BDFA
BATTEN DISEASE FAMILY ASSOCIATION

SPRING/SUMMER 2016

making a difference

from the only dedicated UK charity raising awareness, providing support and funding research into Batten disease

THE BATTEN DISEASE FAMILY ASSOCIATION NEWSLETTER

Contents

Diary Dates

Professor Jon Cooper's Lab Day at King's College London
10 May (see page 23)

Eurordis Membership Meeting
Edinburgh, 26 - 28 May 2016

Batten Disease Awareness Day
3 June 2016

Ben Nevis for Batten
24 - 26 June 2016

London 10k
10 July 2016

NCL 2016
Boston, USA, 5 - 8 October 2016

BDFA Family Conference
Holiday Inn, Stratford Upon Avon
18 - 20 November 2016

Don't forget we are on Facebook and now you can also find us on Twitter @BattenDiseaseUK

Can you help us achieve our vision: a world without Batten disease?

The BDFA is looking for individuals with the time, skills and commitment to support our vital work by becoming Trustees. We need candidates with demonstrable understanding and/or experience of the governance of a charitable organisation. In particular we would welcome candidates with any of the following experience/skills:

- Experience of fundraising, PR, marketing, networking and campaigning

The role can be based anywhere in the UK and commitment is expected to be as follows:

- Attendance at each bimonthly Board meeting (6 evenings per year)
- Attendance at the annual AGM and Family Conference
- Additional work averaging around 2-4 hours per month, on a flexible basis, to carry out related duties

Further information:

- The term of office for each Trustee is 3 years (with the option to stand for an additional 3 years)
- The role is voluntary and the Charity may reimburse out of pocket expenses incurred in the course of carrying out the role in accordance with the Charity Commission regulations.

A full copy of our strategic aims can be viewed on our website.
If you are interested in applying or would like to know more please contact Andrea West, BDFA CEO, **andreawest@bdfa-uk.org.uk** or **01252 416110**.

Letter from our CEO

I write my newsletter piece this time from California where I have kindly been invited to attend the LSD World Symposium 2016. For my four years in this role I have viewed other patient organisations attending this conference packed with research innovation in their particular disease with nothing on the programme about Batten disease. This year was very different and it is exciting to see so many more projects being presented both in posters and oral presentations.

The highlight of this week has been the presentation of such positive BioMarin data from their CLN2 enzyme replacement trial in London, Hamburg, Rome and Ohio. I watched Dr Angela Schulz present this data with much pride but also remembering those children and families for whom this will have come too late. We remember all of them when momentous achievements like this are made. Mel Hall, mum of Matthew who died in 2009, told Matthew that she would always fight this disease, and on hearing about the data told me:

"When I made Matthew my promise as he drifted away, this news is all I wanted to hear"

The report on this data and the rest of the Symposium can be found on page 19. From San Diego I will travel to San Francisco to meet with the BioMarin team, to give presentations on our work and the experiences of CLN2 families to the staff at their offices and to tour the manufacturing plant where the trial drug is being made.

We are sad to announce that our Batten Clinical Nurse Specialist at Great Ormond Street Hospital, Katie Hanson has left her role to take on another job at the hospital. This role remains incredibly important for us and we hope to have a new person in post very soon to continue to support families and grow the service offered.

Once again we give you a newsletter that is packed full of updates on our work, research and family stories. If there is anything you would like to see in the next edition please do get in touch via **admin@bdfa-uk.org.uk** as we would love to hear from you.

Andrea West, Chief Executive
01252 416110 / andreawest@bdfa-uk.org.uk

Letter from our Chair

Wow! What has happened to the last six months? I continue to be amazed by how much is going on and how much we achieve. The BioMarin announcement is a very exciting development and shows that we really are making progress. No one is getting carried away but it just shows that we are scoring goals along our journey.

Another major milestone of the last six months is Sophia kleine Holthaus's PhD – it is exciting work and we are all extremely grateful to Sophia for the effort she has put into producing such a fine piece of work.

As Andrea says, it is sad to report that our first CNS is leaving us. Katie, on behalf of myself and my fellow Trustees we say thank you and we wish you all the best for the future.

I continue to be so excited about how we are all raising awareness of Batten disease – whether it be at No.10 with the Prime Minister, a hospice with a member of the Royal Family or running a local fundraising event – so many people are doing so much to spread the horrors of our rare disease and again when I think how far we have come in the last six months it makes me feel enormously proud of the whole Batten family.

Somebody who has been at the core of our growth over the last six years has been Ellen Bletsoe. She recently made her first joint fundraising speech with her daughter Laura, at a lunch in the City of London. It was incredibly brave of both of them and we are so grateful for the £30,000 this lunch raised. A huge thanks must also go to Tim Martin who, in the space of 18 months, having never heard of Batten disease or the BDFA before, has raised over £50,000 and we are very grateful for his continued amazing support.

I wish all of you an enjoyable summer and please keep spreading the word, baking your cakes, running or cycling long distances and achieving amazing things in the hope that one day none of us will have to live with the horrors of Batten disease. We still have a way to go but we are making progress together and we can all be very proud of what we have done and what we will continue to do.

Michael O'Connor, Chair
michaeloconnor@bdfa-uk.org.uk

BioMarin – CLN2 Enzyme Replacement Trial

At the Annual World Symposium in San Diego the worldwide Batten community received some positive news from the US pharmaceutical company BioMarin about their data from the CLN2 (Late Infantile Batten disease) enzyme replacement trial. The trial has been ongoing in sites in London, Rome, Hamburg and the USA and the BDFA has been involved with our partner patient organisations in those countries to support families enrolled on the trial.

