Batten Disease Newsletter

ISSUE 31 AUTUMN/WINTER 2015

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

BDFA
BATTEN DISEASE FAMILY ASSOCIATION

Batten Disease Family Association Registered Charity No. 1084908

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Dear Member

From our Chief Executive


Andrea

Dear BDFA members, friends and colleagues,

When I sit down to write my piece for the BDFA newsletter it always gives me the opportunity to take some time and look at what has been achieved over the past few months. A wise friend always says that sometimes we are so focussed on getting to where we want to be that we forget how far we have come. The end of our journey is a much longed-for world without Batten disease but we sometimes forget that we get there by all the single steps taken by our fundraisers, members, supporters and families who live with this disease and its legacy every day. This newsletter gives us the opportunity to recognise all of those efforts and each step that brings us closer to our goal.

Over the past few months we have had the opportunity to work closely with our friends and colleagues from across Europe and the rest of the globe. We are all so much stronger and louder if we fight this fight together so we were delighted to host the meeting of the Batten Disease International Alliance and the next planning meeting of the European Education Project in London in June. More about these meetings can be found on pages 6 and 19. Two members of our team, Barbara Cole (BDFA Education Advisor) and Heather Band (BDFA Scientific Officer) also attended the BDSRA conference this summer where they were able to share our UK work and learn from our friends and colleagues in the US.

I am delighted to introduce a regular feature in our newsletter (and soon to be on our website) by Katie Hanson, our Batten disease Clinical Nurse Specialist. We introduced Katie to you in our last newsletter and she will use her slot as an opportunity to answer questions that she is regularly asked by families during the course of her work. She can of course also be contacted at Great Ormond Street Children's Hospital on 020 7405 9200 ext 0460 or by email (katie.hanson@gosh.nhs.uk).

A busy summer saw us launch the first UK Batten Disease Awareness Day on 5th June and hold our 10th Annual Walk in the Cotswolds. We were overwhelmed by the support shown to "Turn the World Orange" and our Awareness Day finished with the Spinnaker Tower in Portsmouth being lit up in orange as our Beacon for Batten. Friday 3rd June 2016 will be the date of next year's Awareness Day and we will keep you posted as we get nearer that date. More details and photos can be found on pages 10 and 11.

I hope you enjoy reading our latest newsletter and we would love to hear feedback on what you would like to see in the next edition. Please contact us by emailing **admin@bdfa-uk.org.uk**. As always, we cannot do any of this without you, so together we will always keep our eye on a world without Batten disease knowing that each of our steps is bringing us closer.

Best wishes,

Andrea

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

From our Chair


Mike

Dear all,

In our Spring newsletter, Andrea and I wrote about the new members of our staff team. I am very pleased to report that the staff have settled in well and are greatly enhancing our work. As a Board we consistently challenge ourselves on every penny spent by the BDFA so it has been very pleasing to see the benefits that investment in our infrastructure and staff team has made.

As I write this report it is sad to see a major UK charity, Kids Company, being forced to close. It is not for me to speculate on the reasons for this closure, but one thing we are led to believe is that the Charity was not holding enough cash in reserves with the result that they had to close immediately impacting greatly on their staff and the children who desperately needed their services. In the time I have been involved with the BDFA I have always been impressed by the way in which it has been run. Under the leadership of Julie Pickering, Pauline Docherty and now myself we have always been prudent in our financial management and never begun a project without the funding in place. As a Board we have always discussed the critical need for reserves. Recently we dropped this from 12 months to 9 months due to our significant financial growth but this is something we will continue to review to ensure the sustainability of our work.

Yet again I'm pleased to report that one of our large member events was a great success – the 10th Annual BDFA Walk and camping weekend. I know it was greatly enjoyed by all and that for some of our members it was a great opportunity to catch up with others who are sharing the same difficult journey or to remember those who are sadly no longer with us. Our whole staff team did a fantastic job pulling together the wonderful weekend. Unfortunately what started out 10 years ago as a fundraising event for the BDFA has now become a rather large expense so next year we plan to do something different which we hope will achieve the same for our families but will also return the weekend to being a fundraising event. I know the fundraising team are already busy planning so watch this space for more news.

Finally I am going to finish with more fundraising news as we have had some Herculean efforts in the past few months. Duncan Brownnutt and Rod Wark cycled 500 miles on a tandem to the Arctic Circle. Not only was it a heroic physical challenge but it was also a very emotional one and we congratulate them on their enormous efforts.

I do hope you have had an enjoyable summer and I look forward to seeing many of you at the BDFA Family Conference in October.

Best wishes,

Mike

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

Front cover image: 10th Anniversary Walk Balloon Release. Photo by Theo Roose

The Year Ahead

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 its vital work by becoming Trustees and helping achieve its vision of a world without Batten disease.

The Board of Trustees is responsible for governing the BDFA and providing strategic leadership to plan its future direction and services. Trustees are elected by the Charity membership at the AGM.

We need candidates with demonstrable understanding, expertise and/or experience of the governance of a charitable organisation. In particular, this year we would welcome candidates with any of the following experience/skills:

- Knowledge of the challenges faced by families affected by Batten disease
- Experience of fundraising
- Experience of 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, usually in London)
- Attendance at the annual AGM and Family Conference (usually October)
- 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)
- Trustees receive no remuneration for their services
- 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, 01252 416110, andreawest@bdfa-uk.org.uk**

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

Contents

Diary Dates 2015/16

19 November 2015	Kidz & Adultz up North, Manchester
27 - 29 January 2016	British Paediatric Neurology Association Annual Conference, Sheffield
10 March 2016	Kidz & Adultz in the Middle, Coventry
3 June 2016	Batten Disease Awareness Day
5 - 9 October 2016	NCL 2016, Boston USA

CLN2 Burden of Disease Project

We are currently working with BioMarin to deliver the CLN2 Burden of Disease Project. This study aims to find out more about families who have been affected by CLN2 in order to influence government policymakers to deliver quality services and treatments to families living with this disease. On 24th April 2015 a group of families who have children living with the disease as well as bereaved families came together in Sheffield to share their experiences. It was lovely to see members of extended family there as we know the vitally important role they play in helping families manage the devastation of this disease.

Some families brought their children with them as childcare was provided. The children enjoyed playing with a parachute, making crafts and drawing.

The day was facilitated by researchers who interviewed families and led the discussions. The families were very open and honest about the effect that the disease has had on their lives and some of the discussions were very emotive.

The morning discussions were about the effects the disease has on the caregivers as well as discussion about the disease symptoms and the families' journey to diagnosis. In the afternoon some families were interviewed and others looked at the questionnaire which will be sent out to other families. They were asked to comment on the questions and make changes if necessary. Having the families' perspective on the questionnaire was very helpful to the researchers as they could then plan for the remainder of the study.

We would like to thank all the families that attended the focus group. Research projects like this one are invaluable to us in helping to gain important insights and to help us lobby government for services and potential treatments.

The questionnaire and interview process will be open to all CLN2 (Late Infantile Batten disease) families in due course and we will be in contact with you in Autumn 2015.

Harriet Lunnemann
Support & Advocacy Partner

Disability Living Allowance

Recently, with the support of Greg Mulholland MP, we were able to meet with Justin Tomlinson who is the Minister for Disabled People and his team at the Department of Work and Pensions. During the meeting we raised the issue of Disability Living Allowance for families living with a diagnosis of Batten disease. Based on your feedback, we do not believe that the current application process works for affected families.

