

Batten Disease Newsletter

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



BATTEN DISEASE FAMILY ASSOCIATION

Bringing light to Batten's

Issue 29 Autumn/Winter 2014



Batten Disease Family Association

Registered Charity No. 1084908



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From our Chair and Chief Executive



Mike

We think it is fair to say that the last six months have thrown us some real challenges to manage. We feel we have become a victim of our own success and keeping up with the pace of our expansion over the last few years has been particularly challenging recently.

We have nothing but admiration for our incredibly committed and passionate staff who have been massively over-stretched in the last few months. It should be very pleasing to all of us that they continually go out of their way to serve us all.



Andrea

To manage these challenges, Andrea is actively looking for additional quality staff members to support the membership's needs. We have found it particularly difficult to find high quality staff to meet the needs of families in the recruitment of a new Support and Advocacy Partner.

Recruiting to this role remains our highest priority but we must have the right person for this vital role supporting families. It has been our concern that without someone in this role, many will feel the core value of the Charity is being lost. We can assure everyone that this is not the case and we remain determined to fill this role very shortly with a quality individual who will raise the profile of this element of the Charity back to its rightful place.

The one thing that staff and Trustees have needed but haven't had is enough time. We sincerely hope that we can build up our workforce to give all our staff sufficient time to carry out their roles to the best of their ability and to further build on this strong foundation.

In addition to a Support and Advocacy Partner, you will see from page 3 that we are appointing our first Clinical Nurse Specialist at Great Ormond Street Children's Hospital. This is a very exciting development for the Charity and is the culmination of hard work and commitment on the part of our supporters and staff.

We have also consolidated our fundraising team with extra administrative support for our regional volunteer fundraising groups. We would like to thank Naked Fundraising for their work and commitment over the last two years and wish the team well for the future. They have always been committed and passionate in their fundraising for us and this is not goodbye but more of a farewell as we hope we'll use Naked Fundraising on specific projects in the future.

As the team grows, we have needed to look at how we can work more efficiently to maximise our time and resources. We

are looking forward to investing in our first office based in Farnborough, Hampshire which will provide a much needed central base for staff to consolidate the BDFA's current services and help us to develop further.

Since our beginnings in 1998, our Trustees remain central to the culture and direction of the BDFA. Every BDFA Board member is committed to their role and you will have the opportunity to meet with them at the BDFA AGM and Family Conference in November. Many of our Trustees are very stretched for time because they work full-time and/or care for the needs of their affected children.

Whilst we have an incredibly diverse and talented team, it is always a challenge for everyone to perform their vital governance role for the Charity. We know we ask this at every opportunity but if you have the time and the inclination to join the team, please speak to any of the Trustees or to Andrea. Your support and most importantly your time would be extremely welcome.

We continue to develop a reliable funding base for our services and we have again beaten our budget and delivered record income to enable us to continue to deliver our services to families. Our list of supporters continues to grow as does our geographical spread and reach.

It would be wrong to finish without mentioning Heather Band, our Scientific Officer whose patience and perseverance has been incredible this year. She has played a major role in the appointment of the Clinical Nurse Specialist and continues to consolidate the BDFA research agenda both in the UK and worldwide. I'm delighted that through the generous support of Sir Ian and Lady Kathryn Botham and Beefy's Foundation we have been able to fund the fourth year of research into Gene Therapy for the Eye under Dr Sara Mole and Professor Ali (page 19).

Many challenges lie ahead for the BDFA team but they are good challenges to have and we remain in a positive place. Please do come forward if you have time, ideas and the inclination to support an amazing organisation.

With thanks to you all, and hoping to see as many of you as possible at the Family Networking weekend. If we don't, we hope you winter well and if you're a fundraiser please do keep up all your amazing work or if you'd like to become one please do call one of the team.

Best wishes,

Andrea

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

Mike

Michael O'Connor, Chair

Front cover image: The Robinson Family. Photo by Ben Marlow

Welcome to our New Board Members



Briony

BRIONY LUMB

Briony is an architecturally-trained Director of Business Development who has worked globally both client-side and for professional services firms, prior to setting up her independent consultancy in the UK. Her work has taken her to live in South Africa, the USA, Holland and Singapore before settling with her partner in West Sussex.

Her interests include architecture, kayaking and travel. She has come to the BDFA via fellow Trustee Shaun Andrews, who she knows professionally.



Catherine

CATHERINE SERMON

Cath is a Campaign Director at a charity called Business in the Community, where she has worked for over 15 years. Over this time she has worked with a wide range of companies, charities and government departments to achieve social change.

As godmother to Lily O'Connor, she is passionate about supporting the BDFA and excited to join the Trustees Board. She lives in Cambridgeshire with her family.

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Breaking News

After much hard work fundraising by our supporters and ongoing commitment from our staff team and Trustees, we are delighted to announce that the BDFA has now funded a Batten disease Clinical Nurse Specialist at Great Ormond Street Children's Hospital in London.

We have always viewed this role as a critical first step in developing consistent, strong services for families living with an NCL diagnosis and we also hope that it is the start of a strong collaborative working relationship between the BDFA and GOSH. Recruitment for the role is underway and the BDFA will play an intrinsic part in that process to represent the voice of families.

We look forward to introducing the successful applicant to you all in the very near future and working together to drive forward a robust service for families living with Batten disease in the UK.

The BDFA Small Grants to Individuals Scheme



The BDFA appreciates that families are often faced with financial challenges when caring for someone with Batten disease.

The BDFA aims to provide assistance to help families in the UK cope with these circumstances and has a Small Grants to Individuals Scheme (maximum award is £500). This is designed to provide a rapid practical response to difficult situations.

