

Batten Disease Newsletter

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



BATTEN DISEASE FAMILY ASSOCIATION

Bringing light to Batten's

Issue 27 Autumn/Winter 2013



Batten Disease Family Association

Registered Charity No. 1084908



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From our Chair



Pauline and James

Dear Members,

Another October, another AGM! And a new venue this year. We are looking forward to the weekend at the RNIB Centre in Coventry. It looks to be a wonderful facility and the Team have lined up an interesting and varied programme of events - for both the adults and the children.

As always, the Team are ever busy as the demands for our services and support continue to grow. A truly exciting development this year is the BioMarin trial and we are pleased and proud to have been closely involved in facilitating meetings between representatives of the company and a number of families to discuss the many aspects that a potential trial would involve.

Since then, we have continued to work with them on the many family-centred aspects required prior to a new treatment going to clinical trial. Please see pages 10 & 11 for more information on this and our other research activities.

Let's be honest, Batten disease is no laughing matter. Over the years I have shed a fair few tears, but what has been a silver lining to the Batten's cloud is the wonderful people I have met along the way and the fun that can still be had. A wonderful example of this was the Walk held in June - amazing weather, good company and just great fun. Then there was Battenfest - another highly successful fundraising and fun event - you can see what I mean on pages 6 & 7!

Your Board of Trustees are determined to continue to grow the BDFA. We must evolve if we are to continue to make a difference. I am pleased to say that we are in the process of welcoming new Trustees with the skills we need to help shape and drive our long term strategy.

We are determined to continue to improve the services we can offer all of our children and young people plus their families and friends and of course continue to support research, as finding a cure to this evil disease is a key objective. Your support is vital so please continue to help us by raising awareness and funds throughout the coming months.

Best wishes,

Pauline

Pauline Docherty, Chair



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



Front cover image Laura and Thomas Bletsoe at Battenfest.

From the Chief Executive



Andrea

It doesn't seem that long since I was writing my piece for our last edition of the BDFA newsletter but we are now into Autumn and what is always a very difficult period of the year for our families and their vulnerable children and young people. We wish that you all stay well throughout the difficult winter months.

Again we have a packed newsletter, but for me one of the key moments was reading Debbie Norris's heartfelt poem about her family's journey with this devastating disease. I know that when this poem was published on our Facebook page, it touched many of you, so thank you Debbie for so bravely putting pen to paper and describing the daily challenges that you all face.

We are delighted to welcome Professor Paul Gissen to our Medical and Scientific Advisory Team. You will see that he brings with him extensive medical and research experience and I know will be a real asset in bringing the necessary services and resources to the families we work with.

As many of you will already know, BioMarin has started their CLN2 Trial with Dr Angela Schulz in Hamburg, Germany. The BDFA has worked alongside BioMarin and the MPS Society to make this trial a reality and to support families who are taking part. You can read more about this in our research section and Dr Ruth Williams will be attending our Family Networking Weekend with members of the BioMarin team to update us on how the London site for the trial is developing.

In this issue, you can also read about Steve Harrison who valiantly cycled from Land's End to John O'Groats to raise awareness and much needed funds for our work. Special mention must go to his helpers in Ashleigh and Alisha Lennon and we have some wonderful photos of all of their hard work on page 5.

The BDFA only succeeds in its work with the contributions of the whole team. This is a particular challenge as our Family Support Officer, Matt, is away from the office at the moment. We wish Matt well and hope he is able to return to work soon. As always, we continue to need skilled people to volunteer for our Board of Trustees. The Board have made some changes to the duration, frequency and location of their meetings to enable more people to attend. If you would like to know more about how the Board oversees the work of the charity please get in touch - your support will be very welcome.

Finally, we are all looking forward to our Family Networking Weekend and AGM which this year is being held at the RNIB Pear's Centre in Coventry. We have a packed programme on education for children with visual impairment, research updates and clinical updates in addition to a fabulous children's programme of circus skills, arts and crafts, and music workshops. I look forward to seeing many of you there and meeting friends old and new.