We were able to show Justin Tomlinson specific ways in which the process did not work for those families who have children and young people diagnosed with Batten disease. The process is lengthy and it is difficult and families are having to reapply for their Disability Living Allowance every few years. Justin Tomlinson has now said:

- Children and young people who are receiving the higher rates of both the care and mobility components should never have to re-apply for Disability Living Allowance.
- If your child or young person's needs change you can update your claim by making a call to the DWP and you do not have to fill in another application form.
- When young people are transferred over to the new Personal Independence Payment at 16 years old, they should not be invited to a face to face interview but their parents can fill out a paper form on their behalf instead.

After the meeting we asked each of you via an online questionnaire to give us valuable evidence from your family's own experience. For example, despite assurances from the ministers, all of the families who filled in the questionnaire have had to re-apply for Disability Living Allowance when their child or young person was receiving the higher rates of the care and mobility components.

We are now awaiting a response from the Department of Work and Pensions and Justin Tomlinson in reply to the evidence gathered from the questionnaires.

If you have had or are having any problems with the DWP with either Disability Living Allowance or Personal Independence Payments please contact Harriet Lunnemann, BDFA Support and Advocacy Team (**support@bdfa-uk.org.uk or 07876 712553**).

From the Clinical Nurse Specialist


Katie

Well, I have been in my CNS job for nearly six months now, and what a privilege it is to work with such wonderful people. There has been lots going on in the background as I have settled into my job, and I have had to expand my knowledge in the Batten disease world and have met some wonderful families and children in the process.

I hope more families are feeling that they can contact me now and know how to get hold of me – if any of you feel you need any help with anything health related, or are even unsure about how to get an answer to an unrelated health problem, please do not hesitate to contact me.

The role of a Clinical Nurse Specialist is to educate, advise and work very closely with other nursing and medical teams for the best interest of a child and his or her family. My role is to act as an advocate for the child and family and to be a main link to other healthcare professionals across the country.

As any family who has a child with Batten disease knows, the amount of people involved in your child's care is phenomenal. There are physiotherapists, occupational therapists, speech and language therapists, neurologists, paediatricians, carers, teachers, school nurses – the list seems endless. But as a qualified paediatric nurse of nearly eight years, I hope I can tie all these things together for you and make life a little bit easier.

I have enjoyed attending BDFA events and watching the amazing work the charity does. My highlight was the BDFA 10th Annual Walk in June. The thing that struck me most was how all

these people are so different and some only knew about Batten disease through the friend of a friend, but they all came together for the same reason and had the same willingness in their heart to spread the word about Batten disease.

I hope this article will be a regular occurrence in each newsletter, and I aim to provide an update of what I have been doing as well as including a health related topic each month. This month I am talking about fussy eating in childhood.

Best wishes

Katie

Katie Hanson
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020 7405 9200 ext 0460

Fussy Eating – also known as Selective Eating – in Childhood

I've got to admit, all I hear from my parents are nightmare stories about me and my fussy eating as a child. I went through a phase of eating ONLY boiled carrots. I couldn't get enough of them, to the point they were picked out of every meal just so I could eat them. Then, the following week I was apparently spitting them out and trying to hide them.

Some may not agree with this but, believe it or not, fussy eating is not just a trait of early stage Batten disease but is normal in many children. However, in a child with Batten disease, it may well be more of a challenge to get your child to eat the foods you want them to eat as well giving them enough fluids because they have their own likes and dislikes.

As we know, certain types of Batten disease have traits that are linked to autism. The web link below is an informative booklet written by a dietician and can help a wide range of those who are dealing with fussy eating, and not just autism or Batten disease.

What I'm sure we all know does not work is coercion and force feeding. This is not recommended for a variety of reasons, and will normally have the opposite effect to the desired one. Other hints and tips can also be found in the booklet.

The main thing is that your child receives the correct nutrition and hydration, and if you have any concerns about your child's nutritional health your GP will be able to refer you to a dietician or nutrition consultant for more help.

Please see the following link to access the booklet:
http://www.zoeconnor.co.uk/2012/10/extreme-faddy-eating-selective-eating-advice-sheets/

Katie will also have a page on the BDFA website **www.bdfa-uk.org.uk** which will have a Frequently Asked Questions section and useful resources for both families and the professionals working with them.

Fire Up a Cure: BDSRA Conference, Chicago 9th-12th July 2015


Barbara Cole

It was my great privilege to be invited to attend the BDSRA Annual Conference in July in Chicago. Families began to arrive and I saw many tearful but happy reunions. Many extended families came and it was evident that the conference was hugely important to them all and the 'high spot' of the year. Distances are huge in America and many families had driven for hours or even days with all the equipment and medication needed for their child. Siblings met up with other siblings and were soon off exploring the resort together. This opportunity for them to meet up, have outings and fun together and to be able to share their experiences was obviously important to them.

Following Registration the detail on badges made it easier for families to 'seek out' other families affected by the same CLN type. A golf tournament, a trip to a baseball game and a full day BDSRA Board meeting all took place on the first day. I attended the new family 'mixer' where newly diagnosed families were introduced to their 'buddy family' and could ask questions and familiarise themselves with the conference programme. Some had very recently had the diagnosis of Batten disease, others had taken some time after diagnosis to be 'ready' to attend and all were warmly and lovingly welcomed.

Jarrett Payton, a motivational speaker, provided an entertaining and thought-provoking first session. Some sessions were attended by everyone, including updates on research and clinical trials and there was a programme of smaller group sessions for newly diagnosed families, bereaved families, mothers, fathers and grandparents and for each CLN group.

Together with two experienced educators, Jo Willer and Leanne Lepato, I had been invited to join the Education Panel for a session entitled "Classroom Quality of Life: Strategies for Parents and Teachers". It was wonderful for Jo, Leanne and me to share ideas and expertise and we plan to continue this collaboration. The session was well attended and there were lots of questions from parents and lively discussions. The issues that challenge children and families affected by Batten disease are similar on both sides of the Atlantic. We discussed how to support children and young people in the later stages of the condition, home schooling, transport issues, getting individual support and how to make sure that achievements – however small – are recognised and celebrated. The importance of music and music therapy was highlighted as well as the need to support young adults no longer in education.

The team from the University of Rochester Batten Center attended the conference as did other scientists and researchers. Pets as Therapy dogs and miniature ponies were greatly enjoyed by the children and young people. The Batten Kids Care team did a wonderful job of keeping everyone actively involved and everyone dressed up in their best outfits for the conference Banquet on Saturday evening. The band got everyone on the dancefloor and encouraged children and young people to 'take the microphone'. It was wonderful to see and hear affected children and young people and their siblings on stage singing their favourite songs. Sunday morning's programme included a Service of Remembrance and a very helpful 'decompression' session for new families. Families then began their long journeys home and I had to pack my bags and leave too.

It was a wonderful experience and I am very grateful to the BDSRA and the BDFA for their joint funding of my trip. Next year's BDSRA conference is in St Louis. I do hope my new friends in the US will say 'Meet me in St Louis' in 2016!

Barbara Cole
BDFA Education Advisor

European Union Education Project Update

The European Union funded Education project is now in its second year and has brought together many experts working with children and young people with CLN3 (Juvenile) Batten disease from Norway, Germany, Finland, Denmark, Scotland, England and the United States. Schools, Patient Associations and specialist Batten disease services are working together and already much has been achieved.

The second project meeting was hosted by the BDFA in London in June. The project group worked hard developing a survey to be distributed to families and professionals in all the participating countries this Autumn. It will be followed up with in-depth interviews with a representative group of families and professionals. The aim of the survey and interviews is to find out as much as possible about the educational experiences of children and young people with CLN3 and to build on that information throughout the project. We hope that as many families and professionals as possible will participate in this important first phase of the project.