If you wish to make an application or discuss the process and your eligibility, please contact the BDFA Support and Advocacy Partner.
(0800 046 9832 / support@bdfa-uk.org.uk)

Dates for your Diary 2014

OCTOBER 2014

22 - 25 October - NCL2014, Cordoba, Argentina

NOVEMBER 2014

8 - 9 November - BDFA AGM and Family Networking weekend, Coventry

20 November - Kidz Up North

EventCity, Barton Dock Road, Manchester M17 8AS

MARCH 2015

19 March - Kidz in the Middle

Jaguar Exhibition Hall, Ricoh Arena, Coventry

JUNE 2015

4 June - Kidz South, Rivermead Leisure Complex, Reading
Some of the largest, free UK exhibitions dedicated to children and young adults with disabilities and special needs, their families and the professionals who work with them. For free tickets for these events, call 0161 607 8200

16 June - PSDL Lab Day at Kings College London (date tbc)



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



Farewell from Naked Fundraising



After two and a half years of supporting the amazing BDFA fundraisers, we at Naked Fundraising are sad to announce that our contract comes to an end at the end of October.

We would like to thank all the charity's supporters for their incredible enthusiasm and inventiveness – long may you continue to raise vital funds to help find a cure for this terrible disease.

To all the parents we have come to know, we wish you all the very best for your journey. You and your children are amazing. Be strong.

Nikki, James, Annabel & Emma



Great Scotland Swim



Back in May this year I played cricket as part of an invitation cricket team for a close friend against his local side in Hampshire. After the game he told me about the daughter of one of our team's players who has Batten disease. I had heard a little about the disease but had no idea of its severity and our conversation put into crystal clarity the importance of our own ongoing health and family wellbeing.

My son and I decided to enter the Great Scotland Swim in August to raise money for the BDFA in the hope that one day there will

indeed be a cure for this most dreadful, cruel and despairing disease. My son, Sam, who has just had his fifteenth birthday, did the half mile swim and I (yes, probably a golden oldie at 50!) swam two miles in the beautiful Loch Lomond which is at the mouth of the Trossachs and National Park, a 45 minute train ride east of Glasgow.

Conditions started off near picture perfect with still glassy water, no wind and 16 degrees water temperature. By early to mid-afternoon, however, there was wind which created choppy waves and made for a pretty challenging outbound leg. The swim was very well organised with 13 different "waves" through the day from 09:00 to 15:30.

For each wave there were kayakers strategically positioned to ensure that swimmers did not go "off piste" and that they safely completed the course. Each wave had acclimatization and warm-up instructors to get swimmers in the mood. We understand that some 3500 swimmers took part over the course of the day.

As an open water swim venue, we can both thoroughly recommend it and I think the "genie has come out of the bag" for Sam as he would like to do another open water swim (there seems to be plenty of choice of events including Big Chill events at www.chillswim.com) and he is also looking at sprint triathlons!

We are also delighted to have beaten our fundraising target of £1000 and have now raised over £1400 for the BDFA. We hope it helps the BDFA in its vital work.

Stephen Lucas



On 20th December 2013 we were given the news that Ellie Mae & Caleb had Late Infantile Batten disease. Ellie Mae had started with seizures 18 months earlier and due to some developmental concerns we had been told it was unlikely to be just epilepsy but rather a symptom of something else.

When Caleb's seizures started we were desperate to find the cause so he could be treated before any developmental issues started but unfortunately this wasn't possible and we are now focussed on caring for them both as much as possible.

From the start we were blessed with a lot of support from family and friends. Our doctors have all been excellent in assessing the impact on us as a family when planning tests and appointments. And my colleagues have been amazing. I was given three weeks off over Christmas to spend with the family and in the New Year they started fundraising to send us to Disney World for a holiday to help us gain some wonderful memories with the kids.

Our Church supported us financially when the NHS wouldn't and practically with meals and cakes to keep us going. Both sides of our families are also always here for us.

As we have received so much support from those around us I wanted to do something to fundraise for the BDFA to help them support others. Last April I completed a month of running where I was sponsored to run five miles every day. I hate running and hadn't done any for 20 years so it wasn't an easy challenge.

Sponsorship was going well but really took off when I invited our Church friends to join me on Easter Monday. Twenty five people

turned up and one of them had invited Greg Mulholland, our local MP. Greg ran with me and arranged for photos at the end which his team got into the local papers. The story was covered in four separate newspapers and that weekend the sponsorship went up by over £1000. Altogether I raised £6000 for the BDFA.

In September my company arranged another series of events for employees to raise money for a charity of their choice.

I completed my first (and hopefully last) marathon on 25th September, setting off from Kew Gardens and running along the Thames to the Olympic Park in Stratford. I didn't have a lot of time to train but managed to get through it in one piece and collected another £2000 for the BDFA.

Duncan Brownnutt



Both our cousin's children, Ellie Mae and Caleb, were recently diagnosed with Late Infantile Batten disease. The BDFA have been a great support to them by helping them apply for funding when needed and by talking to the children's school and pre-school, to help them help the kids.

We wanted to raise money for Ellie Mae and Caleb to have an all-expenses-paid return trip with their Mum and Dad to London (from Leeds) and visit a number of London attractions. A share of the money would also go to the BDFA to help increase awareness of the condition and search for a cure.

So we wanted to think up a challenge with a London theme and our geeky side got the better of us. We decided to attempt to visit all 270 Tube stations in 24 hours. No, we weren't going to be collecting train numbers or wearing kagools – we just wanted a challenge that was unusual and had a London theme. The challenge was only just possible, and involves taking the first train out at 5.00am, and getting back before the network shuts down at 1.00am the next morning. And if you think that sitting down on trains all day isn't much of a challenge, just consider those hard tube seats, in sweltering summer heat, travelling the distance from London to Paris in a day.

Richard & Michael Brownnutt

In February 2014 Andrew and I decided to fight back against the lack of research into our twin boys' condition. Little did we know we would become the first CLN5 family in the UK to fund a research project to try to help our children.



