administration because we had not formally withdrawn them from the school. Added to that, the Local Education Authority appraised the school of the circumstances, confirmed there were epilepsy trained staff at the school and, more importantly, with the assistance of the Educational Psychologist, generated a full Statement in 8 weeks.

'we have been fortunate to have received coordinated wrap-around care that seems to have covered all aspects of our changing lives'

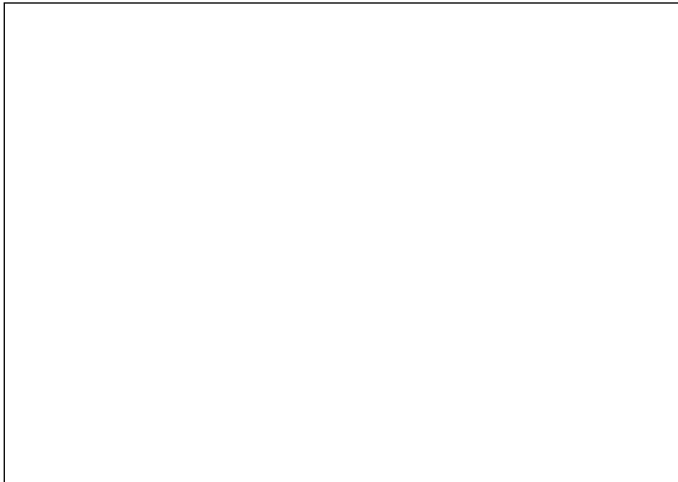
The provision of social care has been equally efficient. Early engagement from a very supportive and proactive social worker has resulted in an excellent care package that provides 40 hours of care per week. The social care worker and the clinicians all talk to the Occupational Therapist and the OT has been equally proficient at organising a specialist car seat, a buggy, a chair for the bath, with more to come to assist as Harry's disabilities increase. This formidable team are also assisting us in choosing a specialist school for Harry to which he will start to make a gradual transition after the summer. Other individuals who have been involved are the physiotherapist, the Sapphire Nurse, a dietician, Child Occupational Therapist, a speech and language therapist, and a psychologist for us. As we write, Harry is recuperating from having his gastrostomy which had been arranged in February but which we delayed until the last possible moment. But again, this process was efficiently organised.

As an Army family, our housing is provided by Defence Estates, an agency of the Ministry of Defence who are mandated to provide for our needs. Soon after diagnosis, we engaged with DE to alert them to the possible requirement for the provision of specialist accommodation and pulled together a meeting between DE, the OT and us. The result of this has been an agreed adaptation to the house which will provide for disabled access, a specially adapted bedroom, sluice / bathroom and area for the carer to rest for overnight stays. The OT is critical in setting the requirement and DE should be obliged to meet the criteria set in the OT report. Building should start soon.

Given the circumstances, we have been fortunate to have received coordinated wrap-around care that seems to have covered all aspects of our changing lives. And all of this is a direct result of the independently agreed multi-agency approach that runs in Lancashire, and the importance the Authority seems to place in children's welfare. It makes the difficult journey that little bit easier – so hats off to professionals in the Central Lancashire NHS, NHS Foundation Teaching Hospitals and Lancashire County Council.

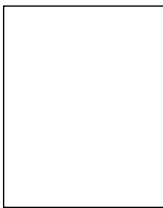
Anthony Millsop

Editor's footnote: Sadly, at the time of going to print, little Harry's condition had seriously deteriorated. Our thoughts are with the family and we thank them for their continuing permission to share this article.



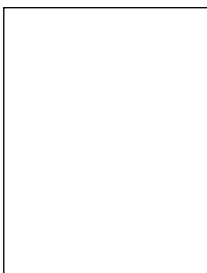
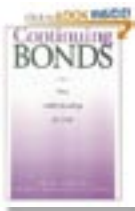
The BDFA's special Remembrance Book

As part of our support to families at every stage of the Batten's journey, we believe families deserve the best emotional care in such a crisis and offer a listening ear when you need it so you have space to talk about and remember your child. If anyone would like to make a tribute page to be added to our special Remembrance book to share good memories of their child, please get in touch so we can arrange for further information and for a page to be sent to you. The Book will be available at our Family Conference in October. We can also put you in touch with other families who have lost a child through Battens. Here, one Mum, shares her experience with us...