After the meeting the project group were invited to visit Dr Sara Mole at UCL to hear the latest update on CLN3 research. This included a talk by Sophia Kleine Holthaus on progress in gene therapy to treat visual failure in Batten disease, a very relevant research project funded by the BDFA. The day was very informative and was much appreciated by all who attended.

In September I will be attending a workshop in Norway where we will be working on the second phase of the project – the development of educational tools to improve the education opportunities of children with Batten disease and to support life-long learning. The group, including teachers who work day to day with our children and young people with CLN3 in schools, will be sharing their expertise along with professionals with special interest in the field.

Barbara Cole
BDFA Education Advisor
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New College Worcester

New College Worcester is a national residential school and college for young people aged 11 to 19 who are blind or partially sighted. Many of our students also have additional needs, and the College currently supports two students with Batten disease.

Situated in Worcester, just south of Birmingham, most students at the College are either weekly or full boarders although there are also a few who travel to school every day. Funding for places is through the student's own Local Authority, so students at New College Worcester come from far and wide across the UK and have a range of different skills and attributes, and support and specialist teaching needs.

Staff at New College Worcester are experienced in supporting young people with Batten disease, who can receive extra support in the classroom and the residential setting, plus intensive support for braille and literacy if required. The curriculum can be adapted in line with these young people's individual needs, and they are encouraged to participate in sports and activities with their peers.

Every student at NCW has a tailored programme, carefully planned and delivered to meet their individual needs in three key areas; academic achievement, independent living skills and in extracurricular activities at the College and in the community. The College philosophy is based around outcomes, wellbeing and learning. Whilst offering the National Curriculum for academic study, the College has an extended curriculum focusing on independent living and mobility, which is extended into the residential provision of the College. Out of school activities and sports build on self-esteem as well as being fun with friends and play a crucial role in the development of confidence and preparation for life beyond the College.

While it can be difficult for parents of children with Batten disease to consider a residential setting for their child, many of the benefits of the College are because it is residential, facilitating a 'waking day' which is beyond the standard school day. Not least is that young people are able to be young people, spending time with friends and where nobody is excluded from a social life outside the school day. Many students have experienced social isolation prior to joining NCW, which is upsetting for both parents and young people. At NCW no student is excluded, no matter what their situation or condition.

The College has three junior houses for students in Years 7 to 11, each with several bedrooms. Each house has dedicated residential staff led by a Senior House Parent. The houses have study rooms, a sitting room and communal kitchen area and are often alive with parties, theme nights and birthday celebrations. Constant links with home are encouraged and students can visit home or have parents stay for the weekend whenever they wish.

There is also an extended 'home weekend' every half term for students in Years 7 to 9, so that they can enjoy a full weekend at home with their family.

Sixth Form life is relaxed and fun. As students reach Sixth Form they are supported by care tutors in the Sixth Form hostel and house which are organised to ease the transition to university, Further Education and independent living. There is a common room with TV/DVD player, stereo, pool table and games as well as shared living areas. They start to do more for themselves, including budgeting, shopping, cooking, washing... even the dreaded ironing! Experienced, knowledgeable and qualified staff are on hand to provide support, advice and encouragement while students develop these skills. Many students go on to Further Education and have successful academic careers. Many find employment and live independently.

The College also offers a range of events over the course of the year for parents, teachers and those supporting a young person with Batten disease. Most of the events are free of charge, and others have a nominal charge to cover costs. Parent Information Days are available where parents can get a feel for the College by taking a tour and having a chat with a member of staff without any commitment. For those who wish to spend a longer period at the College, Family Days are a fun way to get involved, with a range of activities available around the building and grounds for the whole family.

For those supporting young people in mainstream school, Outreach Open Days offer the chance to spend time with the NCW teachers and learn some useful teaching techniques in particular subject areas. In addition to these there are also courses for young people to help with revision for GCSEs or in planning for the transition to university.

Information on all the courses and events can be found online at **www.ncw.co.uk/events**. Information about New College Worcester can be found online at **www.ncw.co.uk** or by telephoning **01905 763933**.

An Introduction to Abeona Therapeutics

Greetings Families! I am writing to introduce Abeona Therapeutics and share more about myself and the role I'm serving in. Abeona Therapeutics is an organisation focused on developing gene and plasma therapies for rare diseases. Our lead programs are an AAV9 gene therapy for Sanfilippo Syndrome (mucopolysaccharidosis (MPS) type IIIA and IIIB). Similar to NCLs, these are inherited lysosomal storage disorders where an enzyme is lacking or not functioning properly. The company was created through a close collaboration with a dozen Sanfilippo foundations to progress the work of Dr Douglas McCarty and Dr Haiyan Fu from Nationwide Children's Hospital in Columbus, Ohio.

Since the acquisition by PlasmaTech Biopharmaceuticals in May of this year, the company has returned to the Abeona Therapeutics name and added two more programs. Our scAAV9 CLN3 gene therapy was licensed from the University of Nebraska Medical Center for Dr Tammy Kielian's research. We are working on pre-clinical studies, planning our meetings with the FDA, and the timeline for clinical trial. The animal data from Dr Kielian (dedicated aunt to her niece diagnosed with CLN3) is exciting and we are pleased to be taking it forward.

The most recent gene therapy added is to treat Fanconi Anaemia; an inherited blood disorder that often results in bone marrow failure and cancer. This has been licensed from the University of Minnesota based on Dr Jakub Tolar's work. The AAV delivered CRISPR/Cas9 allows for very specific modification or repair of the DNA. This is also in the pre-clinical stages.

My role with Abeona is that of Vice President, Patient Advocacy. It was recognized very early by Abeona leadership that our relationships with patients, their families, and the foundations should be one of the highest priorities. Additionally, there is a commitment to providing education and information on gene therapy, the clinical trial process, and sharing progress for our proposed therapies. Prior to joining Abeona, I served as Vice President, Client Relations for Aldevron, a contract manufacturer for gene therapy bioreagents, genetic vaccines, and molecular diagnostic controls. My educational background is in Biotechnology, Chemistry and Microbiology. I live in Minneapolis, MN with my husband and daughters (12 and 9 years old).

I'm looking forward to participating in the BDFA Family Conference in October. Until then, you are welcome to write or call to introduce yourself, share about your child(ren), and ask me questions. My purpose is to serve the Batten community as a resource for information, provide support to families, and help our company bring forth a therapy for CLN3.

Abeona THERAPEUTICS
Working together to find a cure.

Michelle Berg,
Vice President, Patient Advocacy

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Batten Disease Awareness Day 2015

The first ever UK Batten Disease Awareness Day was held on Friday 5th June 2015. Up and down the country, people were turning the world orange with cake sales, dress down days, running races and generally having fun whilst letting everyone know more about Batten disease. From Rock FM to Great Ormond Street Hospital, from Sara Mole's laboratory in UCL to Colchester Zoo, from Glenwood School to Harwich Primary, you all did your bit to get the word out there.

One highlight came down on the South coast. The team at the iconic Spinnaker Tower in Portsmouth agreed to turn the tower orange for the evening, and people for miles around could see our orange beacon. Boris the Bear turned up to support, but could not be persuaded to abseil down this time... He had a busy day too. He met up with Wilbur from Aerobility, a charity giving people with disabilities the chance to fly light aircraft. In his special orange flying suit, Boris even had the chance to get behind the controls himself.

As with all things BDFA, our social media pages were flooded with posts, shares and tweets, giving all our followers a flavour of what everyone was getting up to. There was the opportunity to download a banner and logo to turn your own pages orange. Our "Did You Know…" slot on Facebook was particularly popular with many reposts, retweets and new followers. We shared facts with you about Batten disease every hour on the hour and were even retweeted by Stephen Fry, Allan Carr and Ian Botham. In all, we reached over 50,000 people – what an achievement.