At the BDFA Family Networking Weekend in October 2013, there were sessions organised for families to meet UK researchers and during a session on CLN5 and CLN8 we learned that research projects being carried out in other types of Batten disease could also be applied to rarer forms including CLN5 which our boys Freddie and Louie have. But there was no funding available at present... all that was needed to start was around £20,000.

To Andrew and me this seemed a huge amount of money to raise ourselves. But equally it seemed a small amount of money to progress research towards therapies and ultimately a cure. Time is not on our side. Over the following few months we wondered if we would have the time and strength to raise the money as well as caring for our boys. However, their smiles had returned since losing their vision which spurred us on.

Andrew had applied to run the London marathon and his place had been confirmed for April 2014 – this would be a start! He asked people to sponsor him at his local gym and a lady named Suzy came forward and offered to help set up a JustGiving page.

We were hoping to raise around £1,000 and told her about the £20,000 needed to fund a project. When she learned how cruel Batten disease is she wanted to help us reach our goal. Her friend Maureen also offered her help and this resulted in our own campaign. We decided to call it “Battle Batten” with 100% of the money raised being donated to the BDFA for research into CLN5.

Within a matter of weeks we had our own website and a reporter from our local paper adopted our campaign. Our local community has really pulled together to help – more so than we could ever have imagined.

There have been letters containing money from strangers through our letterbox, charity football, a ladies' headshave, cake sales, Tesco bag pack, sponsored no-alcohol, fit-a-thon, 4x4 blindfold event, 10 mile swim, the list is endless...

We have done things we never thought we could do including public speaking, live radio and numerous TV interviews. Each time we had to think of Freddie and Louie.



The money raised so far has gone beyond what we ever believed possible – £40,000 in six months. And there is still more to come. Andrew is running the Robin Hood Half Marathon blindfolded and we are holding a “Battle Batten” Ball on 8th November to raise further funds and celebrate everyone's achievements.

The BDFA offered to organise a call for research proposals for CLN5 and this was launched on the BDFA website in September 2014 - <http://www.bdfa-uk.org.uk/research-rfp-cln5-disease>. Submitted projects will be reviewed in November, with a project chosen in December. For more details, see p16.



Research Studies and Clinical Trials for NCLs

There are currently three major clinical research initiatives seeking to enroll patients meeting the eligibility criteria.

- The Department of Genetic Medicine at Weill Cornell Medical College in New York City is conducting a gene therapy study for the CLN2 gene.
- The pharmaceutical company BioMarin is pioneering an enzyme replacement therapy (BMN190) for CLN2 (Late Infantile Batten disease) at centres in Hamburg, Rome and London.
- The University of Rochester Medical Center (UMRC), USA, is currently recruiting US patients for a clinical trial to learn if mycophenolate (CellCept) is safe and well tolerated in children with JNCL.

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 CLN2 (Late Infantile Batten disease), 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 called 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 also 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 Denesy, Research Manager at: dem2026@med.cornell.edu or visit: www.clinicaltrials.gov and please search for "Safety Study of a Gene Transfer Vector (Rh.10) for Children with Late Infantile NCL."

CellCept for Treatment of Juvenile Neuronal Ceroid Lipofuscinosis (JUMP)

This trial is only open to US patients who meet the eligibility criteria. It is based at the University of Rochester Medical Center (UMRC) and is aiming 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.

As the trial is ongoing no data is available to be released at present. So far, 14 patients have completed the trial, a further two are enrolled at present and UMRC are continuing to recruit and enroll the remaining patients. There is more information regarding the trial itself on the website provided by the U.S. National Institutes of Health, www.clinicaltrials.gov.

Heather

Heather Band, BDFA Scientific Officer

B:OMARIN™

BioMarin CLN2 BMN 190 Clinical Trial

The BDFA continues to work very closely with the Californian pharmaceutical company, BioMarin, to deliver the BMN190 clinical trial of enzyme replacement therapy for children with a diagnosis of CLN2 (Late Infantile Batten disease). Alongside the medical teams at each of the trial sites and the home country patient organisations, we have been supporting families to meet the challenges of participating in a clinical trial. We would like to thank the A-NCL Associazione Nazionale Ceroido Lipofuscinosis in Italy and the German patient organisation NCL-Gruppe for their continued work in their own countries to support families and medical teams involved in the trial.

Since our last update the Bambino Gesù Hospital IRCCS, Rome has begun recruiting to the trial and Dr. Nicola Specchio (Head of the Epilepsy Surgery Unit at the Hospital) will be the Principle Investigator for the study. The Bambino Gesù Hospital has expertise in caring for children with complex and challenging medical disorders and is an established centre for the diagnosis and management of children with Batten disease.

Trial recruitment has ceased at the Evelina London Children's Hospital and it is hoped that another site will open at Great Ormond Street Children's Hospital in London in the near future, with Professor Paul Gissen as the Principle Investigator. The trial continues in Germany at the Children's Hospital, University Medical Centre, Hamburg-Eppendorf where children are being recruited to the study with Dr Angela Schulz.

The trial is enrolling approximately 22 patients for a treatment duration of 48 weeks and families have been admitted to the trial centres from across the world. The BDFA has produced a patient information leaflet for the BMN190 trial which can be obtained from admin@bdfa-uk.org.uk. We have also produced a general leaflet describing the process of clinical trials.

We recognise the decisions around clinical trials can be very challenging for families. The BDFA is able to support families considering these issues and to put them in touch with other families who have taken part in clinical trials. As always, all questions about a child's medical care should be directed to the relevant health professional.

For more information about the trial or if you have any questions please contact: Andrea West on 01252 416110 or by emailing andreawest@bdfa-uk.org.uk



Linden Lodge School



Linden Lodge School is situated in very pleasant and leafy grounds in Wimbledon, London. It provides a high quality educational experience for children and young people registered visually impaired, including those who are multi-disabled, visually impaired, deaf/blind and with profound and multiple learning difficulties.