In summary these results showed:

- The trial measured decline of children's motor and language abilities and showed 80% reduction in progression of the disease at one year when compared to the data that is available for the natural progression of this disease.
- 87% of children on the trial experienced a slowing down of the progression of the disease.
- This means that based on the CLN2 rating scale (measuring mobility and language with a normal score being 3 for each) we would expect children to lose 2 points over 48 weeks. Children on the trial lost 0.43 points on average.
- The MRI results for the trial also showed that brain volume loss was less in the children on the trial compared with the natural disease progression.
- The drug, Cerliponase Alpha, is administered to children through a port directly in the brain in a 4 hour infusion every two weeks. The data released shows that the process was generally safe and well tolerated by the children.
- When the data was further examined for the 9 children who have entered the extension part of the trial it suggested that the benefits have continued with children showing an improvement of 0.2 points at 72 weeks compare to an expected loss of 3.13 points.
- Of those 9 children, 3 have gained a point, 5 have remained stable and 1 has lost a point.
- 4 children have now been receiving the drug for 88 weeks and the data has shown a loss of 0.5 points compared to expected loss of around 3.83 points.
- BioMarin plans to submit applications for the licensing of this drug to the regulatory authorities in Europe and the USA in the middle of 2016 and there is the hope that they will receive the official responses in the first half of 2017.

BioMarin's statement on compassionate use of Cerliponase Alpha is as follows:

"BioMarin is planning to implement an early access program to provide access to treatment for additional CLN2 patients prior to obtaining marketing approval. An early access program will be limited in scope and number of participants, and will be conducted under a protocol. We expect that the program will be conducted at centers that have participated in the cerliponase alfa study. Those sites have experience administering this drug directly to the brain and would ensure continued patient monitoring. The program is expected to begin in Q3 2016. The exact timing will vary by country of the sites participating. The overall scope, eligibility criteria and details of this program are still being determined. BioMarin must adhere to specific legal procedures for each country and has begun these preparations at risk with the goal of being ready to dose patients in Q3 2016. BioMarin will provide additional details about the scope and timing of this program as they become available."

The BDFA is actively working with the regulatory authorities within the UK and Europe to ensure that this good news is translated quickly into an effective treatment for those children who need it.

Alongside such positive news we are all remembering those families for whom these developments have come too late. We send our heartfelt thanks to them for the invaluable contribution that every one of their children has made to get us to where we are today.

The full BioMarin press release can be found:
http://investors.bmrn.com/releasedetail cfm?ReleaseID=958565

Sense is a national charity that supports and campaigns for children and adults who are deafblind. It provides tailored support, advice and information as well as specialist services to all deafblind people, their families, carers and the professionals who work with them. In addition, it supports people with a single-sensory impairment with additional needs. Working alongside the team at the BDFA, its Children's Specialist Services can offer support with particular issues, including:

- practical ideas on suitable play/leisure activities
- information on massage, story massage and personal stories
- parent/carer groups or family events – to meet other families, share information and get support
- support at times of crisis
- resource libraries which lend sensory toys and equipment (currently only available in some areas)
- communication and practical teaching strategies and intervention approaches
- support with individual Education Health Care Plans
- support for Annual Review and Child in Need meeting process
- mentoring for local MSI teachers
- information and advice service
- Sense holidays

Support offered will be time limited, depending on the level of family need. The aim is to start with what a child is able to do and build on that.

More information about Sense is available on their website **www.Sense.org.uk** or by contacting the Information and Advice Team on **0300 330 9256** or **020 7520 0972** (email: **info@sense.org.uk**) who will put you in touch with a member of your local area Children's Specialist Team.

Family Resource Corner
Sensory Shoebox Activity Kits

Jo Willer is a retired special educator who has a 9 year-old granddaughter, Kate, with Late Infantile Batten disease (CLN2). Here she shares her experience with sensory shoebox activity kits – a tool to help provide stimulation to non-verbal and non-ambulatory children with varying abilities. Kate is blind, unable to walk or talk and has minimal use of her arms and hands.

Jo began making sensory box activity kits when Kate's respite caregivers didn't know how to play with her or keep her busy. Clear plastic shoeboxes containing a variety of items for an individualized task are used with simple instructions and adapted for use with each child.

The caregiver is encouraged to describe the steps of the activity, what it looks like and what the child will feel by using lots of descriptive words to help the child understand and make it fun and interesting.

Jo takes a brush and exfoliating glove over Kate's arms, face, hands and sometimes legs and feet to stimulate and awaken her neurological system. She also uses a kit with a strong magnet and magnetic bingo chips which are separated by colour or counted while moving the magnet across them. She helps Kate grasp the wand and pick the chips up, alternating hands to work both sides of the body. The magnet makes a fun clicking sound as the chips are picked up and they listen closely and laugh when they get a 'bunch of 'em' picked up at once!

By involving the child as much as possible in holding items, turning switches, saying words, the activities can be done whatever the child's ability.

Further ideas for what to include in a sensory shoebox activity kit can be found on the following websites: **http://www.shoeboxtasks.com/ http://www.pinterest.com/karachambers/teacch-task-ideas/**

BDFA Education Training Days

The BDFA ran two education days in March in Manchester and Mansfield. A wide range of professionals from across the north of England attended, including class teachers, teaching assistants, a music therapist and a teacher of the visually impaired. The training was well received.

We were able to educate professionals about all types of Batten disease as well as providing information on the key educational issues. We talked about the importance of visual impairment support as well as Education, Health and Care Plans and what should be included in these.

"The day gave us lots of ideas to try in class"

Katie Hanson, the Batten Clinical Nurse Specialist, led a session on epilepsy and movement disorders in Batten disease. She made education professionals aware of the difference between epilepsy and dystonia and also gave more information on other types of movement disorders within the disease process.

We were joined by Barbara Moore from Sense who led a session on sensory stories and massage. We all had a chance to have a go at a massage story and saw how simple it is to make our own sensory stories. Barbara and two members of the Sense team were able to give us all more information about Sense and their range of UK-wide services, and professionals found it very useful to meet them.

"Fantastic information, broken down so we can understand"

Networking was also a big part of the days. All professionals found that being around others working with children and young people with Batten disease was helpful. They were able to learn from each other and to gain new ideas that they could use within their own settings.

The feedback from everyone was excellent and they valued the day and the information they received. We will be running another Education Day at Linden Lodge School on Friday 10th June 2016. Please let your children's school and other professionals know about this event and contact **support@bdfa-uk.org.uk** for further information.