To raise funds as well as awareness, a text donation service was available. It has already totalled £700 but is still open for donations. Just text **BDFA15 £2** to **70070** to donate £2. It may seem a small amount but £10 pays for the BDFA Freephone helpline for one month.

10th Anniversary Annual Walk

After much planning by the fundraising team, BDFA "Team Orange" finally hit the road from Farnborough to the Cotswolds for our 10th anniversary BDFA walk and camping weekend on a sunny Friday afternoon in June.

We arrived with our cars, a van full of provisions, some dogs, some children and some partners and husbands to find the caravanning element of the party already set up with camping tables with tablecloths and flowers and early evening drinks on the go. The marquee and the bunting went up and gradually tents appeared and that evening everyone got to know one another over fish and chips on the hay bales.

Well before 7am the following morning the smell of bacon was enticing people out of their tents ready for a day of walking. The hall at the start point was soon a sea of orange T-shirts as the young, the young at heart, seasoned walkers, dogs, teenagers and much smaller children all geared up for a walk.

After a group photo we walked through Bourton-on-the-Water and headed off into the fields. We went up hill and down, through long grass and short, over many a stile (and some barbed wire!) and through plenty of kissing gates. Some were fitter and faster than others but, as a member of the rear party, there was great determination by all to get back for the lunch stop. Never has a large piece of delicious bread and a choice of fillings been more welcome.

After lunch the more determined headed off to add to the seven miles they had already walked. They were rained on and came close to some cows and swans but heroically returned back at camp in the early evening with smiles on their faces. Others found gentler amusements in the afternoon including ice-creams, tearooms and pubs in Bourton as well as the Motor Museum to see Brum.

Back at camp the evening's entertainment was beginning. Other non-campers joined us for an orange balloon release, each with a label in a loved-one's honour or memory. It was very moving to watch the orange balloons float gracefully away into a beautiful blue sky. We then enjoyed more expertly barbequed food and a special 10th Anniversary cake before settling down to an evening of music and entertainment for all ages from the band "Grace Hates Monkeys" who so kindly volunteered their time and amazing musical talents. There was something for everyone as we sat on the hay bales in the marquee and rested our weary feet as we chilled and darkness fell. "Let it Be" from Frozen was aimed at the young but it was remarkable to see the enthusiasm of some of the older people at this point…..Even the campsite owner found an excuse to come over and join us.

Tired out from walking and fresh air, people gradually headed into their tents for the night. Some tents were much smaller than expected and there were a good few issues with lilos going down – those in caravans may have been smug at this point but those under canvas just appreciated their bacon sandwich and cup of tea as they emerged the following morning all the more.

The BDFA staff team is grateful to everyone who helped and participated in the weekend and in particular their hardworking families who gave so much to the weekend, as well as to Marion Giles-Donovan, Ian Frazer Smith and their amazing family of musicians. After ten years of walks we will now be planning other get-togethers which may be easier for our less mobile families to join. We hope all those who came along enjoyed their weekend.

One of our younger participants had this to say:

"I liked the band because I liked the song "Why the sun shines". I liked the camp site because it was nice playing there. The walk was the best because it was seven miles long and because you got to wear orange T-shirts."

See overleaf for photos of the walk and Awareness Day


BDFA
ELLIE'S ARMY
John & Bridie
BATTEN'S


BATTEN'S
BDFA

Family Story

On 12th January 2011, we were told that at 37 weeks my waters had broken and to prepare to be induced that afternoon. In the early hours of the next day our perfect tiny 6lbs 5oz baby boy was born. We named him Ollie. Although I had complications after the birth, Ollie was checked over and was said to be a healthy baby.

During the first two years of Ollie's life he spent a lot of time in and out of hospital with breathing problems before he was eventually diagnosed with asthma. Even so, he hit all his milestones and was walking before the age of a year to keep up with his older brothers.

After Ollie's second birthday we noticed a speech delay and he became increasingly clumsy. Each time we mentioned this to health care professionals, we were told not to worry and that he would catch up.

Whilst trying to work out the best plan for Ollie, I went into labour 14 weeks early with our baby girl. After countless drugs and going into anaphylactic shock, labour was stopped and our princess hung in there until 38 weeks. Amelia was born on 4th March 2013 after a very difficult birth where we nearly lost her more than once. Weighing in at 6lbs 13oz she was the final piece to our perfect family.

Following breathing problems, Amelia was diagnosed with laryngomalacia. At 14 weeks old she contracted bronchiolitis and she was admitted to hospital where we nearly lost her again. This princess is a true fighter.

For the next eighteen months life was good. In fact looking back now it was perfect. We had four very happy and healthy children. Ollie had been attending speech and language therapy but no one was concerned and we had been assured that he would catch up by the time he reached school age.

On 9th September 2014 Ollie had his first seizure. This day was and still is the most frightening day of my life. Ollie had a normal day at preschool but had come home with a raised temperature. A trip to our GP confirmed a urine tract infection. Later that evening while bathing, Ollie had his first seizure. While pulling him out of the water I realised he was not breathing. Ollie's dad performed mouth to mouth resuscitation and brought our little boy back. Ollie was rushed to our local hospital by ambulance where we were told he had had a febrile convulsion. He recovered and was allowed home the next day. Feeling relieved and thankful and believing the nightmare was over, little did we know this was just the beginning.

From then on Ollie began to have weekly seizures but with no signs of raised temperature or infection. In October an E.E.G. was carried out confirming generalised epilepsy and Ollie was prescribed anti convulsive medication. His doctor decided to do an MR scan which showed a small abnormality in the cerebellum and we were reassured that this was nothing to worry about. Ollie was referred to a neurologist. At the appointment with this neurologist, instinct told us something was not right. The questions asked and the examinations carried out made us worry. We walked away from the tests with no answers but more worry while waiting for blood test results to come through.

Throughout all this Ollie was his normal, happy and lovable self; he continued to attend preschool and was thriving. As parents we wanted to know what was going on, so we made the decision to pay for a copy of Ollie's medical records and reports. Reading

through the records we came across a list of tests requested by the neurologist. This was the first time we had come across the name Batten disease. Like other people we googled the name and what we found was alarming. We could not believe there was anything so cruel, but at the same time we felt relieved that Ollie didn't seem to fit the signs and symptoms described in the article. A couple of weeks later we received a phone call asking Mike, Ollie and me to meet with the neurologist in 2 days' time. My gut instinct told me something was wrong but my brain said the doctor would not be asking us to bring Ollie with us if it was bad news.

That day will stay with me forever. I can remember the room, the colour of the walls, even the smell. As we sat in that small room with Ollie on his daddy's knee we were told our child has Batten disease, that there is no cure and that life expectancy is between six and twelve years. Ollie is just 4 years old. Our world fell apart in that moment. We were given a leaflet and something about having to each give a blood sample there and then to check for the mutant gene. We were then left alone with no reassurance or further hospital appointment and made our way home. Trying to process what we had been told as a family, we decided to get away for a few days. A short break spending time together as a family turned into a nightmare as Ollie had 14 seizures in the space of a few hours. It was then we decided to stand up for Ollie and get him the best care possible.

We began to research and found a specialist in London as well as the amazing support system at the BDFA. Exactly four weeks later we travelled two and a half hours by train to London to meet the specialist. After a long discussion we decided to get our youngest child, two year old Amelia, tested for Battens after learning she had a 25% chance of also having Batten disease.