It is a Specialist Regional Centre for children aged between two and nineteen years of age and currently receives pupils from 28 Local Authorities throughout southern England. Pupils can attend as either day pupils or as weekly boarders (Monday to Friday, term time only).

Over the past ten years Linden Lodge has continued to develop as a centre of excellence and has built a close affiliation with the BDFA, its families and young people. The school's facilities, and its knowledge and understanding of Batten disease ensures that they are able to meet the complex needs of this group of children and young people.



Linden Lodge has continually reviewed its curriculum offering different approaches to best suit children's needs. The school is well versed in planning for children at all stages of the disease and is well equipped to manage multiple learning difficulties and complex health needs. It benefits from an on-site, integrated multi-disciplinary team including physiotherapists, occupational therapists, speech and language therapists as well as nursing care.

The school prides itself on The Family Partnership Model of working which promotes exploration, understanding and detailed planning and partnerships with families, believing this ensures Linden Lodge is a truly special place to learn.



The school has amazing facilities. All teachers are qualified Teachers of the Visually Impaired (or in training) and therefore use specialist approaches to help pupils develop or maintain their sense and understanding of the world around them. Staff use Braille, Moon and Objects of Reference (both standardised and personalised) alongside augmentative communication aids.

The site also includes a Learning Resources Centre housing a sensory library, a swimming pool, a hydrotherapy pool and a movement development centre where pupils engage in specialist approaches including Rebound Therapy. The gym and fitness suite are adapted and additional therapy input is highly valued, including music therapists, a movement therapist and input from a yoga and massage therapist. Specialist teaching facilities are also provided in the art room and the food technology suite which are both well used.

Richley House is the residential element of the school and has a staff team (including waking night attendants) to ensure opportunities to develop and maintain independent living skills. The school ethos is to be constantly reflective about the needs of its families and pupils. It is fundraising for the Isabel Family Centre which will be a space where families can be supported including time with the family counsellor, with other families and with support organisations such as the BDFA.

To further build on the strong relationship with the BDFA, Linden Lodge hosted a BDFA education training day in October and look forward to more collaborative working in the future. For more details about the school please visit the **website <http://www.lindenlodge.wandsworth.sch.uk/>**

NEW Fundraising Toolkit

We often speak to people who have never been involved with fundraising before but are keen to help. We have therefore produced a toolkit to get you started and to help you to think of everything you will need to do.



Fundraising Toolkit



Top tips to make your fundraising easier

Fundraising is fun and rewarding but it can be hard work especially if you have never done it before.

Hopefully this toolkit will steer you in the right direction and keep you on track.

It takes you through everything you need to run a successful event including fundraising ideas, how to get started, how to stay safe and how keep it legal.

It is available in electronic or hard copy. Simply call the BDFFA office on 01252 416323.



WE ARE NOW COLLECTING USED STAMPS

WE CAN TRANSFER THE STAMPS INTO DONATIONS FOR THE BDFFA, THE MORE STAMPS WE HAVE, THE MORE WE GET.

ALL STAMPS SHOULD HAVE NO MORE THAN 1CM OF ENVELOPE AROUND THEM AND SHOULD NOT BE TRIMMED SO THAT THE PERFORATIONS ARE DAMAGED

ACCEPTED STAMPS

UK OR FOREIGN
1ST OR 2ND CLASS
USED OR MINT
ON OR OFF PAPER
COMMEMORATIVE

PLEASE SEND YOUR STAMPS TO
FUNDRAISING STAMPS LTD
59 MITRE COPSE
BISHOPSTOKE
EASTLEIGH
SO50 8QE



BDFFA Annual Walk



Lucy

The weather was typically English – we needed ice-creams to cool down and (BDFFA!) umbrellas to keep dry at times – but we were a bright presence on the landscape in our orange T-shirts as we enjoyed walking through the beautiful Hampshire countryside.

We comprised a varied bunch of adults, children, dogs, pushchairs, newcomers as well as die-hard loyal walkers who have been on each and every one of the BDFFA walks that have taken place.

There was all the fun of camping, wonderful home-from-home food provided at regular intervals, a quiz as well as animals and fun at Butser Farm. There was time to be active and time to just catch up with old friends and make new ones.

Circumstances meant that I could only be there for one day but my five year old is already looking forward to camping and coming for the whole weekend next year! Thank you to our Fundraising team for all their hard work in organising such a lovely weekend.

Lucy

Lucy Roose

Ben Marlow



Ben

I became aware of Batten disease and involved in the Batten Disease Family Association through my friendship with Ben Pickering. Ben had CLN2 Batten disease and sadly passed away in 2009.

I am a volunteer photographer and videographer and have given my time to the BDFFA in this capacity.

I produced a video of this summer's BDFFA walk which can be found on YouTube (<http://youtube/nlOqXktMMHs>) as well as the photos that can be seen in the centre spread (pages 10 and 11).

I have also taken photos at BDFFA Lab days at King's College & UCL, and for previous newsletters as well as other publications. I am looking forward to working with Paul Robinson at the forthcoming AGM and networking weekend in November.

Currently I am studying for A-Levels in Media, English, Art, Psychology and Critical Thinking. Ultimately I would like to go to university and further my passion for media production.

Ben

Ben Marlow

B DFA Annual Walk







Making the most of precious time

Hello, we are the Hadman family. Our beautiful daughter Effie was born in the summer of 2010 and was, at first, a perfectly healthy child. She met all of her milestones, even exceeding some of them. She loved to go to the local farm, run around at the park and play with her friends. In December 2012 when Effie was two-and-a-half, she seemed run down and anxious. We thought she may be coming down with a cold, but in reality it was much worse.