Best wishes,

Andrea

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

Update from the Fundraising Officer



Nikki

Summer seems to have departed once again and as I type it is pouring with rain. I am not trying to sound like a weather reporter but the seasons have such an effect on fundraising that I am always sorry to see the good weather go.

We had two amazing events during the summer – our annual walk at the end of June closely followed by Battenfest, our first ever music concert thought up and run by our Trustee Ellen Bletsoe and her dedicated helpers. Not to harp on about

the weather but we had two of the hottest most beautiful days for these events.

The Walk saw 130 of us set up camp at Queen Elizabeth Country Park before setting off for a warm-up walk of approximately 3 miles. In the evening we had a BBQ and quiz (won by the Bletsoes). After a good night's sleep we set off on THE walk - 5½ miles to Uppark House for a picnic lunch and then back. We provided a shuttle service for those who were unable to complete both ways although the majority of people managed the full 11 miles.

Watch out for details of next year's Walk appearing on the website as it will be even bigger and better. Special mention must go to Zach and Molly who raised over £3336 in sponsorship.

Our fundraising supporters are extraordinary people, going to such lengths to raise money to help us keep our services running. Without your help the BDFA simply would not exist. Please keep up the amazing work, keep the ideas flowing and help us to reach out to even more families affected by this hideous disease.

Nikki

Nikki Radcliffe, Fundraising Officer
07584 300086 / fundraising@bdfa-uk.org.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 Family Support Officer:
(0800 046 9832 / support@bdfa-uk.org.uk)

Contents

Dear Member.....	2
BDFA Team and Dates for your Diary.....	3
BDFA Team	4
Biking for Battens	5
Battenfest & Annual Walk	6-7
BDFA News	8
International Collaboration.....	9
Research.....	10-11
Our Path to Batten Disease.....	12

Dates for your Diary

OCTOBER 2013

28-31st October 2013 BDFA Siblings and Family Education Event at West of England School

NOVEMBER 2013

21st November 2013 Kidz up North free exhibition for children and young adults with disabilities and special needs, their families, carers and the professionals who support them. For tickets and programme contact 0161 607 8200 email: info@disabledliving.co.uk

JUNE 2014

10th June 2014 PSDL Lab Day

June 2014 (date tbc) BDFA Annual Walk

OCTOBER 2014

22-25th October 2014 NCL 2014, Cordoba, Argentina

18-19th October 2014 BDFA Annual Conference and AGM (venue tbc)



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Update from the Research Officer



Heather

Following on from the excellent lab open day at the RVC, we have co-hosted two more lab open days, at the PSDL Lab. They were a great success and we had very positive feedback from all who attended, both from families and professionals. We hope to extend this to enable families to visit other groups in 2014 and the PSDL will welcome visitors again on 10th June next year. NCL 2014 will be held

in Cordoba, Argentina, and preparations are well underway. Registration will open online in December.

We have received excellent 6-month interim reports from the projects funded at the start of 2013, featured in the last newsletter (Issue 26 Spring/Summer 2013 P18 and 19) – Prof. Hofmann, for her work on Enzyme Replacement Therapy, in a mouse model, for CLN1 disease; Prof. Cooper and Dr. Williams on Cell based system for drug discovery in JNCL; and Prof. Gerst, who has a PhD student looking at the downstream effects of CLN3 in cell based systems. These projects are of 1 year duration and full reports are due in early 2014, and will be available on the BDFA website.

Our programme of co-funding PhD studentships has proved very successful and our congratulations go to Lotta Parvianen (Dr. Brenda Williams) and Thomas Kuhl (Prof. Jon Cooper) from Kings and Kim Wager (Dr. Claire Russell) from the RVC, who have recently been awarded their doctorates. We hope to continue and extend research funding in this way for further studentships in 2014.