A few weeks later we received a call from the genetics clinic asking us to bring our two older boys aged six and seven years to be tested. Mike's dad decided to come with us saying he had a bad feeling and it turned out he had good reason. On arrival we were asked to leave the boys in the waiting room as the doctors wanted a word. For the second time our world fell apart as we learned our beautiful princess also had Batten disease. Walking out to my boys in the waiting room, trying to hold myself together and to act normal must be the hardest thing I have ever had to do. Mike could not look at them and had to leave. It was down to me to keep it together for them. The boys were not tested that day; there was no sane reason for them being there.

Ollie and Amelia and every other child with Batten disease deserve a chance of living life to the full. We continued our research and found help overseas from other families and medical professionals. We know Batten disease is rare as everyone we speak to has never heard of it. We decided to set up a Facebook page called Ollie's Army Battling Against Battens Disease to raise awareness of Battens in the hope that this will mean that one day there will be a cure.

Ollie is starting to struggle with his balance and we have been to so many appointments and seen so many different professionals that I have had to make a list of who is who. Amelia has recently been assessed and we are proud that she is meeting all her milestones. The most important thing now is to make every moment count. We don't bother with our old worries and will continue to fight for our children and raise awareness of Batten disease. We are still waiting on the results of our two older boys and we pray they get the all clear. Every day we spend as a family of six is a gift and we will never take this for granted.

We are extremely lucky to have the amazing support of our family, friends and even some well-known celebrities. We are currently planning a Family Fun Day to raise funds for equipment for Ollie and Amelia as well as raising funds for the BDFA. Most of all the day will be about creating fun memories for all families who attend and support the event.

Lucy, Mike, Danny, Micky, Ollie and Amelia Carroll

Owen 'Bryn' Hitchcock

12 March 2007 - 3 May 2015

How do you sum up in a few words the life of our eight year old Brynnie who passed away so suddenly? Bryn would say "Me won!!" the way he always did racing his brothers to the car.

"Me Won" was Bryn living life to the max – he'd never walk when he could run; he'd never say 'hello' to you when he could knock you backwards with a big hug; he'd never 'smile for the camera' when he could do a massive, screwed-up-face cheesy grin.

And Bryn has won.

He won by passing away while sleeping in his mother's arms, escaping the suffering and pain of the normal conclusion to his disease. He won by having two amazing brothers who loved him so deeply they'd squabble over who could sleep with him.

Our lives have been blessed – surrounded by the loving support of family, friends, and of those warm, enthusiastic people who are so dedicated to making a difference to families like ours by generously giving their time, equipment and emotional support to guide us along our journey.

On getting Bryn's diagnosis we needed to learn to walk in the rain and boy, did it rain. We quickly adjusted, learning to enjoy our four year 'rainy walk'. But Bryn wanted more from us – why be satisfied with 'enjoying' it when we could be 'loving' it? Why just 'walk' in the rain when we could 'sing' in it, and 'dance' in it? So we did.

Those who know our story know that these four years have never been a burden or a limitation, they have always been a pleasure, a gift, a joy. Bryn, our 'eagle' of a boy never once complained as his body betrayed him, and he stopped being able to do the things he loved – the running, playing football, laughing, smiling, chatting, seeing, eating. What an inspiration!

We wanted to keep Bryn safe in our nest forever, but of course Bryn, after so much time sitting and lying down would have been desperate to fly. When the moment to take that first flight comes, even eagles need a push. We didn't know it was time for Bryn to fly, but God knew. He knew it was time for Him to give Bryn a push, for Bryn to fly.

There is a verse in the Bible that seems like it was written for Bryn … and for us: Isaiah 40:31 says: *'but those who hope in the Lord will renew their strength. They will soar on wings like eagles, they will run and not grow weary, they will walk and not be faint.'*

I know Bryn would want to be celebrated, not mourned. To be remembered with love and fondness. He'd want us to look forward, to live with great energy and enthusiasm. He would want us to see the joy that is in all of our lives and take satisfaction, pleasure and reassurance from that. He would want us to race to the car. He would want us to be able to say 'Me Won!'

As Bryn's parents, in knowing him and sharing his life with him, we've already won. Winnie the Pooh put it very well: "How lucky I am to have something that makes saying goodbye so hard."

Bryn has definitely won. He's flying – and he's loving it. So now we need to keep living our lives with the same joy, energy and enthusiasm that Bryn lived his with. We don't want Bryn to rest in peace – that's not him. We want you to fly Bryn-Bryn, fly!!

Dylan James Robinson-Sinclair

22 May 2007 – 21 March 2015

He was my North, my South, my East and West,
My working week and my Sunday rest,
My noon, my midnight, my talk, my song;
I thought that love would last for ever: I was wrong.

The stars are not wanted now: put out every one;
Pack up the moon and dismantle the sun;
Pour away the ocean and sweep up the wood.
For nothing now can ever come to any good.

Remembered by and missed dearly by Mummy and little sister, Lyla

Ellie Mae Brownnutt

12 December 2008 - 7 May 2015

Ellie Mae lived only six years but she lived life to the full. She was happy and loved every day. She was a cheeky, determined, joyful girl who made everyone smile. She fought to be independent but accepted without frustration when she needed help. She wore her heart on her sleeve, loved her friends and family openly and would light up as soon as they came near.

Her favourite things were her family and friends, closely followed and sometimes exceeded by Peppa Pig and a host of Disney characters, most importantly Winnie the Pooh and Jess from Toy Story. She loved a cuddle and to sing songs. "Row Your Boat" was a favourite that even when she could no longer speak, she could still ask for and enjoy it as much as when she was a toddler. Being silly with her Grandads was one of her favourite pastimes, or stories and songs with her Grandmas.

Whenever you were feeling down, five minutes with Ellie Mae would cheer you up. She loved bouncy castles and fairground rides, balloons and cake, donkey riding and swimming. From babyhood, she enjoyed books and stories and her favourite books were loved until they disintegrated. She would spend hours playing with her small toys, putting hats and glasses on and off them.

She hated chickens and loved pigs and trained Caleb to chase the former away for her, which made a visit to None-Go-By Farm a rollercoaster of emotions. She was always busy with something.

Ellie Mae and her little brother Caleb shared a special bond, often shown by an affectionate cuddle followed by a quick whack across the face, all in love. From her first day at preschool and then at school she was always involved and determined to have fun, but always overjoyed to see Mummy or Daddy at the end of the day.

She leaves a space that cannot be filled.

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 has a special remembrance tree on which each child or young person has a ceramic star. If you would like a star for your child or young person please email **support@bdfa-uk.org.uk**.

- **Emotional Support:** The BDFA helpline is available for all family members and friends to access for 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.**

Support Resources

Dreams Come True is a national UK charity that works hard to make dreams come true for children and young people aged between 2 and 21 years old with serious and life-limiting conditions.

When a child or young person is faced with a serious or life-threatening condition, their dreams can sometimes be overlooked. That's where we come in.

We enable these courageous youngsters to fulfil a personal dream – often at a time when they need it most. A dream can be anything such as meeting an idol, feeding the animals at the zoo or even having a family holiday.

For some, a dream might simply be to enjoy the everyday activities that most of us take for granted. That's why we are also happy to contribute towards specialist equipment such as wheelchair swings, computers and purpose-built trikes for disabled children.

Anyone can nominate a child or young person for a dream. Please visit our website for more details and to download an application form.
www.dreamscometrue.uk.com/nominate

Aerobility is a registered charity founded in 1993 offering disabled people, without exception, the opportunity to fly an aeroplane.

For some, just that amazing first flight is enough to change their outlook on disability forever: ***"If I can fly a plane, what else can I do?"***

Others, realising that the fantasy of flight is truly in their grasp, decide to continue their training – many all the way to securing a Private Pilot's Licence and some even with an outlook to commercial flying.