We would like to thank Fountaindale School for hosting one of the training days at its site.

Box4Kids

Seriously ill and disabled children are now being nominated for the Barrie Wells Trust's Box4Kids Initiative 2016.

The Barrie Wells Trust is a children's charity founded and funded by entrepreneur and philanthropist Barrie Wells. Box4Kids invites seriously ill and disabled children to experience a once in a lifetime VIP event at a major UK sporting or entertainment venue.

To date over 2300 guests have received an amazing Box4Kids experience including shows at the O2, Royal Albert Hall, MEN arena, and sports at Premier League football clubs, Wimbledon, the Jockey Club, Wembley, and the Millennium Stadium.

40 hospitals across the UK nominate children for Box4Kids events including Great Ormond Street, Alder Hey, Sheffield Children's Hospital and Noah's Ark Children's Hospital, Wales. It is hoped many more hospitals will come on board this year.

2016 is set to be Box4Kids' biggest year to date, boasting even more events and new venues including its first ice hockey box. Last year's trend of following S Club 7's tour around the country looks to be repeated with boxes already donated for two X Factor Live Tour performances and three Little Mix 'Get Weird' tour dates.

The Barrie Wells Trust looks forward to welcoming children from the Batten Disease Family Association to their events this year.

More details can be found at **www.barriewellstrust.org**.

EURORDIS
Rare Diseases Europe

Dear patient representative,

If you would like to complete our survey "Acting on the Treatment Information you Have" please do so by the **end of May 2016**.

EURORDIS invites you to disseminate this survey on the use of any drug or device to the largest possible group of patients in your network.

Please find the survey here:
https://www.surveymonkey.com/r/YVY9PX8

This study received positive feedback from the INSERM Ethics Evaluation Committee/IRB00003888 on 9 June 2015.

EURORDIS thanks you in advance for your assistance.

Sincerely,
François Houÿez
Treatment Information and Access Director/
Health Policy Advisor

www.eurordis.org +331 56 53 52 10

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www.bpdesigns.co.uk

Hello, my name is Anna Pickering. I am 16 years old and my brother, Ben Pickering was a child with Batten disease. To be more precise, he was diagnosed with Late Infantile Batten disease aged five, meaning he couldn't walk, talk, eat nor see and this happened when I was only 3 years old and didn't have a clear idea what was going on. Now I am older, I understand more about what my brother had and I feel it's important to spread my knowledge and tell everyone what it was like having a brother with Batten disease.

When Ben was diagnosed, I was oblivious to the situation. I was just starting to walk and talk myself. So I had no knowledge of how to either grow up with a sibling with Batten disease or how to introduce myself into the world. I am a person who tries to view everything from several perspectives. This helped me as a child, as I never treated Ben differently to anyone else. Even though he was my big brother with Batten disease; to me, he was just my big brother, Ben.

By the time I was seven, it was clearer to see that something wasn't right with him, and that there was pressure on each of my parents to cope with both me and my brother. My parents never restricted their love or our family experiences – we would still go on exciting holidays around the world, exploring different places and even though Ben couldn't experience it the way I was experiencing it, I always knew he was happy to be with the people who loved him the most.

> "We would laugh and we would smile, and it is such a beautiful memory to have, and I will cherish it all my life."

I have many memories with my brother but one sticks with me every day. When Ben was about six, and I was four, we would sing 'Row, Row, Row Your Boat' while sitting and moving side to side in the fire gate in our lounge. We would laugh and we would smile, and it is such a beautiful memory to have, and I will cherish it all my life. My grandpa used to film Ben and me as we were growing up, and watching the videos reminds me of the love we had in our family. It really was infinite. And that's the beauty of it. Although we had a son/brother with Batten disease, it really did give our family a strong relationship. We called ourselves 'Four Sides of a Square'.

My friends at school, and out of school, were always so supportive of me and they never treated me any differently, which is what I wanted. I didn't want to be seen as the girl with a disabled brother, I wanted to be seen as a normal schoolgirl. There were ups and downs coping with school work and family life and I wanted to keep them balanced so I could keep growing through my life. Overall, school life wasn't a problem for me.

> "The charity would host events and conferences where I could talk to people about my feelings and thoughts about growing up with a sibling with Batten disease."

Although I loved my family so much, there were times when I felt I couldn't take it all. I felt Ben was the priority in my family and this made me sometimes feel unwanted. Obviously I knew that my parents loved me dearly, but sometimes I felt alone and with no one to talk to. Generally I keep my thoughts to myself and I don't share them because I don't want sympathy or to get other people down. I felt if I told my parents they would feel upset or disappointed in themselves, and I obviously didn't want that at all. I always found a way to get round it, and move it to one side so I could carry on growing. I couldn't talk to my friends because I felt like they wouldn't understand what I was going through, because my life was so different to theirs.

But this is where the BDFA helped. The charity would host events and conferences where I could talk to people about my feelings and thoughts about growing up with a sibling with Batten disease. But we didn't only talk at

these events; there would be games, shows, music, crafts, animals, everything... and I enjoyed every moment of it. Not only that, but I have made so many friends within the charity who I will cherish and love forever. We all help each other get through the difficult parts of growing up with our sibling, and we are always there for each other. Without these events, I feel like I would have just broken and not been able to cope with growing up overall, so I really do appreciate everything the charity does.

Unfortunately, my brother passed away, aged 12, in November 2009 when I was only ten. The morning before he passed, I remember hugging him before I went off to school and saying 'Goodbye'. At this point, I think my parents knew he wasn't going to make it through the day. And then, I clearly remember coming home and finding out that he had passed away and I ran into his room and I lay next to him crying because he really did change my perspective on life, and now he was gone. However, he never went away from me because he will always be with me, in my heart and soul, forever.

Six years on, my perspective on life is unique and people always ask how I have this outlook. I know it is from my brother. Ben gave me a varied view on life. He taught me that you should never restrict yourself or give up on a fight. I believe that if it wasn't for Ben, I wouldn't be the person I am today and I am so happy that I had such a beautiful childhood growing up with him. I will always be so proud to be called his little sister and I hope he is proud of me and what I am achieving.