Today, I am comfortable with my grief. I am fully engaged with my fulfilling life as a practising counsellor, family support co-ordinator in an adult hospice, and a licensed lay minister in the Church of England. But I am also entitled on occasions to still feel sad and shed some tears over the life and death of my beloved youngest daughter, Lindsay (1983-1992), who had Late Infantile Battens Disease (CLN2).

To my mind, the book 'Continuing Bonds' edited by Dennis Klass (Compassionate Friends) and Phyllis Silberman, published in the late 1990's revolutionised professional thinking about bereavement and affirmed what I already knew as a parent - the grief doesn't end so that we are restored to normal functioning. We re-engage with life whilst absorbing our loss within our being so it becomes part of who we now are, because the emotional bonds to our children cannot be broken and they are never forgotten.



Since Lindsay died in 1992 there have been many milestones in my grief, and not always the obvious ones ending with 0s or 5s. The ninth anniversary was harder than the tenth because she had now been dead more years than she had lived; 17 was as hard as 18 because I had always been so involved with teaching my kids to be responsible drivers; leaving her historically in the 20th Century as

we celebrated the new Millennium was particularly painful. Because the death of a child goes against the natural family order of life, how can I not be aware of her absence at every significant family celebration? She should still be here now, preparing to play her part in her older brother's wedding later this year.

So yes, there will continue to be moments of sadness as the years pass, but I claim them as my right as much as I celebrate the very rich and rewarding paths life has taken because of the experience of living with her, loving her and losing her. Without her I am quite sure I would be none of the things I am today, so I embrace my grief and move forward with her ever present memory as company on my life's journey. This is my mantra, and I hope all those reading this will one day be able to share it's sentiment:

"The folk who look happiest and those who seem bright - with smiles on their faces and feet that are light - are not always those who have lived in the sun, but those who faced darkness and fought it, and won!"

Angela



IN LOVING MEMORY

Our thoughts and prayers are with the families and friends of:

Saul & Savannah-Rose Fraser, aged 7 (Infantile)

Robbie Hollern, aged 6 (Infantile)

Jay Theobald, aged 12 (Infantile)

Supported Networking with other Battens families available through the BDFA

The Batten Disease Family Association have traditionally aimed to ensure that "No family will go through the devastating journey of Batten Disease alone" and network families in similar circumstances to their own - if they wish.

If you would like to join this informal family networking group, which links up families across the UK by Battens type, with a Befriender to support you if you wish, please email our Support Team at: support@bdfa-uk.org.uk for more information and a BDFA Membership Registration Form. One of our Befrienders will then contact you to say 'hello' and give you details of other families in the BDFA you can get in touch with...

Notes from our Medical Advisor

Dr Ruth Williams, Medical Advisor to the BDFA
Consultant Paediatric Neurologist at the Evelina Children's Hospital, GSTT, London

Ruth will be at our Family Conference in October to chat over coffee or sign up for a 1:1 session during the event (see programme on page 12 for further details).

In March, we were delighted to meet Emma Williams, from Matthew's Friends and we wanted to share some information about the ketogenic diet you may have heard about...

What is the ketogenic diet for epilepsy?

Ketogenic diets were first used for epilepsy in the 1920s before anti-epileptic medications (AEDs) were available. The diets were hard to follow and when phenobarbitone, phenytoin and later valproate were discovered they fell out of use apart from in a very few centres. Despite the availability of a very large number of AEDs now, many children will continue to have seizures frequently despite treatment. Children with all kinds of Batten disease (NCL) often fall into this group.