Aerobility provides "experience of a lifetime" trial flying lessons for as many terminally ill and disabled people as possible every year as well as subsidising flying days for other disability charities and giving at-cost instruction and qualification flight training to disabled people.

As a User Led Organisation ("ULO"), Aerobility is run largely by disabled aviators for disabled people. Through various fundraising initiatives and careful management strategies we allow disabled people to fly for a fraction of the commercial rate. Aerobility also provides advice, knowledge and advocacy for disabled people who wish to fly, and is the representative body for disabled aviation within the UK.

For further information, please visit our website:
www.aerobility.com

The Butterfly Wishes Network

The Butterfly Wishes Network was founded in April 2013. We are now in our second year and have already provided photoshoots to children with Batten disease. Our aim is very simple. We are a group of professional photographers who have come together to give something back by providing families of children that are life-threatened with a photoshoot and at least ten digital images for free.

We want families to be able to capture some treasured memories of their family. We realise that families with children suffering from life-limiting diseases may have parents who cannot work as they are caring for their child and therefore cannot afford expensive photoshoots. Or they may just find the task of finding a photographer with enough patience too immense.

Each and every photographer in our network gives the time and talent of their own business free of charge. Even if you choose not to use our service, giving us a Facebook share will help us grow.

We are a non-profit network and we do not have charitable status as we have managed to run on virtually no money at all. Our photographers are all fully insured. Currently our organisation has 280 photographers across the UK, with a few sprinkled in Ireland. But we are growing all the time.

Please visit our website **www.butterflywishesnetwork.co.uk** or like our Facebook page "The Butterfly Wishes Network" for further information.

BATCure

In 2011 the EU Heads of State and Government called on the European Commission to bring together all of the previous EU research and innovation funding under a single common strategic framework. The Commission then launched a wide-ranging consultation involving all key stakeholders which led to the Horizon 2020 programme, now the biggest EU Research and Innovation programme with nearly €80 billion of funding available over 7 years (2014 to 2020).

In response to the call for proposals in the rare disease category, Dr Sara Mole brought together experts from across the EU including Denmark, Germany, Italy, Latvia, Spain, Sweden and the UK. The UK is represented by Dr Claire Russell (Royal Veterinary College), Professor Jon Cooper (King's College London), Dr Emyr Lloyd–Evans (Cardiff University), Dr Tristan McKay (St Georges but soon to take up a new appointment as a Professor at Manchester Metropolitan University), Professor Rob Harvey and Dr Ahad Rahim (UCL) as well as the BDFA.

Large EU grants such as these are divided into Work Packages (WP) and Heather Band, our Scientific Officer, was invited to lead on a WP. This part of the project would involve being the "Patient Voice" to support the scientific studies. Where direct patient opinion and involvement is needed, the BDFA will be fundamental in this process for affected families, individuals and the general public. This work is vital to ensure that we do all that is necessary to move any potential therapeutic advances forward for the benefit of those affected by Batten disease.

It was an important part of the bid process to demonstrate an effective, inclusive, workable method for involving patients across the EU, and enabling them to have equal and effective participation in potential research. This is ground-breaking work, which builds on the BDFA's experience of funding research projects and developing long-standing relationships with NCL researchers and patient groups across the world.

UCL provided the venue and we all met together for two days to discuss preparation of the final bid. It represented some of the best new innovative ideas for moving towards potential therapies in CLN3, CLN6 and CLN7 and was the culmination of an enormous amount of work by Sara Mole at UCL and all the participants.

Over 150 bids were considered for review in the 2nd stage, with the possibility of up to 10 being awarded, and in late August we were delighted to receive the news that BATCure had been funded.

It is testament to the commitment and dedication of everyone involved in this field that we now have a critical European investment of €6,000,000 into NCL research, bringing together key institutions across Europe to drive forward towards a cure.

Cell based systems for drug discovery in JNCL

In the Spring edition (Issue 30 p21) we reported on the progress of this project and the proposed extension as King's College had agreed to provide additional funds to supplement the current research grants awarded in 2013-14 by the BDFA, BDSRA and NCL-Stiftung, Germany. This was in response to a request by the BDFA to allow Dr Greg Anderson further time on the projects, as there had been delays in providing the necessary resources by the University.

Dr Anderson was due to return to King's to start work in the Summer but for personal reasons he has now moved to another position within King's College. The BDFA would like to thank him for all his hard work and wish him well in his new job. We are working closely with Professor Jon Cooper to secure a replacement to complete the development of this potentially valuable research resource by the end of June 2016.

It has been an eventful year as in June Professor Cooper and his team moved to a new purpose-built laboratory next door. You are all welcome to visit next year to see their new home and further details of their 2016 Lab Day and how to book a place will be available in March 2016.

Announcing the 15th International Conference on Neuronal Ceroid Lipofuscinoses (NCL; Batten disease):

Sue Cotman PhD, who is heading up the planning of NCL2016, is an Assistant Professor of Neurology at Harvard Medical School and Assistant in Neuroscience at the Massachusetts General Hospital (MGH). Dr Cotman graduated from the Ohio State University in 1993 and received her PhD from the Biochemistry Program at the same institution in 1999. She began studying NCL (Batten disease) during her postdoctoral training in Dr Marcy MacDonald's laboratory at MGH (2000-2005).

Dr Cotman currently leads an NCL research laboratory located within the Center for Human Genetic Research at MGH in Boston. She and her team are collaborating partners within the MGH-CHGR Joint Program in the NCL Disorders, a clinical and translational research program with a collective mission to integrate research and clinical care for patients of all ages with NCL.

Dr Cotman's research laboratory specialises in developing genetic and cell biological tools to look at the earliest events that result from the NCL genetic defects, which will lead to insights into the functions of the NCL proteins. These tools include mouse models, mouse-derived neuronal cell models, and human-patient induced pluripotent stem cells (iPS cells).

The models are key components for research aimed at identifying disease modifiers, for example, drugs that could then be used for developing NCL therapies. The models are being actively used for drug screening and pre-clinical drug development in a mouse model.

Dr Cotman collaborates with many NCL researchers worldwide including with Dr Sara Mole at UCL. Her laboratory is involved in research to identify new NCL genes which will improve our broader understanding of the NCL disease pathways and NCL diagnosis.

Dr Cotman serves in a scientific advisory capacity for the Batten Disease Support and Research Association in the United States (SAB member since 2011) and regularly writes for their website and presents scientific sessions at their conference as well as for other NCL and lysosomal disease Foundations.

She has always placed a high priority on making sure that patients and their families are informed and given a voice so the BDFA is delighted that she has offered to host NCL 2016.

As travel to NCL conferences can be challenging for families and often very expensive, we are working with Sue and Margie Frazier at the BDSRA on the best way to ensure that families can access the research findings from NCL2016.

Heather Band
BDFA Scientific Officer

An NCL conference is held every two years in a different host country. As many of you will remember **NCL2012** was held at Royal Holloway College, London University and organized by Professor Jon Cooper, Dr Sara Mole and Dr Ruth Williams. Co-hosting this event was a milestone for the BDFA and we have gone on to take an ever-increasing role in facilitating and funding research (see BATCure on page 17).

NCL2012 was a great success with excellent scientific presentations running concurrently with a programme for families, and coming together on the final day for a joint session. The Batten Disease International Alliance (BDIA) was officially launched and we have forged even closer links with our partners, most notably the BDSRA as we both support families and fund research for all forms of NCL.