Love you always, Ben.

The BDFA annual conference is an important part of the year for families with children affected by Batten disease. It is a brilliant chance to catch up with our Batten family, meeting friends old and new.

Saturday was a very busy day, both for parents and children. It was great for us to hear updates on the latest research and to gain new ideas for entertaining our children and making family life more manageable. Our children were having fun playing on the fire engine, stroking animals in the petting zoo and creating their own Boris teddy bear. Effie and George both enjoyed the music and dance class!

The meal on Saturday evening was an invaluable chance for us to socialise with other families, gaining support from other families on the same journey, as well as inspiration and strength from bereaved parents. The highlight of the evening was hearing the 'Hey Charlie' song for the first time. There wasn't a dry eye on our table!

On Sunday the children went to the Sealife Centre in Birmingham with their excellent carers and we had another opportunity to catch up on the latest research into Batten disease and chat to researchers about future plans. We also had time to go into the beautiful Remembrance room and reflect on all the stars, in memory of our beautiful angels.

The BDFA conference is invaluable to us. It gives us a very rare chance to be 'normal' and not feel lonely in a crowded room. When our child is screaming or in seizure, rather than the usual stares, we receive compassion, comments of 'my daughter does exactly the same' and endless support from new acquaintances who we know will become lifelong friends. Many thanks to the BDFA for making this weekend possible.

Carly Hadman

See page 24 for details of our 2016 family conference

VOLUNTEERS

We are so grateful to all our volunteers and fundraisers. Currently we have two volunteers who devote many hours of their time to helping us out in the office. They help us so much with jobs that we do not have the time or expertise to do ourselves and we want to say a huge thank you to both of them. If you feel you could offer time to help us out in any way (either in the office or remotely), please do get in touch.

EMILIA STEFANESCU

I joined the BDFA team in November 2015 as a Social Media volunteer. I spend around 3 hours each day monitoring and being active on the BDFA's social media channels, their blog and website. With a background in advertising, I love the digital environment and I really enjoy pushing my limits by volunteering for a rare disease charity. I have learned a lot about the BDFA's amazing work and it is my pleasure to help with raising awareness and with digital communications.

ROSS SQUAIR

I am Ross Squair and I started working at the BDFA in September 2015. I got the job via a man named Bob Sutton who works for a company called Employability and he thought that BDFA would be a good choice for me to work at. I started by helping prepare leaflets for their annual conference and sorting out paperwork, and after that I did some photocopying and a bit of shredding when they don't need certain papers any more. I was then introduced to DS which stands for Donor Strategy and I do some admin stuff by adding contacts who help and raise money for the charity and also there are people on there who are affected by Batten disease and I like to think that's my favourite part of working at BDFA.

HART Voluntary Action

Volunteer Recognition Event

On 22nd March 2016 we were delighted to be invited to the Hart District Council Chairman's Volunteer Recognition Event as our two office volunteers were nominated for awards for their volunteering with the BDFA. We are thrilled for Emilia and Ross that they have been recognised in this way and we all enjoyed drinks and canapes before the ceremony in which they were awarded certificates and a bottle of bubbly to celebrate their contribution to volunteering in the local area.

They know how much we appreciate them but it was lovely to see others acknowledging their contribution, especially as they were among the youngest volunteers to be nominated. From the BDFA's perspective it was also great to show our presence in our local community and there was much interest in Batten disease and our stand from those present.

"It is an honour to be nominated and to be presented with a certificate of appreciation for volunteer work. It makes the volunteer work even more enjoyable when I am able to help the BDFA Team and to be recognized for it. I would like to thank Andrea for believing in me and my work and for giving me the opportunity to work for the BDFA"

Emilia Stefanescu

Where I used to work either I or another chap used to lock up; whoever was the first to leave would tell the other so he could set the alarm. We would often set the world to rights and it was on one of these evenings that I sensed his 'happy go lucky' demeanour had disappeared. Even to me, it was obvious that something was troubling him. Several times he protested it was nothing, until he finally told me one of his children has Batten disease. At that moment, it meant nothing to me. I had never heard of it although now I've come to realise that this lack of awareness is a major problem that needs to be addressed – but more of this later.

Our subsequent fractured conversation did not really convey the enormity of it and I needed a computer to enlighten me; there is no need to tell you the answer it gave. Over the next few months, we gradually talked about his family and his feelings. His personality had changed dramatically; he was confused, scared, depressed and he was extremely angry with everything and everybody.

"Sitting at the piano I remembered things my friend had told me and I wove these things around a simple tune"

My partner Marion and I often discussed my friend's plight; we wanted to do something to help raise awareness and perhaps earn a few bob for the BDFA along the way. I have some musical ability, and as Marion's daughter, Lily, sings in a prestigious choir, writing a choral piece for them seemed like a plan and *Hey Charlie* was born. Sitting at the piano I remembered things my friend had told me and I wove these things around a simple tune. These are his words, a man in turmoil; I could not have expressed these feelings with such power.

Lily's choir performed *Hey Charlie* at their Christmas performance and put out some buckets and leaflets. We sent the money to the BDFA, told them what we had done and thought little more about it… When Andrea heard a rough recording she declared that it was not just the voice of one man but 'the voice of Batten' and asked if it could be professionally recorded. On the 29th July 2015 we went into Abbey Road studios with the choir and a host of musicians to record *Hey Charlie*. The day at Abbey Road was filmed and both the song and the film were launched at the BDFA's family conference.

Whilst all profits from sales of *Hey Charlie* will go to the BDFA, it would be foolish to think that these would be of any significance; very little is left by the time iTunes, Amazon et al have taken their cut. The best we can hope is that we get it heard by the right people (those with large amounts of surplus cash) and it will move them enough to offload some of it our way.

Celebrities and large corporations tend to support the bigger charities. The clue is in the word bigger – big names, big charities, big advertising budgets. Maybe it's time to collude – we must endeavour in some way to encourage them to put some of their 'peanuts' in our tin and not always into those tins that are already close to being full. We need a 'name!' Does anyone know Johnny Depp?