In January 2013, Effie had her first seizure – a drop attack in the bath. The following day saw three seizures, by the end of the week she was having ten a day and was admitted to hospital. Effie was diagnosed with epilepsy and started on seizure medication. Over the next six months, the seizures became increasingly difficult to control and her medication was regularly increased. Effie's development slowed considerably over the following months. The day before her third birthday in June 2013, she was admitted to hospital again unable to walk, with slurred speech and hallucinating. More medication was added which seemed to help and she was able to walk again, albeit very wobbly with lots of falls. As the year progressed, Effie's ability to walk and talk continued to decline and it was clear that she had started to forget things she had learnt.

In January 2014 we were given the devastating news that Effie has Late Infantile Batten disease. Effie was just three-and-a-half years old. At this time her little brother George was 6 weeks old and we had an agonising wait to find out if he was affected too. Fortunately he doesn't have Batten disease, which was a massive relief for the whole family.

The months after diagnosis were a very dark time for us but we have learnt not to look too far ahead and to focus on what Effie can and loves to do, rather than what she has lost. We are determined to get as many giggles every day, even on days when we don't feel like smiling ourselves.

People tell you to 'make the most of every day', which can be a difficult concept. It's difficult to feel as though you're making the most of precious time when you're waiting around for appointments or exhausted from sleepless nights. We've learnt that memorable times don't necessarily have to be spectacular trips out (although those are fab!), but can be simply relaxing at home with the family or playing in the garden.

Despite all the "Batten's stuff", Effie is still a fun-loving, rollercoaster-loving cheeky four year old who wants to have adventures. The difficult part is for us to judge when these days will go down well and when they're going to cause meltdowns! Since diagnosis, we have been keen for Effie to experience as many different things as possible. Effie can no longer walk and is becoming more and more dependant but by concentrating on what she can do, we've found that Effie adores rollercoasters! It's something we'd never have thought to go on previously, but we're trying to make the most of Effie's abilities while we can.

We applied to the Make-A-Wish foundation to help us make some amazing memories with Effie and originally asked for a family holiday somewhere sunny. However, at the time Effie's seizures weren't well controlled and we were going through a difficult patch of hospital admissions and screaming sessions and felt that trips away were best planned last minute as we never knew what each day would hold in store. So instead we requested something Effie could use every day and which would bring the most enjoyment for the whole family.

As Effie loves the feeling of moving and is a bit of an adrenaline-junkie, we decided to ask for a bike for Effie so that she could go as fast as possible! She cannot pedal herself and as the disease progresses, she will struggle to sit unaided so with this in mind, we requested a Van Raam O Pair, which is the most incredible bike ever!

Effie sits in the (detachable) wheelchair section at the front and we pedal the bike attachment behind. It can be adapted to give head support when needed, although we try not to think that far ahead. It's fair to say that the harder we pedal, the more giggles we get! It gives Paul and me valuable exercise whilst being out and about as a family. We can be outside with Effie getting fresh air and making the most of our time together.

We always take as many photos and videos as possible. George is still young and unlikely to remember the fun times he's had with his big sister. We would like him to be able to see what a truly amazing, brave and fun loving sister he has.



A Child of Mine

A Child of Mine is a registered charity (1152159) supporting families after the worst loss imaginable – when a child dies. The Charity was set up after the death of my own son to cancer in 2010 and his death highlighted the gaps existing in bereavement care. We wanted to address these gaps in some way and also reduce the stigma and taboo attached to such a loss. And so A Child Of Mine was born.

Almost 4,000 children die every year in the UK. Sadly many of their families will slip through the net and not receive the support they need and deserve. As I am sure you can imagine, the death of a child has an incredible impact on every single family it affects, both emotionally and financially and the truth is that they never fully get over losing their child. For this very reason it is absolutely vital that families are supported when they need it, whether that is 6 months, 2 years, 10 years or longer after their loss.

We are passionate about providing a platform for these families to access information, guidance and support when they need it, and from people who totally understand.

Currently we do this through many channels including our website, which is a complete resource centre for anyone affected by the death of a child, at any age or in any circumstance. We have recently had our own set of publications produced which provide valuable guidance for family members including dads, grandparents and siblings. These are distributed via various healthcare agencies across the UK including Great Ormond Street, Birmingham and Alder Hey Children's hospitals.

A crucial part of our work is building strong relationships and partnerships with all of these agencies. We work closely with all of them to help educate on the needs and experiences of the bereaved parents, enabling them to improve the care and the lives of families in the future.

We have started to deliver some direct services to families within our local area. We now offer small financial grants to families



and have set up our first Sunshine and Rainbow's playgroup for bereaved families with other children. These have been huge milestones for us and something that we would like to develop as part of our long term plan. We are looking to develop a befriending scheme, which will offer fully trained outreach support within the community. We feel this is absolutely vital to ensure the health and wellbeing of every bereaved family going forward. There is no greater support than from someone who has already walked the same path.

We are a young charity but extremely passionate about delivering services and making a difference to the families we support. We have already started to make positive impacts in their lives by increasing wellbeing, improving emotional health and by providing access to much needed information during the difficult days that will lie ahead.

To find out more please visit: www.achildofmine.co.uk or you can follow us on our social media pages:

Twitter - @achildofmine

Facebook - facebook.com/AChildofMine



We are a Farnborough-based patient advocacy charity looking to recruit a full-time

SUPPORT & ADVOCACY PARTNER

If you have a background in nursing or social care, a proven track record of providing support and advocacy services and an understanding of the needs of children and young people with progressive and profound multiple disabilities, we would like to hear from you.

For more information please contact Lucy by email admin@bdfa-uk.org.uk or by phone on 01252 416323

www.bdfa-uk.org.uk

Child Funeral Charity



Arranging a funeral for a child is something no-one can imagine having to do. Child Funeral Charity was set up as the result of like-minded people linked to the funeral industry talking about their experiences of trying to support families arranging a funeral for a baby or child.