NCL2014 saw the BDFA sponsor the 14th International Conference in Córdoba, Argentina which was organized by Inés Noher de Halac, PhD. Representatives from all the major Laboratories, Institutes and Clinical Centres gave presentations and it provided an opportunity for the South American families attending to meet – many for the first time – and to share experiences. Andrea West (BDFA) and Margie Frazier (BDSRA) played a leading role in the family day activities.

From 5th to 9th October 2016, Dr Sue Cotman and her team will welcome basic and clinical scientists working on the NCL disorders who will once again come together to discuss state of the art research and progress towards treatments and improved clinical care. **NCL2016** will be held in Boston, Massachusetts, USA, at the Wyndham Boston Beacon Hill, which is located near Massachusetts General Hospital and downtown Boston. More details will be coming soon and will be available on the BDFA website.

Meeting of the Batten Disease International Alliance (BDIA) in London, Heathrow on 16th June 2015

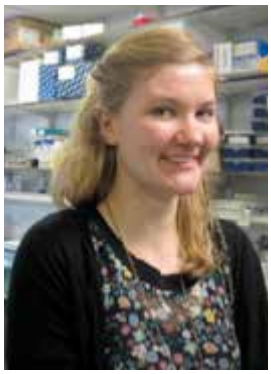
Following on from a very successful International NCL Education meeting at the beginning of the week, the BDFA were delighted to further host our international friends and colleagues for a meeting of the BDIA in London in June.

The meeting was attended by representatives from patient organisations and research foundations from Belgium, Denmark, Norway, USA, Serbia, Germany and the UK. As rare disease groups, we all understand the need to work together to share resources, good practice and expertise to maximise the impact of our work in supporting affected families and driving the research agenda towards potential therapies and ultimately a cure.

As organisations we have differing resources and structures, but we have much to share. To begin that sharing and connecting, a new BDIA website detailing the international organisations and their areas of work will be launched very soon. Currently further details of the organisation can be found on Twitter @BattenDisease and on Facebook.

"Gene therapy to treat the visual failure in Batten disease"

BDFA and Beefy's Charity Foundation fund a £60,000 research award for 2016-7



The BDFA is delighted to announce a further one-year extension to this project in collaboration with a very generous donation from Beefy's Foundation. The BDFA initially funded Sophia Kleine-Holthaus's (photo left) studies for three years in 2012, and then for a further year until December 2015. Her work on this project, in Professor Robin Ali's group at the Institute of Ophthalmology (IOP), has been outstanding in what is a very challenging field.

The BDFA would like to thank her co-supervisor Dr Sara Mole at UCL, Dr Sander Smith and all the team at the IOP for their dedication and expertise in support of Sophia over the past four years. Treating visual failure has relevance for all types of Batten disease and the contribution that the group has made cannot be underestimated.

BDFA research funding aims to support research of the highest quality; often this is the only source of funding to start a project, such as this one in a rare disease. The ultimate aim is that BDFA research awards will enable researchers to apply for larger grants to secure the expansion of the work in the future.

Development of photoreceptor treatment for visual failure

Using a CLN6 mouse model, Sophia's work demonstrated that photoreceptors are affected early in the disease, and that these cells are dying. Photoreceptors represent the majority of cells in the back of the eye that are light sensitive and essential for vision. Other cell types in the retina do not appear to die early in the disease.

However, when she investigated the effect of injecting virus at a range of concentrations, she found that even when CLN6 was delivered and expressed widely the loss of photoreceptor function was not slowed down. This was surprising. It meant that to prevent visual deterioration in these mice it is not sufficient to target only photoreceptor cells.

Looking for a new target

Sophia then investigated which other cell types in the healthy retina express CLN6. Her results showed that whilst it is present in photoreceptors it is also found in the inner retina and more specifically in bipolar cells, a cell type that is involved in transmitting light responses generated in photoreceptors.

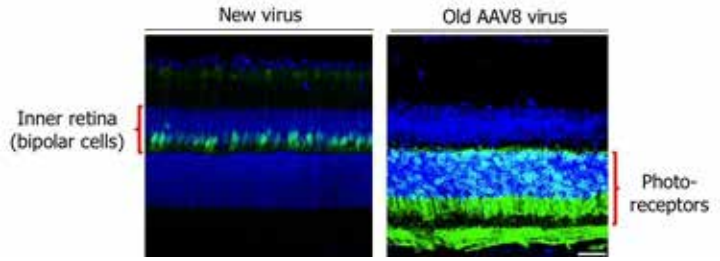
New virus to target inner retina

To target bipolar cells is very challenging because the cells are located within the middle of the retina and so are more difficult to reach by a virus. However a laboratory in America has recently developed a virus that can infect these bipolar cells.

It was therefore of vital importance that this work should continue for the long-term benefit of all those affected by Batten disease.

Although a CLN6 model is currently being investigated, the results obtained will make a significant contribution to future studies and the ultimate aim is to treat visual failure in all forms of NCL.

The BDFA wishes to thank everyone who has raised funds for the Charity so that we can now ensure Sophia can develop the new vector and test it in targeting bipolar cells. Future plans include treating both types of cells as well as testing the effects on retinal function.



Sparks Update...

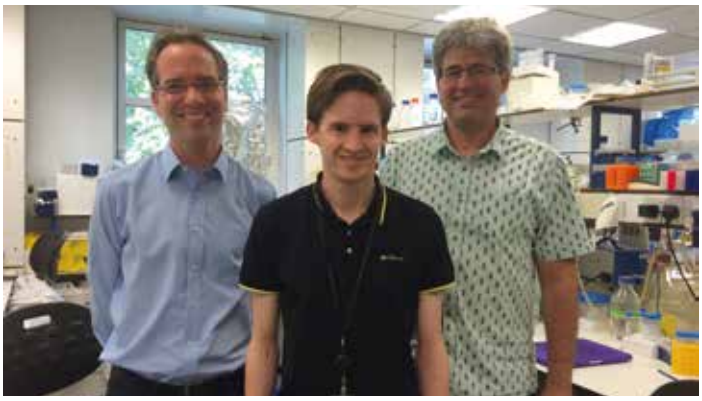
Applications for a one-year research project in CLN1, CLN5, CLN6, CLN7 and CLN8 (see BDFA Newsletter Issue 30 Spring/ Summer 2015 p22) have now been received and the Peer review process is in progress. We expect this to be completed soon and to be able to make an announcement at the BDFA Conference in October.

BDFA, on behalf of Battle Batten, funds research into CLN5 disease

Due to the amazing efforts of the Dawkins family and the many supporters of the Battle Batten campaign, the BDFA is delighted to be commissioning two research projects into CLN5 disease. The BDFA is enormously grateful to the Dawkins family and the Battle Batten campaign for funding this research on behalf of the whole Batten community.

Evaluating potential drug development in CLN5 cell lines

The projects will provide complimentary approaches to studying the mechanisms of CLN5 disease with the ultimate aim to provide a platform to look for new therapeutic targets and potential treatments.



A group of scientists (Dr Robin Ketteler, Dr Daniel Little, Professor Paul Gissen – all pictured above – and Dr Sara Mole, not shown) based at the MRC Laboratory for Molecular Biology (LMCB) have been awarded a grant of £20,000. The MRC LMCB is a Medical Research Council-UCL University Unit and centre for research into fundamental aspects of cell function and their links to human disease. The scientific focus of the MRC LMCB is to develop new molecular understandings of cell function and behaviour through discovery-based research.