If you haven't already downloaded *Hey Charlie* or bought the CD – here's how. Go to **www.imarcomedia.co.uk** or **www.bdfa-uk.org.uk**. You can download from most streaming sites (iTunes or Amazon) and watch the video from Abbey Road on BDFA YouTube channel. In the summer, we will be launching a video about the story of *Hey Charlie* and why it is the Voice of Batten.

Ian Frazer Smith

Remembering...

The BDFA is here to support families at all stages of the Batten journey. We believe that bereaved families deserve the best possible care, information and emotional support to help them at any point that it may be wanted or needed. All of our bereaved families remain a part of the BDFA's network for as long or as little time as feels comfortable to them. We understand that some families may prefer to stop or to minimise the contact that we have with them, and will do so immediately upon request. However, we will always be there should they feel they wish to contact us in the future. If families would like to remain in contact with us then we can offer services to support the whole family.

- **Remembering:** We will always endeavour to support families' wishes to have their loved ones remembered in our biannual newsletter and feel that this process should not be restricted by any concept of time. Our memories are with us forever and therefore we will be receptive to anyone wishing to share their memories of someone. The BDFA will produce a personalised ceramic star for all bereaved families on request which will be hung on the remembrance trees at the family conference each year.
- **Emotional Support:** The BDFA helpline is available for all family members and friends to access emotional support or simply someone to listen. The BDFA can also put families in touch with other bereaved families for an opportunity to share experiences and speak to someone who understands.
- **Bereavement Services:** The BDFA can help families to access further support both on an emotional and practical level. By contacting the helpline, families can also obtain information about resources and support for bereaved siblings.
- **On-going Contact and Support:** The BDFA is here for as long as a family needs us and would like to remain in contact or involved. Some bereaved families stay in touch with us and continue to attend events such as workshops and conferences, both as a support to them and to other families.

We constantly monitor the support offered to bereaved families and consider ways to develop this service. If you have any suggestions or thoughts about bereavement services then please share them with us.

If you would like further information about bereavement support then please email **support@bdfa-uk.org.uk** or call **0800 046 9832.**

In February Harriet Lunnemann and I attended the British Paediatric Surveillance Unit (BPSU) 30th Anniversary rare disease conference in Birmingham. This year they celebrate 30 years of a remarkable institution, which provides a valuable infrastructure for the monitoring and collection of data on rare childhood conditions. The work it has pioneered has informed national screening policies and influenced clinical practice worldwide.

It was an opportunity to network and raise awareness of Batten disease and the work of the BDFA. Our stand attracted interest from paediatricians, students and those involved in the work of the BPSU. The talks were varied and interesting on the work of the BPSU, the value of data collection and Dan Levi from the Care and Action for Tay-Sachs Foundation gave a very moving patient perspective.

On 29th February – Rare Disease Day – I attended the Findacure Scientific Conference at the Royal Institution in London. Rick Thompson, Findacure's Scientific Officer, gave an overview of their work and drug-repurposing programme (www.findacure.org.uk). The morning session featured talks from researchers, pharmaceutical companies both large and small, rare disease patient organisations and experts in the field. These were very informative and presented a wide range of views and ideas on drug repurposing. All agreed that this is a vital area for rare disease research: taking existing generic drugs already licensed for human use, and looking for new uses for them to treat other diseases such as Batten disease.

Heather Band, BDFA Scientific Officer

In the afternoon delegates were invited to submit suggestions for the five minute "Lightning Talks". Our abstract was chosen and so on behalf of the BDFA, the Freeman family and Dr Claire Russell at the Royal Veterinary College, I presented a joint BDFA project, "Fishing for a Cure" – how zebra fish can provide an innovative way to screen selectively from existing drug libraries to look for new treatments for Batten disease. Currently Claire has identified a potential lead compound, which is showing a beneficial effect in her CLN2 model. Claire was at the presentation, which was well received, and we made many valuable contacts. Many thanks to Rick and the Findacure team for a great meeting.

© Rare Disease Day UK/Josh Tucker

Two days later I was off to London again for the rare disease event at the House of Commons, organised by Rare Disease UK.

Alistair Kent spoke about the progress that has been made and the crucial role that the patient voice plays in creating change and improving services. MPs from all parties were represented and in agreement that patients and families must be central to all policies. There was time to meet and talk with other groups, I made new contacts and met up with colleagues old and new: Pat Roberts from Save Babies UK, a fellow member of the LSD Collaborative and Flora Raffai, CEO of Findacure.

These were three great events, epitomising the BDFA's Mission "Together We Will Make A Difference" for those affected by Batten disease.

Heather Band

BPNA Meeting

From the 27 - 29th January 2016, Andrea West and Heather Band attended the British Paediatric Neurology Association annual conference held at Sheffield City Hall. The programme brought together clinicians, pharmaceutical companies and researchers from the UK and across the world. Connections were made with neurologists and paediatricians currently working with children and young people with an NCL diagnosis and we raised awareness of the disease and the challenges of diagnosis with those who are not. This group of professionals is very important to the care and management of children and young people with an NCL diagnosis so the BDFA has a key role in raising the profile of the disease and the experience of families at these meetings.

Whilst it is not always the case at these busy meetings, it was very exciting to see NCL research being presented. The programme included a presentation from Dr Ming Lim from the Evelina Children's Hospital on his work with Professor Jon Cooper on Safe Combination Therapies for Juvenile Batten disease. There were also two NCL poster presentations, one detailing the work of international Batten experts and their knowledge of the natural history of CLN2 disease, and one detailing the real-world experience of the diagnosis of children with CLN2 disease.

We know that many of these professionals will never work with a child or a young person with an NCL diagnosis in the whole of their working career but we want to make sure that if they do they are prepared and able to quickly provide families with the support and information they need and deserve.

PhD awarded to Sophia kleine Holthaus

The aim of the project supported by the BDFA and Beefy's Charity foundation was to investigate the feasibility of gene therapy to treat early visual failure in Batten disease by funding vital research with Dr Sara Mole, UCL, and Professor Robin Ali at the world-renowned Institute of Ophthalmology.