In acknowledging the financial pressures, emotional stress, and tremendous difficulties families in such situations encounter, they saw an opening for an organisation which could help provide that support and decided to do something about it.

Child Funeral Charity (CFC) financially assists families in England and Wales who have to arrange a funeral for a baby or child aged 16 or under. Whilst many funeral directors, the clergy and most celebrants do not charge fees, there are other funeral related expenses that bereaved parents struggle to find. Financial support is available to help with such funeral costs, together with practical advice and guidance.

CFC takes referrals and applications for financial support from professionals who have knowledge of the bereaved family and their circumstances. Therefore if you are a funeral director, celebrant or faith representative, bereavement nurse/midwife, hospice manager, GP, Local Authority Registrar, Hospital Bereavement Officer or Medical Registrar, please refer bereaved families to us.

Calls are also welcomed from bereaved families who may have questions about any aspect of arranging a funeral. Please feel free to call us and we will do our very best to help.

**Tel: 01480 276088 (Monday-Friday 9am-5pm) Email: enquiries@childfuneralcharity.org.uk
www.childfuneralcharity.org.uk Charity No 1156387**



Charlotte's Tandems is a charity lending tandems and tag-alongs to people with disabilities or additional needs free of charge. It aims to enable those who are unable to ride a bike safely on their own to enjoy the wonders of cycling. Our borrowers are both children and adults.

Tandems are a fun and safe way to get out and about as friends, a couple, a family or group. They are available for hire for a couple of months at a time.

We now have nearly a hundred tandems and a dozen tag-alongs across the UK. They are with borrowers for the vast majority of the time but looked after by our volunteer helpers. If you would like to borrow one, please contact us via our website: www.charlottestandems.co.uk.

The disabilities and special needs of our borrowers have included severe learning difficulties, Autism, Aspergers, blindness and visual impairment, Down's Syndrome, Cerebral Palsy, among others.

Our only requirement is that each borrower sends us a photo of them using the bike and some comments.



Family Fund is the UK's largest provider of grants to low-income families raising disabled and seriously ill children and young people aged 17 and under. They aim to help ease the additional pressures families face. They can help with essential items such as washing machines, fridges and clothing but can also consider grants for sensory toys, computers and much needed family breaks together. You can apply to the Family Fund if:

- You live in England, Northern Ireland, Scotland or Wales.
- You are the parent or carer of a disabled or seriously ill child or young person aged 17 or under.
- You have evidence of your entitlement to one of the following: Universal Credit, Child Tax Credit, Working Tax Credit, Income-based Jobseeker's Allowance, Income Support, Incapacity Benefit*, Employment Support Allowance*, Housing Benefit and Pension Credit. If you do not receive any of the above, further information may be needed to complete your application. Please only send photocopies of this information as we cannot return it to you.
- You have permanent legal residency in the UK and have lived in the UK for six months.

*We may need to contact you for further information about your household income. For more details, please visit our website: www.familyfund.org.uk.

Ashleigh Lennon

Born 15th of May 2003

Became an angel

11th of May 2014 aged 10

*Twinkle twinkle little star
How I wonder what you are
Up above the world so high
Like a diamond in the sky
Twinkle twinkle little star
How I wonder what you are*

*When the blazing sun has gone
When she nothing shines upon
Then you show your little light
Twinkle twinkle through the night
Twinkle twinkle little star
How I wonder what you are*



Remembering...

The BDFA is here to support families at all stages of the Batters 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 also has a special remembrance book that is available to all family members, friends and associated professionals so they may enter a tribute to someone who is no longer with us.

- **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.



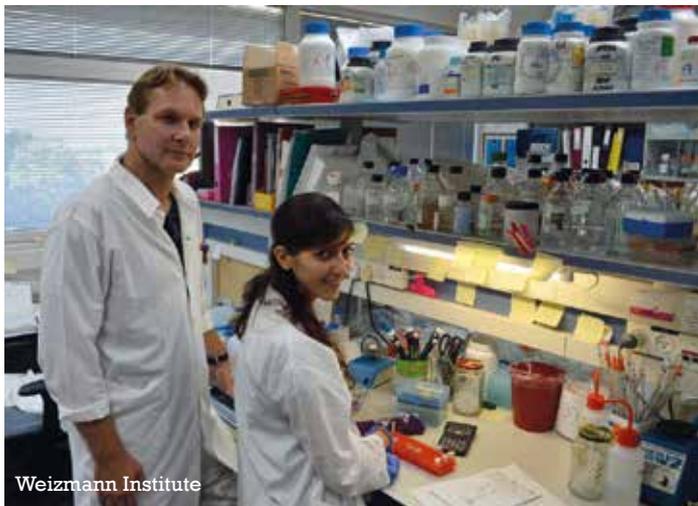
Heather

Introduction

The projects featured in the Research section are very different and were chosen not just for their excellent scientific approaches but also to illustrate the diverse and different projects the BDFA supports.

Researchers, medical experts, health and education professionals must be encouraged and supported to collaborate across all disciplines if we are to achieve effective therapies and ultimately a cure for all types of Batten disease. The BDFA is committed to bringing all these aspects together and our research program aims to directly fund and facilitate research in an innovative way.

We are a major contributor to the American Batten Disease Support & Research Association (BDSRA) Research Grants Awards in 2012-4 (www.bdsra.org) and are continuing to fund a major project at Kings College, London with Professor Jon Cooper and Dr. Brenda Williams chosen from this joint call. Greg Anderson, an NCL-Stiftung/BDFA co-funded Post Doctorate student has been looking at cell based systems for drug discovery in JNCL. The project is due for completion in Spring 2015.