A ketogenic diet is a high fat, low carbohydrate diet. Enough protein is included for children to grow and vitamins and minerals are supplemented. Several versions of the diet exist and a modified version of the popular slimming diet, the Atkins diet, can be used in adults. Ketogenic diet formula feeds are available, so that a ketogenic diet can be given to those with nasogastric tubes or gastrostomies. About 30% children with frequent seizures on medication will have a significant improvement in seizure control on the diet. About 30% of those starting do not continue because they cannot tolerate the strict diet or because of side effects.

Ketogenic diets are not available in all hospitals. They require a team of professionals experienced and skilled in looking after children with severe epilepsy and including ketogenic specialist dietitians. The diets are tailored for each child individually and children are monitored very closely with blood tests and EEGs. About 15 children's hospitals are able to offer ketogenic diets in the UK and most have a long waiting list. At the Evelina, I run one of the largest clinical KD services in the UK and at any time we have 25-30 children on the diet.

More information is available on the following websites and on request from myself:

Ruth.williams@gstt.nhs.uk

Dr Ruth Williams, Evelina Children's Hospital, London
Medical Advisor to the BDFA and Medical Board Matthew's Friends



Charity Number: 1108016

Never in my wildest dreams did I expect Matthew's Friends (MF) to grow at the rate it has done over the past five years. The little charity that I set up in order to support families that wanted to try a dietary treatment to manage their child's epilepsy, took off at a rate that none of us expected.

My education into the world of epilepsy started back in 1994 when my son Matthew was born and he developed seizures at nine months old. My education proved a very steep learning curve as none of the medications that were meant to control epilepsy worked with him and if anything, they made things a whole lot worse. It was only when I put Matthew on a Ketogenic Diet when he was seven years old that we actually saw a vast improvement, this was in spite of the fact that for the previous six years I had been told that it didn't work and I was made to feel terribly guilty for even suggesting subjecting my son to something so awful, all of which turned out to be totally false. The diet wasn't disgusting and it DID work. The devastating effect 6 years worth of uncontrolled and prolonged seizures had on his brain means that today he only has a mental age of about 15 months old. Obviously when the diet reduced Matthew's seizures by nearly 90%, I was highly delighted, but also extremely angry that I had been denied a treatment that could have helped my son a lot earlier and maybe, just maybe he wouldn't have suffered as much brain damage, hence the reason why I started Matthew's Friends. This treatment may not be for everyone but everyone has a right to know about it and a right to try it if the medications are not working and it is the reason why MF works with ALL epilepsy groups. We specialise in just one area – Dietary Treatments for the management of Seizures.

continued...

continued...

Obviously working with so many groups means that we have to be up to date with the latest information in relation to the various syndromes so that we can support our families in the best way possible. My education into Batten's came in the form of the beautiful Rhiannon, Shauna's little girl in Ireland, an incredible family whom I have chatted to over the years in relation to their use of the Ketogenic Diet to help manage Rhiannon's seizures and then, more recently, with Mel and Matthew. I remember vividly the phone calls from Mel when she first found out the diagnosis. We can read what we like in the books as far as Batten's is concerned, but when you are actually talking to the parents who are going through it, obviously things change your perspective and it all becomes very, very 'real'.

In the past five years, I have travelled across the globe promoting the use of dietary treatments for epilepsy and however many conferences I attend and meetings I go to I ALWAYS learn something else. Your own education never stops when it comes to epilepsy and the primary source of my education is the families, the absolutely awesome families that I am very privileged to know. There is simply no education that matches what we can learn from one another, which is why it was so good to come along to the Batten's training day that was held in March in order to meet other families and listen to the presentations that were made on living with Batten's and how best to manage the condition. It certainly increased my education of this syndrome.

So my message to you as far as the Ketogenic Diet option is concerned or indeed any dietary treatment for seizures, is think about it, discuss it with your medical team and for those of you who are not aware, Dr. Ruth Williams (Paediatric Neuro) is also part of the Matthew's Friends Medical Board, as well as your own – so you have a Keto expert on board with you to chat things through with. This option may not be for all of you, it may not work for some of your children but for others it will help and make the quality of your child's life better. You will find out a whole lot more about all the dietary treatments at our website: www.matthewsfriends.org – you will have to excuse it at the moment as we are in the middle of a huge re-vamp of the site and completely new one is on its way with lots more information – so keep checking in. We also have a busy and very supportive forum if you have any questions and please don't hesitate to e-mail me at: emma@matthewsfriends.org, I will try and get back to you as soon as I can.