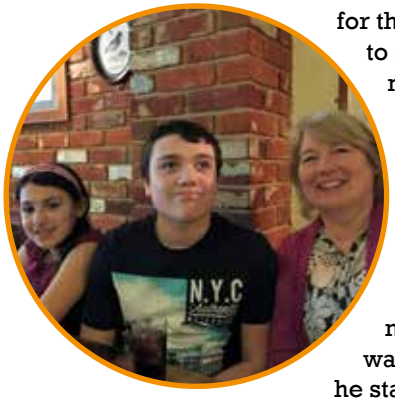
Sophia kleine Holthaus began her studies by developing a model to study how the disease affected sight, and then testing for effective treatments. Her work has been exceptional and has shown novel results in that different cells within the eye are affected more than was previously thought, and at different stages in the disease. Discovering this has allowed for the development of a targeted therapy.

Our congratulations go to Sophia who was awarded her PhD on 22nd January 2016. Also present were her supervisor Dr Sara Mole, Dr Sander Smith and all the members of Professor Robin Ali's group. We extend our thanks to all of them for their help and support of the project.

Sophia's work has continued beyond her PhD and she is currently testing new potential vectors – ways to deliver a "good" copy of the gene – to the place where it is needed in the eye in her model system. Her current work is vital in bringing viable therapies to treat vision loss a step closer.

We all have different ways of coping with what life throws at us. Writing poetry has always been mine. My inspiration, as always, is my grandson Jordan.

Jordan was born a happy, healthy baby in September 1999. I remember feeling overwhelmed with love for this little bundle as I held him in my arms for the first time. He went on to meet all his childhood milestones. Always a cheerful little chatterbox who loved Thomas the Tank Engine and travelling on trains.



Then, just before he started primary school, my daughter noticed he was bumping into things and he started sitting right in front of the television. She took him for an eye test and he was prescribed glasses which didn't help at all. Eventually he was referred to Great Ormond Street Hospital, where he was diagnosed with Retinitus Pigmentosa and we were told he would gradually lose his sight.

Two years later Jordan had his first seizure. His eyesight was rapidly declining, his seizures were increasing and behavioural problems began.

Fortunately my daughter was told about the Wolfson Behavioral Clinic at GOSH. Jordan was then referred to Guy's & St Thomas' Hospital, where he was officially diagnosed with Juvenile Batten disease at the age of twelve. We were heartbroken.

Now aged 16, Jordan's speech and mobility are slowly declining but he is still my loveable rogue.

Debbie Norris

Our Path through Batten disease (The middle years)

The road is very rocky,
our pathway very steep,
the ups and downs of this disease
will often make us weep.

But we keep on going,
there is no time to rest.
Awareness and a cure
has now become our quest.

We have had to be accepting
that the hopes and dreams we had
will all be very different now,
but we try not to be sad.

We'll just celebrate life differently,
adapt our dreams and hopes.
Living each day one by one –
this is how we'll cope.

It's now about the little things –
comfort, love and peace.
Doing what we think is best
to combat this disease.

It's not an easy journey
with all its ups and downs.
Batten disease is always there
trying to break our hearts or break us down.

Sometimes we think it's just too hard
to survive another day,
but then a ray of sunshine breaks through
to help us on our way.

A smile, a twinkle in his eyes,
a reminder why we are here –
the unconditional love for him
which we will always hold so dear.

Debbie Norris

BATCure Kick Off Meeting

The first meeting of the BATCure group took place at UCL in London from 27th - 29th January 2016. Over 30 scientists, research students and clinicians attended from Denmark, Germany, Italy, Latvia, and Spain, Sweden and the UK. Expectations were high and it was great to finally be starting on the project.

The BDFA is an integral part of the project to provide the patient voice and Heather Band, BDFA Scientific Officer talked about the BDFA's work package. Several groups within the consortium are new to the field of Batten research and were keen to hear about family experiences and how important research is to our community.

Developing new therapies for Batten disease

BATCure Consortium

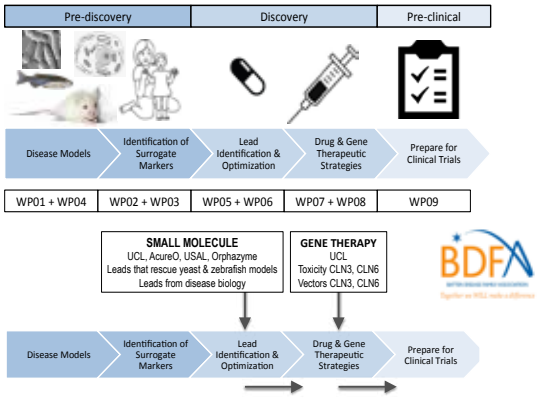
Coordinator - Sara E Mole

MRC Laboratory for Molecular Cell Biology, UCL Institute of Child Health and Department of Genetics, Evolution & Environment, University College London, London WC1E 6BT, UK. s.mole@ucl.ac.uk

PARTNERS BATcure Kick-off meeting UCL, Jan 2016

PHC-14-2015
666918
36 months
1/1/2016-31/12/2018
€5 995 769

CONCEPT CLN3, CLN6 and CLN7



This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 666918.

12th Annual World Symposium 2016 and visit to BioMarin

I was privileged to attend the World Symposium for Research into Lysosomal Diseases in San Diego. Whilst this is a very important research conference in the world of lysosomal diseases, the lack of any research relating to Batten disease on the programme meant that the BDFA had not attended in the past. It was testament to the hard work of all of the researchers, and our supporters who help us fund much needed research, that this year's programme saw presentations on CLN2 (Late Infantile Batten disease) and CLN3 (Juvenile Batten disease). This is a trend that we want to see continuing and growing as we drive towards more clinical trials for all forms of the NCLs. Thanks to the support of our BDSRA friends and colleagues, we were able to have a joint conference stand. It was rewarding to have a large number of researchers and clinicians visit us and show interest in our work and the experience of families living with an NCL diagnosis.