Weizmann Institute

The collaborative PhD studentships program has been very successful with Davide Marotta and Marianna Vieira due to complete their studies in Dr. Sara Mole's lab at UCL by the end of the year. Internationally, Osnat Cohen-Zontag in Professor Gerst's lab at the Weizmann Institute in Israel is now in her second year of studies. The funding for this project is a truly international collaboration between the BDFA, NCL-Stiftung, the BDSRA and Beyond Batten Disease Foundation (US) all contributing funds over the three years.

The amazing fundraising efforts of one family – Andrew Dawkins and Sarah Finney – whose story is featured on page 6 have led to the BDFA's research grant call specifically for research into CLN5.



With the help of family and many dedicated friends and supporters, the "Battle Batten" campaign has raised £20,000 and aims to increase this to £50,000. All the funds are being donated to the BDFA to provide a research call specially for CLN5 research in the UK.

The request for proposals (RFP) went out to researchers in September and the projects submitted are now undergoing the Peer review process. The results of this will be known in late November and the successful project will be chosen in December. It is hoped work will start by the end of the year or early in 2015.

On page 17 we have an example of our Small Research Grants scheme. These grants are intended to be for start-up or "proof of concept" studies, usually in areas where no other funding would be readily available.

At the successful conclusion of such a project the researcher can then seek further funding from other grant awarding sources. In 2012 Sarah Kenrick and SeeAbility were awarded £1500 and have completed their project and are now able to report on their findings.

The BDFA places a very high priority on the development of an International Registry and Database for all the NCLs and has provided support and contributed to the preliminary work required to establish this in the UK. The letter from Dr. Ruth Williams on page 17 invites all affected families to participate, if they wish to, and the BDFA would like to acknowledge all the hard work of Dr. Williams and her colleague Dr. Eleanor Claydon in successfully obtaining approval for UK data to be added.

Page 19 features an update on one of our major projects. In 2011, £100,000 (with the UCL Impact Award Scheme) was awarded to Dr. Sara Mole and Professor Robin Ali for a PhD studentship based at the Institute of Ophthalmology, UCL.

Sophia Kleine Holthaus is nearing the completion of her studies and expects to submit her thesis in the New Year. This is a major piece of work and she is to be congratulated on her achievements in answering many fundamental questions on eye therapy in Batten disease.

The project results have been reviewed and with the generous funding from Beefy's Charity Foundation and funds from the BDFA we are delighted to be able to offer her funding to continue and expand her investigations for another year.



Sarah Kenrick (Registered Manager) and the research team at SeeAbility Heather House have been hard at work on an exciting new research project that should further our understanding of CLN3 Disease.

Unique in the UK, Heather House has a world-renowned reputation as a leading nursing care home for young adults with CLN3 Juvenile Batten disease. The dedicated and professional staff team has combined experience with practice and for over fifteen years has contributed to the advancement of nursing techniques supporting people with CLN3 Juvenile Batten disease.

This new research project, funded by the BDFA, will demonstrate how the experience of best practice at Heather House can further our understanding of how to manage epilepsy in order to minimise some of the most distressing symptoms of the disease process.

The project draws upon a wealth of historical and current data on epilepsy and its management through medication to analyse what works and how reducing the rate of epileptic activity can have a positive impact on the disease process, quality of life and promote longevity.

This will also look in more detail at differences between male and female patients/sufferers and by age groups.

Sarah Kenrick will be presenting the findings of the research during the BDFA Family Conference 2014. If you would like any further information on the project please contact Sarah Kenrick by email at s.kenrick@seeability.org.

Matthew Bradberry, Researcher

Matthew has been working and volunteering at SeeAbility Heather House for over six years.

Prior to supporting Sarah Kenrick on this research project he has obtained a large amount of experience of quantitative analysis in social sciences to postgraduate level at the University of Warwick.

Recently he has been promoting the social model of disability in public affairs with Remploy.



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



Beefy's Charity Foundation & the BDF A give boost to Batten disease research

UK researchers from the UCL Institute of Ophthalmology and the MRC Laboratory for Molecular Cell Biology are currently working to improve the sight of children affected by Batten disease.



As part of this project the BDF A fund Sophia Kleine-Holthaus, a graduate PhD student working with Dr. Sara Mole, who leads research on Batten disease at UCL, and Prof. Robin Ali at the

UCL Institute of Ophthalmology. This research team is developing a treatment that involves inserting healthy copies of the missing Batten disease gene into the cells of the retina to help them to function normally.

Dr. Sara Mole says "This work is a culmination of twenty years research on Batten disease which began by identifying the first gene causing this disease in 1995, and thirteen more since then. We are still working to understand what these genes do but that does not stop us from developing new ways to bring therapies to help those families affected by this devastating and distressing disease".

The BDF A are committed to continuing to support this vital work to bring the goal of a potential treatment closer. Earlier this year we were delighted to learn that Ian Botham's Beefy's Charity Foundation had awarded a generous grant of £20,000 to enable us to continue to fund this research for a further year.

Professor Robin Ali together with eye surgeon Mr. James Bainbridge have already successfully treated patients with a condition called Leber's Congenital Amaurosis (LCA), another rare inherited eye disease, in the first clinical trial of its kind in the world.

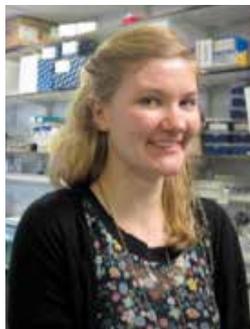
This treatment has been shown to be safe and can improve sight. The findings were a landmark for gene therapy technology and could have a significant impact on future treatments for many eye diseases, including Batten disease.

For further information about this or other BDF A funded research please contact BDF A Scientific Officer, Heather Band, email: research@bdfa-uk.org or 01243 672660.

Breaking News...

The BDF A is taking a leading part in a consortium of UK and European scientists headed by Dr. Sara Mole at University College London who are in the process of applying for a Rare Disease EU grant for 5.5 million Euros. The first round of submissions is in October 2014 and if successful a full bid will be made in February 2015.