It was an absolute pleasure attending the training day and thank you for a warm welcome and thank you for educating me. By my sharing the information I learnt from that day to the rest of the MF team, we will be able to provide a better support service for you. From everyone at Matthew's Friends we send you lots of love and trust in us when we say that if you decide to try the dietary option, we will support you all we can.

Take Care

Emma

Emma Williams

Founder/CEO Matthew's Friends, Parent Representative – KetoPAG

Hello, from our new Advisor



We are delighted to welcome Shauna Bates, as our new Social Care Advisor to the BDFA...

Hello, my name is Shauna Bates, and I have recently taken on the position of Social Care Advisor for the Batten Disease Family Association. I am very excited about this position and hope that I will be able to offer help to any of you who may need it.

I am a qualified Social Worker and have been practising for 7 years. I live in Derry, in Northern Ireland with my husband Alan, and my 9 year old daughter Rhiannon, who has Late Infantile Batten Disease.

Although I live in Northern Ireland, I want to assure you that any legislation that applies to the main land UK is almost identical to that used in Northern Ireland. In my experience, any legislation required to challenge various issues often refers to the U.N. Convention of Human Rights, which applies to everyone in any E.U. country.

I hope that I will be able to help in many ways. I can give advice on any social care issues, any difficulties that you may have accessing assistance, and help with any professionals involved in your child's life. I am also available if you feel that you just need a chat – I understand that having a child with Batten Disease can be overwhelming at times, so feel free to get in touch through the BDFA's national helpline.

Regards

Shauna

Shauna and Sam, our new Family Support Officer here at the BDFA, will be working to give the best possible information and advice we can to empower families. We are ever grateful to all our Advisors who dedicate valuable time to responding to enquiries. Using their professional expertise in Batten's in this way makes a real difference. They are also an essential component of BDFA events designed to help support other professionals and families. If you would like to meet Shauna and our other professional Advisors, come along to our Family Conference in October (see page 12).

Heather Band, Trustee and BDFA Research Team Coordinator, reports...

So much has happened in the last few months – it has been an exciting time for the Research Team of myself, Heather Sickelmore and Julie Pickering, our Chair. We have attended scientific meetings, started new collaborative projects and continued to award grants for Batten disease research (see the Research section of our website for more details on BDFA-funded research). The most significant event was the biennial NCL congress in Hamburg in June (pages 10-11), including the very exciting news that the next NCL meeting in 2011 will be in the UK. So, please read on to find out the latest research news.

Could you help?: We are hoping to recruit new Trustees, preferably with a scientific background to join our Research Team. If you are interested or would like to find out more, please get in touch with me at: research@bdfa-uk.org.uk or via the BDFA office.

BDFA supporting research – more research grants awarded

In response to our successful second call for grants in the Autumn 2008 and Spring 2009 rounds, the BDFA's Research Team have now awarded 9 grants totalling over £30K in Research Grants for Batten Disease to Research Scientists in the UK since our new research grant award scheme started in Spring 2008. We also allocated €5k in sponsorship of the international NCL 2009 meeting in Hamburg in June. We are committed to continue to increase our funds made available for research and it is envisaged that another call for grants will be made in Autumn 2009. For details of these new awards and all current BDFA-funded Research areas please see our website.