Dr Tammy Kielian presented her exciting research on gene therapy for CLN3 disease and we look forward to hearing more about how this is translated towards clinical trials by Abeona Therapeutics. Abeona Therapeutics will be attending the BDFA family conference in November 2016 to talk about how this work fits into their development pipeline.

Dr Angela Schulz presented the data for the first time from the phase 1/2 BioMarin enzyme replacement therapy trial for CLN2 disease. You are can read more on this positive news and what it means to the CLN2 community on page 4.

When the conference finished I travelled with Dr Margie Frazier from the BDSRA to meet with a group of Californian families. I was warmly welcomed and it was clear that they experience many of the same challenges as families living in the UK.

I finished my visit with three days at BioMarin's headquarters near San Francisco. I presented the work of the BDFA and UK family experiences to over 100 employees before touring the labs where samples from the trial are analysed, and the manufacturing plant where the enzyme is produced. It was clear that everyone working on this project is hugely committed, from the administration staff to those working overtime in the factory. We are very grateful to the whole team and hope that our combined effort will mean that this treatment is available to as many children as need it, as quickly as possible.

Andrea West

NCL 2016 BOSTON

The 15th International Conference on Neuronal Ceroid Lipofuscinosis (Batten disease)
Boston, USA

Date: 5 - 8 October 2016

Venue: Wyndham Boston Beacon Hill

website: www.nclboston2016.com

Emyr Lloyd-Evans, PhD, School of Biosciences, Cardiff University

When did your interest in the lysosomal storage diseases start?

About 15 years ago when I was doing my Masters degree on Gaucher disease in Professor Tony Futerman's lab at the Weizmann Institute in Israel. Professor Fran Platt from Oxford made a visit while I was studying there and I went on to complete my PhD in her lab in 2002. During my time there I was able to work on projects that eventually helped to develop a successful therapy for Niemann-Pick type C (miglustat). It was a great project to be involved in and fuelled my interest in developing therapies for lysosomal diseases.

Tell us about your current research.

In Fran's lab we were looking at the role of calcium in cells. This can act as a form of communication between cells. We were the first group in the world to show that this was different in the lysosome of a human disease. This work led to my Fellowship at Cardiff in 2010, an opportunity to set up my own group and continue the work.

Through contact with Batten research charities I became interested in NCLs, seeing a potential for overlap. Was calcium signalling not working in these diseases as well?

How has funding from the BDFA helped?

Greatly. The funds from the BDFA, on behalf of Battle Batten, allowed us to apply for matched funding from Cardiff University for a PhD student. Without support from charities many rare disease projects never get off the ground.

What do you see as the greatest challenges in the NCL research field?

Not knowing enough about the function of disease-causing genes. If we understand what is going wrong, then we can better target early events in disease and find better therapies.

From the laboratory to the clinic – how can this be improved?

The impact of our research is constantly assessed and this is a very good thing as it keeps our focus on what matters to patients and families. Pharma's attitude is changing – they have to give something back to the community and therefore have rare disease teams. The changes in European law allowing for easier orphan drug designation and have made it easier to bridge the gap. Once one drug is successful – like Miglustat for Niemann-Pick Type C – many more follow. It's the domino effect.

What do you like doing outside work?

We have a dog and I like to go for long walks near where I live in Cardiff. It helps clear the mind and keeps me fit. I am a keen photographer and love taking photos, but maybe this also has connections with what I do at work!

Plans for the future?

Being part of the EU Horizon 2020 funded consortium, co-ordinated by Sara Mole, working on the BATCure project for CLN3, 6 and 7 over the next 3 years. We are keen to bring our expertise into research for all NCLs. CLN8 is a good candidate and because of its biology it is of great interest to us, and a collaboration with some of my former colleagues may be the next step.

UCL

NCL Resource - A gateway for Batten disease - www.ucl.ac.uk/ncl

This website (maintained by Dr Sara Mole, University College London) serves as a global gateway for all those who have an interest in or are affected by Batten disease and for anyone who wishes to find out more.

NCL International Registry

DEM-CHILD

In November 2015, Andrea West and Heather Band travelled to Germany to meet with our global clinical and research partners working on the DEM-CHILD Batten disease International Registry. The meeting was organised by Dr Angela Schulz, MD at the Children's Medical Centre in Hamburg-Eppendorf. Productive discussions of how this international collaboration can move forward took place with groups from Europe and worldwide. The aim is to ensure that data and information about all forms of Batten disease can be captured and analysed.

Why are registries important?

Having accurate and up to date information is vital for successful clinical trials but there are other benefits that can often be overlooked. Understanding and building up a picture of what is happening in the disease assists proactive interventions for patients, enables delivery of better supportive care and improves quality of life. Studying current data identifies indicators of early disease, helping to improve early diagnosis (see page 22).

How is the BDFA involved?

The BDFA brings the patient voice to the discussion and the challenges of patient involvement, including the issues that were highlighted in the registry session at the BDFA 2015 family conference. We are proud to be funding partners in this work along with the BDSRA, Hope for Bridget and Noah's Hope. Thanks to your fundraising efforts we can fund a researcher, based with Dr Schultz in Hamburg. This will facilitate faster interpretation of the data collected, making a significant difference to our current working knowledge of Batten disease and providing vital information for potential future clinical trial development.

How can you help?

The letter opposite details how UK patients can participate. To take part in the registry, or receive more information on what is involved, please contact Dr Ruth Williams, the lead clinician for the UK.

Evelina London Children's Hospital

Guy's and St Thomas' NHS
NHS Foundation Trust

Dear Parents/Guardians,

We are writing to you to let you know about an International Registry and Database for children diagnosed with Neuronal Ceroid Lipofuscinoses (NCL), also known as Batten disease.

NCL are a rare group of progressive diseases that mainly affect the brain and cause symptoms such as epilepsy, movement disorders, dementia and blindness. In general, symptoms worsen with time but the age at which symptoms start and the speed at which the disease progresses is very variable. The diagnosis is usually made by examination of the patient, MRI brain scan and blood tests. The diagnosis is then confirmed by genetic analysis.

At the moment we cannot predict how the disease will progress in any one individual. It may depend on many different factors, including the person's genetic makeup, their environment and lifestyle.