The group will use cells derived from patient skin biopsies with the ultimate aim to identify possible treatments for CLN5 disease. Professor Paul Gissen says:

"It is not possible to study how a mutation in the CLN5 gene directly affects the cells that die because these cells are located inside the brain. However, as a result of recent discoveries, scientists can now take skin cells from a patient and turn them into special cells, called Induced Pluripotent Stem cells or iPSc that can be used to make any cell in the body including nerve cells. This makes it possible to study the CLN5 mutations in the very types of cells that die in the disease. In this project we aim to produce nerve cells from patients' skin cells and then use them to study the effect of many different drugs. We hope to be able to identify some drugs that can be used to develop new treatments for CLN5 disease."

The work will be undertaken by Dr Daniel Little, who already has experience working on producing iPS cells from patients with various diseases and then using these to investigate disease mechanisms and test potential therapeutic approaches.

Uncovering fundamental differences in the cell biology and biochemistry of CLN5 disease

Dr Emyr Lloyd-Evans (photo, right) has worked in the field of research into lysosomal diseases for the last 15 years. As part of his Masters degree he worked with Professor Tony Futerman on Gaucher disease at the Weizmann Institute in Israel, before moving to the University of Oxford to do his DPhil and post-doc research on Niemann-Pick disease type C with Professor Fran Platt. Since the end of 2010 he has been establishing his own research group at Cardiff University where he continues to research the mechanisms underlying abnormal cell function in lysosomal storage diseases. The lab currently has two post-docs and three PhD students all of whom are working on projects related to lysosomal diseases from fundamental cellular work through to zebrafish models and drug testing.

Emyr's lab has a developing interest in the neuronal ceroid lipofuscinoses (NCLs). They are particularly interested in uncovering cellular processes that are not working correctly in the NCLs. Using this information they try to piece together the cascade of events that occur within the cell following loss of gene function so that they can target therapies to the earliest possible disease event. This CLN5 disease project that has been funded by the BDFA on behalf of Battle Batten will initially aim to characterize whether essential cellular processes such as energy production, transport and recycling of nutrients, and cellular signalling are working correctly or not in CLN5 disease cells. The ultimate aim is to attempt to use information of any potential differences between healthy and CLN5 affected cell lines for drug development.

The funding provided by the BDFA has been matched by Cardiff University enabling the project to be converted into a three year PhD. There was an amazing response to the advert with some excellent applicants and in July candidates were interviewed for the PhD. The successful candidate is Katie Shipley (photo, left) who is currently doing an MSc at UCL where she is working on a Parkinson's disease project. Katie is an incredibly enthusiastic student who already has many of the skills and techniques the Lloyd-Evans lab is looking for and we are sure she will do a great job with this project. We all look forward to welcoming her to the Lloyd-Evans lab and the NCL community in October.

DEM-CHILD

The DEM-CHILD registry project led by Angela Schulz, MD, of Germany's Children's Hospital and University Medical Center Hamburg-Eppendorf aims to collect the world's largest, clinically and genetically best characterised set of NCL patients.

The map shows the current partners who will be meeting in Hamburg in November to continue to work together to develop and expand the project. Patient organisations are an integral part of the consortium and our Scientific Officer, Heather Band, will be representing the BDFA.

Registries are of vital importance for a number of reasons. Scientists and pharmaceutical companies are more likely to conduct research on a given rare disease if they find a patient registry in place. Registries also assist in understanding the disease, recruiting patients for clinical trials, tracking genotype and phenotype associations, safety monitoring, identifying novel biomarkers and modifiers, and developing novel therapies for NCLs. The collection of Batten patient data will also assist in achieving earlier diagnosis and proactive interventions for those affected, increasing quality of life and supportive care.

The BDFA and BDSRA as well as many other family foundations recognize the importance of this work and are providing financial support to the project.

The letter opposite details how UK patients can participate and, if you have not already done so, please contact Dr Ruth Williams to find out more about the project.

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 wishes to find out more.

Evelina London Children's Hospital

Guy's and St Thomas' 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

Research Studies and Clinical Trials

There are currently two major clinical research centres seeking to enrol patients to participate in ongoing clinical studies.

Weill Cornell Medical College

CLN2 Gene Therapy study at Weill Cornell Medical College

The Department of Genetic Medicine at Weill Cornell Medical School is conducting a gene therapy study for the CLN2 gene. Patients with the diagnosis of Late Infantile NCL, who meet the eligibility criteria, would be enrolled in the study for 18 months. The study aims to determine whether gene transfer surgery, in which an experimental drug call AAVrh.10.cuhCLN2 is administered to the brain, can be achieved safely and whether the procedure will slow down or halt the progression of the disease.

There is no compensation for participating in this study. The costs of travel and accommodation are not covered. There are no costs associated with the tests and procedures that are conducted under the scope of the research study.

For more information please contact Gerardine, Research Manager, at ger3001@med.cornell.edu.

Please search for "Safety Study of a Gene Transfer Vector (Rh.10) for Children with Late Infantile NCL." on the **www.clinicaltrials.gov**

UNIVERSITY of ROCHESTER MEDICAL CENTER

CellCept for Treatment of Juvenile Neuronal Ceroid Lipofuscinosis (JUMP)

It must be stressed that this trial is only open to US patients who meet the eligibility criteria. The clinical trial at the University of Rochester Medical Center (UMRC) is to learn if mycophenolate (CellCept) is safe and well tolerated in children with JNCL. The JUMP study focuses on evaluating CellCept and its effects on the symptoms of JNCL.

The trial is ongoing and to date 19 patients have been enrolled. No data is available at present but once all the patients have completed the trial they aim to present their findings by the end of the year. There is more information regarding the trial on the website provided by the US National Institutes of Health, **www.clinicaltrials.gov**.

Heather Band
BDFA Scientific Officer

BIOMARIN

BioMarin BMN190 Trial for CLN2 (Late Infantile Batten disease) Update

Following the press release from the Californian pharmaceutical company BioMarin in January 2015 providing preliminary data from the Phase 1/2 BMN190 trial in CLN2 (Late Infantile) Batten disease, the trial continues at the three European sites in London, Hamburg and Rome alongside sites in the USA. The BDFA reported this press release and its encouraging results in the last edition of the newsletter and it is available on the BDFA website.

The trial is now fully enrolled, a number of the children have completed 12 months on the BMN190 study and have entered the extension phase which has followed on from the main study. The Phase 1/2 pivotal study is an open-label, dose-escalation study in patients with CLN2 (Late Infantile) Batten disease.

The primary objectives are to evaluate the safety and tolerability of BMN190 or cerliponase alfa and to evaluate effectiveness using a CLN2 disorder-specific rating scale score in comparison with natural history data after 48 weeks of treatment. Secondary objectives are to evaluate the impact of treatment on brain atrophy in comparison with CLN2 natural history after 48 weeks of treatment and to characterize pharmacokinetics and immunogenicity.

The study enrolled 24 subjects at five clinical sites for a planned treatment duration of 48 weeks. Complete results are expected in Q4 2015.

The BDFA continues to provide support and advocacy to all of those families enrolled at European sites on the trial and will continue to work very closely with BioMarin, European regulatory bodies and partner patient organisations to ensure the availability of an effective treatment as soon as possible.

If you would like any further information about the trial please contact Andrea West on **01252 416110** or email: **andreawest@bdfa-uk.org.uk**.

BDFA

BATTEN DISEASE FAMILY ASSOCIATION

Bringing light to Batten

Hey Charlie

BDFA Charity Single

Hey Charlie is a moving song composed by Ian Frazer Smith to represent families affected by this devastating disease and be the "Voice of Batten"

Recorded this summer at Abbey Road Studios with the beautiful voices of Cantate and with enormous thanks for the kind sponsorship of Keith and Gillian Banks

All profits from the sales will go to the BDFA to fund its work supporting families and fighting for a cure