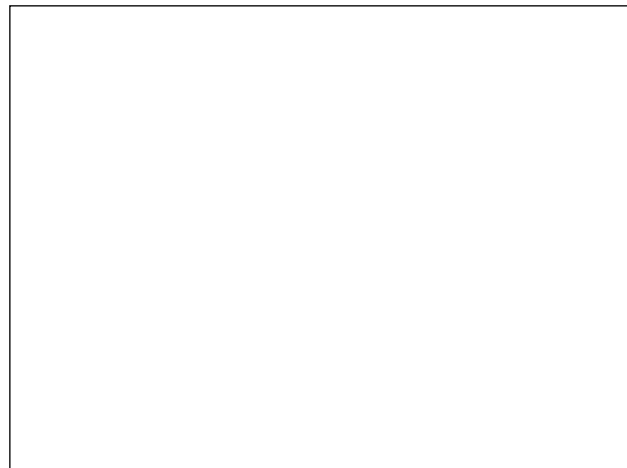
BRAINS for BRAIN

European Task Force on Brain and Neurodegenerative Lysosomal Storage Diseases

Third European Workshop
Frankfurt, Germany March 2009



B4B is a group committed to promoting scientific projects and to bring together scientists and clinicians from the fields of LSD and experts on the Blood Brain Barrier. Hence the name of Brains4 brain! A very important research area to consider when looking for the best approaches for potential therapies for Batten Disease. I was invited to attend the meeting in March and it was an invaluable opportunity to increase my knowledge, meet other scientists from the wider LSD scientific community and raise the flag for BD. You can find out more about the groups work in more detail www.brain4brain.eu



Jon and Heather raising awareness for Batten disease in Frankfurt



“HOT OFF THE PRESS” FROM NCL 2009

Pediatric Storage Disorders Lab Open Day
at King's College London, June 2009
Supported by the BDFA

This event went well, with a small group of families making the most of this opportunity to meet the scientists, our Medical Advisor and members of the BDFA team to catch up with what is happening in the NCL Research field. Here is one JNCL parent's perspective, Heather Sickelmore, who is also a BDFA Trustee on our Research Team...

I always enjoy the lab day. For me, hearing about the latest research gives me hope for the future. It's as simple as that. It's difficult to summarise what it's like to have a child diagnosed with Batten disease in a few words - I think, only those parents and carers that have experienced it can have any real idea of what it's like. But the lab day gives me an opportunity to listen, learn and hope that these very dedicated and clever people will one day have a 'eureka' moment and solve the mystery.

I am interested in genetics and I am gradually learning more and more - but (and I guess this is the scientist's experience as well), the more I learn, the more complicated it can seem sometimes. Having said that, every discovery moves us towards the ultimate goal. The lab day is also a great opportunity of meeting other parents and carers and members of the BDFA.

Heather



Historical perspective on NCL2009

As an NCL 'old-timer' I have been asked to provide a brief historical perspective on NCL2009. This meeting followed the pattern established since 1980 of holding NCL scientific meetings on either side of the Atlantic every two years. A full list can be found on: <http://www.ucl.ac.uk/ncl/meetingspast.shtml>. Hosting a meeting is a lot of work for the local organisers, which this time was shared between two clinicians

Prof Alfried Kohlschutter and Dr Angela Schulz and one scientist Prof Thomas Braulke. Prof Kohlschutter has had an extremely longstanding clinical and scientific interest in the NCLs, and although retired he was particularly keen to host this 12th international NCL meeting. He first hosted the meeting in 1992, which was before any of the NCL genes had been identified and therefore before much of what was presented scientifically this year could be foreseen – a sign of how much progress has been made. This 1992 meeting was a much smaller affair, and was the first NCL meeting I attended.

One of the things I liked about this meeting was that although its focus needs to be scientific progress, uniquely two of its hosts were clinicians who are also interested in research. The meeting was successful in that it attracted approx. 150 attendees, with 42 talks and 58 posters presented. To accommodate so many oral presentations talks had to be kept short, which meant they had to be focused – always a good thing. Posters were up for almost the whole time, even so it was impossible to read and discuss all in detail. Many novel findings since the last meeting were presented. Individually, each presentation, whether oral or poster, is extremely detailed and this scientific detail is of interest to only a very small number of attendees. However the significance of results reported is of interest to many attendees. For some, the NCLs as a whole are of interest, for others only results pertaining to one particular gene or mode of study are of interest. This is typical for a scientific meeting.