An International Batten Disease Registry has already been established and we hope that the information we collect from UK families will contribute to this international project and increase our understanding of how the disease progresses and why the progression is so variable between different people. Currently we do not have good treatments for these diseases, and we also hope that the information we collect from children and families worldwide will in time help us develop and test treatments for these diseases.

We would like to include as many people with NCL in the database as possible. The more information we are able to collect from different people with the disease, the greater our understanding of the disease will become.

We are looking to collect information from your child's medical records as well as the results of any tests that your child may have had, and we may also ask your permission for researchers to use existing samples of biological material (i.e. blood, skin cells, other cells) that may have been taken during the period of confirming the diagnosis. We will not ask your child to undergo any extra tests or procedures for the purpose of this study.

If you would be willing to help us with this study, please contact me on **0207 188 3998** or **ruth.williams@gstt.nhs.uk** for further information.

Yours sincerely,

Dr Ruth E Williams,
Consultant Paediatric Neurologist,
GMC 3057036

Looking for new "Biomarkers" in Batten disease

A group of researchers led by Dr Kevin Mills at the UCL Institute of Child Health, are undertaking a project to detect blood and urine biomarkers for Batten disease. Biological markers or 'biomarkers' are measurable substances in human tissues and body fluids. There are many different types of substances that can be measured, including proteins, lipids or other metabolites. In neuro-metabolic disorders (such as Batten disease) the concentration of many of these may increase or decrease. In Dr Mills' research a technique called mass spectrometry is used to find the most important molecules to study and the best way to measure the differences.

The aim of this project is to use this information to develop a new rapid test for diagnosis. If successful, this technique could also be used for monitoring the effectiveness of new treatments.

In this project Dr Mills' group will address the following questions:

- Can we use a biomarker to design a new, easy, cheap and quick test?
- Are there any markers we can identify/measure very early in the disease?
- Can we detect differences between affected people and those who are carriers of mutation or "mistake" in the same Batten disease gene?
- What is the relationship between the mistake in the gene and the problems this causes in cells?
- What kind of disease mechanisms are involved in Batten disease?
- Does a newly discovered biomarker open up the possibility of a new treatment option?

Initial steps have already been made after successfully recruiting affected families into this study following the BDFA 2015 family conference. The promising preliminary experiments discovered several potential biomarkers in the urine that will be further investigated. Based on these initial findings the group are seeking further funding to expand the study.

The BDFA is a co-applicant for the project (represented by the Scientific Officer, Heather Band) as a key element for the success of the project depends on the recruitment of affected families. More information on how to take part in the study will be available on the BDFA website shortly.

The Research Group comprises Dr Kevin Mills, Dr Barbara Csányi, Dr Sara Mole, Professor Paul Gissen, Dr Apostolos Papandreou, Dr Simon Heales, Dr Wendy Heywood from UCL & Great Ormond Street Hospital, with Dr Ruth Williams at the Evelina Children's Hospital.

Dr Barbara Csányi is a young researcher with a special interest in neuro-metabolic diseases and with a strong clinical background. She graduated in Hungary and worked in Spain for many years as a paediatric neurologist. She is currently working as sub-investigator with Professor Gissen on the first human enzyme replacement therapy trial on Late Infantile Batten disease at Great Ormond Street Hospital. Barbara will play a leading role in the project in the area of patient recruitment, sample collection and analysis.

Dr Kevin Mills' main research interests are the study of the disease mechanisms, pathogenesis and treatment of rare genetic metabolic and lysosomal diseases in children. He is leader of the Proteomic, Metabolomic and Lipidomic Group which compromises 23 post-doctoral Fellows, research assistants and PhD students. Their focus is patient-driven, translational research and aims to establish rapid, sensitive methods to study, diagnose and monitor the treatment of patients from Great Ormond Street Hospital and the National Hospital for Neurology and Neuroscience. They specialise in biomarker discovery and converting those with clinical relevance into rapid, multiplexed, diagnostic medical tests. An example of this research is the development of a simple, rapid and more specific test for pre-symptomatic kidney disease in another lysosomal disease (Fabry disease).

Can gene therapy treat CLN5 Batten disease?

In a project funded by the BDFA and managed by Sparks, Professor Jonathan Cooper aims to determine whether gene therapy could be used to treat a type of Batten disease called CLN5 disease.

Current situation

The Neuronal Ceroid Lipofuscinoses (NCL) commonly known as Batten disease, is a group of rare, fatal neuro-degenerative conditions, affecting cells in the brain gradually, causing a loss of physical and mental abilities. Depending on the type of Batten disease, diagnosis usually happens in childhood, between five and ten years old, but can be much earlier. It is an inherited condition, passed on through one of 14 faulty genes. Children are born apparently healthy before developing seizures, loss of vision and loss of the ability to walk, talk and eat. Children and young adults will eventually become completely dependent on their families and carers, before dying prematurely.

One potential way of treating Batten disease is gene therapy. Gene therapy works by introducing a healthy copy of a gene to cells so they can then make a functioning copy of the protein that is missing. A harmless virus is often used to carry the copy of the gene into the cell. For one form of Batten disease – CLN2 disease – gene therapy is currently being tested in a clinical trial. It is thought that gene therapy may also be beneficial for other forms of the disease.

How this project will help

Professor Jonathan Cooper is going to investigate whether gene therapy could be used to treat a different form of Batten disease – CLN5 disease – building on a successful and long-standing collaboration with Professor David Palmer and his group at Lincoln University, New Zealand. They have been working on using gene therapy in an animal model of CLN5 Batten disease, and this new research project will seek to determine if the treatment can prevent the death of brain cells, and if there are any unintended consequences that might be harmful in the long term.

Ana Assis, Research Assistant

BDFA
BATTEN DISEASE FAMILY ASSOCIATION
Together we WILL make a difference

BDFA Family Conference

18th - 20th November 2016

Holiday Inn, Stratford Upon Avon

Meet with clinicians, education and social care professionals and other families

A full programme of activities for all children with qualified care staff

Registration now open

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