Gene therapy to treat visual failure in NCL PhD project undertaken by Sophia Kleine-Holtthaus



Loss of vision is a significant symptom of Batten disease. It is one of the first symptoms in JNCL and therapies addressing this would improve quality of life for those affected. The development of a treatment for the eye should also provide valuable information on possible treatments to improve brain function.

This PhD project sought to explore the feasibility of developing gene therapy for Batten disease in the eye. For this work a mouse model was used that is similar to CLN3 (Juvenile) in that it presents with vision failure due to a genetic defect in the CLN6 gene.

The first aim of the project was to investigate the progression of the loss of vision in the mice to narrow down when gene therapy treatment would be most effective. Next, viral vectors were produced that carried healthy copies of the affected gene and these were injected before any onset of the disease. The virus was then taken up by photoreceptors, these are the cells of the retina that are most affected and predominantly die in this model.

The second aim was to investigate how much of the healthy new gene would be required and if it had any adverse effects on the eye. In the course of this work the administration of very high doses of the virus was found to give good uptake but had detrimental effects on vision and the survival of cells in the retina. Lower viral doses, which resulted in a less widespread treatment of the retina, overcame these problems but did not show any of the required beneficial effect on vision or cell survival.

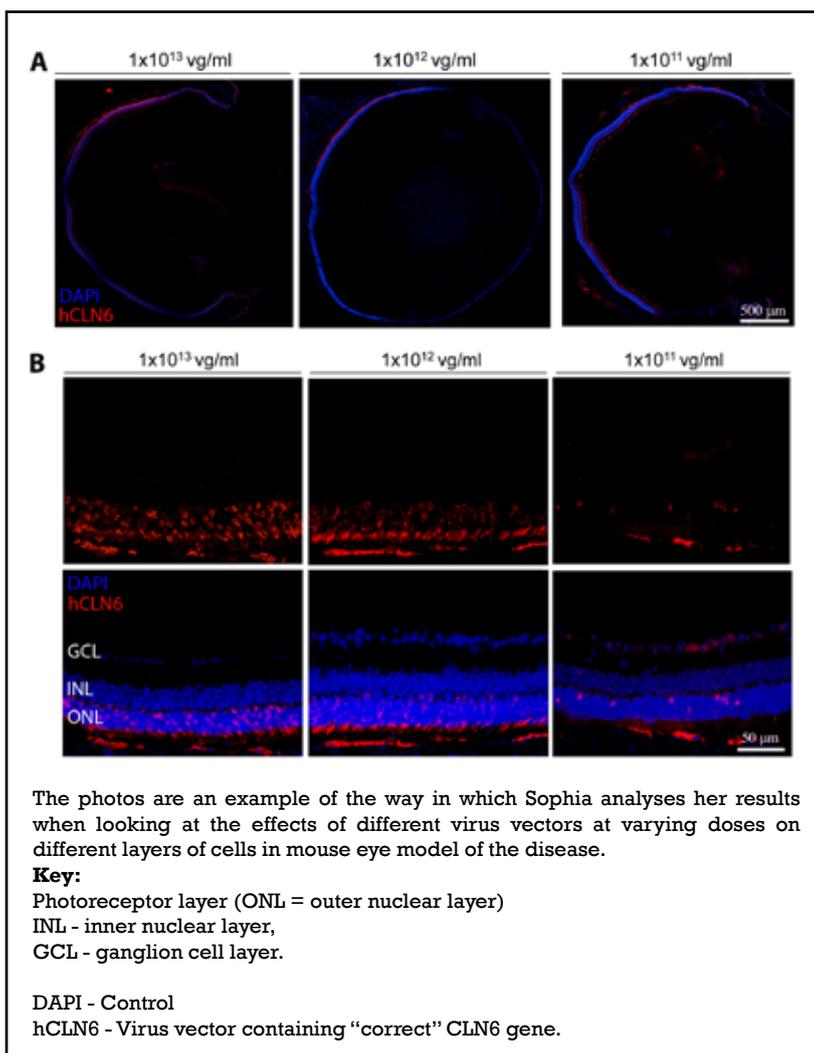
Other strategies were employed to enhance the delivery system, giving high doses of a modified virus to introduce a widespread treatment of the retina but a low expression of the healthy gene in individual photoreceptor cells to avoid toxicity but no improvement was found in the vision. This technique had proved successful in other eye conditions, as researchers in Prof. Robin Ali's lab have shown positive effects where the photoreceptor cells are dying due to a genetic defect. The treatment of these cells by gene therapy improves vision. This might mean that other eye cells also need to be treated to give beneficial effects in Batten disease.

Sophia then looked at other cells within the eye and was able to show that the CLN6 gene is expressed to a higher extent in a cell type in the retina called bipolar cells than it is in photoreceptors. Bipolar cells are cells that receive electrical stimuli from photoreceptors for the next stage of the journey of the electrical impulses towards the brain. Defects of bipolar cells are very rare and only a handful of human eye conditions exist where they are known to be affected.

Generally, bipolar cells are not as sensitive to changes in their environment as photoreceptor cells which could explain why there is no obvious death of bipolar cells in the diseased mice. It is therefore possible that the lack of healthy CLN6 in bipolar cells leads to changes in the retina that photoreceptors react to and die.

To test this hypothesis, Sophia will focus her attention in the next year on investigating whether bipolar cells undergo changes in the CLN6 mice. There is still much work to be done and it is important to investigate the potential involvement of bipolar cells as the CLN3 gene was found to be present in bipolar cells in a CLN3 mouse model as well.

Bipolar cells are hidden away in the middle layer of the retina, a location that is much harder to reach and treat than photoreceptor cells, so Sophia will validate whether recently discovered viruses could be better for this task. This work is very valuable and important not just for CLN6 and CLN3 but in that it provides a platform for future work in gene therapy for the eye in other NCLs.



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