We look forward to hosting the next NCL meeting in the UK. As well as breaking the alternating Atlantic pattern, perhaps this next meeting will break the format in other ways, as well as being one full of new findings yet to be made. However, whether the UK can produce anything equivalent to the young unicyclists who were advertising their 'Wheels of Emotion' attempt to break the Guinness Book of Record for continuous unicycling (24 h) to raise money for Batten disease (see YouTube <http://www.youtube.com/watch?v=hs3VNXCo-oo>) remains to be seen – consider this as a challenge to be discussed and perhaps taken up!

Sara Mole

Dr Sara Mole, Reader in Molecular Cell Biology

MRC Laboratory for Molecular and Cell Biology, University College London
www.ucl.ac.uk/lmcb

Dr Ruth Williams & Heather Band enjoying the evening session held at the Zoological Department at Hamburg University.

Congratulations to Dr Ruth Williams for winning a prize for her poster "Juvenile NCL: What do we tell the children?" in recognition of her and her colleagues', Melinda Edwards and Sarah Kenrick, work on this important subject.

In a very generous gesture, Ruth kindly donated the €500 prize money to the BFDA to enable the production of a brand-new information sheet for families on this challenging topic which we hope will be out in the Autumn this year. Thank you from all of us at the BDFA.

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News from NCL2009, the 12th International Congress on Neuronal Ceroid Lipofuscinosis (Batten disease) Hamburg, Germany June 3-6, 2009

**Dr Jonathan Cooper and Members of the Pediatric Storage Disorders Laboratory,
Neuroscience, Institute of Psychiatry, King's College London**

Every two years there is a large gathering of scientists and clinicians who are working to understand Batten disease (BD). This is the one major opportunity for everyone working in this area to meet together, update each other with the latest scientific findings and what these mean. Most of the work that is presented is very new and hasn't yet appeared in a scientific publication, so it's a good opportunity to find out what is actually happening and what direction research is likely to go in the future.

Although many Batten's parents and representatives from parent organizations do attend the conference, the NCL Congress is at heart a scientific conference and unfortunately little concession is made for non-specialists. For example in Hamburg there were more than 40 talks, often given in 'science speak' and nearly 60 posters presented with very technical jargon. This can make it difficult to pick out the nuggets of what is important and useful from this vast amount of information.

Our report is meant to do this for you and concentrates on four key areas and describes what we learnt in each at the NCL2009 conference. These are: a) what goes wrong at a genetic level; b) understanding what goes wrong inside cells and in the brain; c) understanding the effects this has on people; d) how close are we to being able to fix these problems?

The full report can be downloaded from the PSDL website (<http://tiny.cc/i2WlIp>), but brief highlights include:

- Supposedly rare forms of BD are actually present all over the world
- Some of the genes mutated in BD make proteins that can be in more than one place and do several different things
- Getting proteins to the right place at the right time is critical for brain cells to work properly
- The synapse, via which brain cells talk to one another, is especially vulnerable in BD
- The glial cells that should help brain cells work properly are key players in BD
- There are several new BD models ranging from fruitflies and zebrafish, through mice, dogs and sheep
- Knowing more about how the diseases affect people will be crucial to judging the success of therapeutic interventions
- Several experimental therapies are being tried in animal models
- Two of these (gene therapy and stem cell therapy) have reached a clinical trial in human BD and immunosuppression for juvenile BD will be next (in the USA).

Taken together, the news from the NCL2009 meeting tells that a great deal more is known about the different forms of BD. Although progress may seem frustratingly slow, we have moved forwards a long way in our understanding in recent years. Although this has finally led to clinical trials of some approaches, we must remember that we are unfortunately some way from having any reliable means to cure these devastating diseases. Nevertheless, we all hope that more progress will be made before the BD research community meets again in the UK for NCL2011.



The Batten Disease Family Association warmly invite you to attend our annual Family Conference and AGM on Saturday 10th October 2009 at Heather House/Apollo Hotel, Tadley in Hampshire

'Supporting the Family'

The BDFA is the only registered charity in the UK consisting of a national network of parents, carers, friends, relatives and professionals in communities across the UK who are actively committed to supporting all families affected by all types of Batten Disease. We therefore welcome everyone to our 9th national conference as an invaluable and unique opportunity to come together for educated support. We aim to make the conference a highly informative and enjoyable experience; one which will be of benefit to the whole family. It is intended to give you practical tools and advice and the opportunity for you to be able to share your experiences with others whilst your children and siblings are cared for by specialist staff. We have also lined up good value 4* overnight accommodation. We will also be having a great social evening with a family get-together and evening meal on the Saturday night. We hope this will enable you to make the most of this opportunity wherever you are living in the UK.

We have an exciting agenda as detailed. We welcome our Guest Speaker, Anne Caldwell from The Children's Trust Tadworth, to share her professional experience with us on social care issues and how families can access resources. You can tailor your conference experience to your needs through options to: sign up for individual 1:1 sessions with UK experts in Batten Disease who are also Advisors to the BDFA; choose workshops related to the main talk; join in the afternoon meet-up groups or just stay and chat! Throughout the conference there will be information and library materials displayed covering a variety of topics around research, support and education, with members of the BDFA available to support any individual concerns and questions you may have. **We very much hope you can attend and look forward to seeing you in October...**

Supporting the Family

Event programme

9:30 *Children's and young people's activities start*

9:30 Registration & Coffee - 'Meet the BDFA's Advisor'
- 10:00 session sign ups

Optional 'Meet the BDFA's Advisor 1:1' sessions start for families as per times and rooms on the sign up sheet - in parallel to the main programme

10:00 **Welcome and Introduction to the day**
- 10:05 Marion Bayliss, BDFA Support Team Trustee

10:05 **How a Social Worker can help support the family**
- 10:30 Speaker: Anne Caldwell, Social Worker, The Children's Trust

10:30 **Workshop 1 Discussion Group for JNCL Families:**
- 11:10 *Which: Home or Residential Care? choices for JNCL families*
Facilitator: Anne Caldwell
Or

Workshop 2 Discussion Group for Professionals and the Family:
How to help LINCL/INCL families with social care issues
Facilitator: Shauna Mellon, Social Worker and LINCL mum and Social Care Advisor to the BDFA

11:10 **Swop-over and refreshments**
- 11:20

11:20 **Workshop 3 Discussion Group for INCL/LINCL Families:**
- 12:00 *Which: Home or Residential Care? choices for I/LINCL families*
Facilitator: Anne Caldwell
Or

Workshop 4 Discussion Group for Professionals and the Family:
How to help JNCL families with social care issues
Facilitator: Shauna Mellon

12:00 **Annual General Meeting of Registered Members and the BDFA Management Committee**

12:00 Welcome, Approval of AGM minutes 2008 and Resolutions

12:05 BDFA Summary and Future Vision - Chair's Overview

12:10 BDFA Summary and Future Vision - Team reports

12:25 Annual Report and Future Budget - Future Growth

12:35 Nominations for Trustees seeking election and re-election to join the Executive Committee and Trustee teams

Close

12:45 Buffet Lunch break

- 2:00

Afternoon Optional Forum sessions

'Meet the BDFA's Advisor 1:1' sessions continue
Children's activities continue

2:00 **BDFA Family Befriender Forum**
- 2:15 Facilitators: Sam Simpson, Family Support Officer
(funded by the Roald Dahl Foundation) Or

2:00 **Newly-Diagnosed Family Forum - JNCL**
- 2:30 Facilitators: Heather Sickelmore and Barbra Higgins
then

2:30 **Newly-Diagnosed Family Forum - I/LINCL**
- 3:00 Facilitators: Jane Parkinson and Julie Pickering
Or

2:30 **Adult Sibling Forum**
- 3:00 Facilitator: Marion Bayliss

Refreshments

3:00

- 3:30

then

3:30

- 4:00

then

4:00

- 4:05

4:30

'Meet the BDFA's Advisor 1:1' Sessions end.

Evening

7:00pm onwards **Family Get-together meal** in the Brassiere, at the Apollo Hotel, Basingstoke. Children's entertainer at 7:00pm.