Sophia Holthaus is now in her second year of study on a BDFA funded studentship at UCL (Dr. Sara Mole and Prof. Robin Ali) on Gene therapy to treat the visual failure of Batten disease, and gave us all an update on her excellent progress at the UCL Lab Day.

Research funding will continue and we are looking to fund new projects and to develop innovative ways to support Batten disease researchers and to recruit new researchers to the field in the future. Please contact me if you wish to learn more about our research programme or have any research questions.

Best wishes,

Heather

Heather Band, Research Officer
01243 672660 / heatherband@bdfa-uk.org.uk

NCL Resource - A Gateway for Batten disease

<http://www.ucl.ac.uk/ncl/index.shtml>

This site (maintained by Dr Sara Mole) serves as a global gateway for clinicians, families, researchers and those offering professional support, who have an interest in or are affected by Batten disease or who wish to find out more.

Welcome to...



The BDFA are delighted to welcome **Professor Paul Gissen** to their team of Medical and Scientific Advisors. Professor Gissen is a Consultant in Inherited Metabolic Diseases (IMDs) at Great Ormond Street Hospital for Children, London, UK and a Wellcome Trust Senior Research Fellow. Having obtained his medical degree from the University of

Glasgow, Dr Gissen completed his Paediatrics training at Manchester, Sheffield and Birmingham Children's hospitals; including subspecialty training in IMDs.

Paul completed a PhD in human molecular genetics in Prof Eamonn Maher's group at Birmingham University. His research achievements so far include the identification of more than 10 novel genes mutations which are responsible for inherited paediatric disorders. Paul's research group is investigating molecular and cellular basis of intracellular trafficking disorders and developing novel methods for diagnosis and treatment of inherited metabolic disorders (IMDs).

The Batten Disease Family Association welcomes its new patron, Andrew Strauss



The BDFA are delighted to announce that Andrew Strauss has become a BDFA Patron. Andrew Strauss is one of the most respected and highly rated cricketers in the world and he has been involved with the work of the BDFA for a number of years including both the highly successful "Batting for Battens" events.

Strauss has gained international recognition for the way in which he continues to conduct himself both on and off the field. With bat in hand the 32-year-old has forged the reputation of being a brave, resourceful and dependable opener. As a batsman he leads by example, making valuable contributions in tricky situations when his side needs them most. In his entire cricketing career, he has never shirked responsibility, working tirelessly at his game and regularly giving the England team the platform it requires at the start of a Test innings.

Off the field Strauss has done an enormous amount to promote the game too. When dealing with supporters and the media he is always polite, honest and happy to give of his time.

We look forward to working together to raise awareness and continue the fight against Batten disease.



Biking for Batters

My name is Steven Harrison. I am 24 years old and live in Linthorpe, Middlesbrough. In August I cycled from Land's End to John O'Groats which is 874 miles and raised approx. £5,000 for the Batten Disease Family Association.

This ride is undoubtedly something I will vividly remember for the rest of my life. Scotland in particular offered some breathtaking routes, especially on the last day of the journey. It will be a while before I get a chance to ride on roads more rewarding than those.

You see people on the TV or read about them in newspapers, who have reached the summit of Everest or crossed the Arctic Circle on skis. This was my Everest. For me personally, I approached this as my own once-in-a-lifetime challenge with the added bonus of it being for an exceptionally good cause.

For a long time leading up to the ride I was worried that I just wouldn't be able to do it, but in the end one of the most gratifying things I took away from the journey was being able to silence my own biggest critic – me.

It was heartwarming to see just how much support we were receiving as the ride went on. It really felt like we were achieving something special. There were numerous highlights along the way – meeting a lady whose child had died 11 years ago of Batters, having a visit from little Frank's grandparents and his brother and sister – they really lifted my spirits after a day of riding in driving rain and made me realise what it was all about.



Another family who made me realise the same were the Lennons of Lancaster. What a family! Though I was a bit gutted when the girls beat me in a head to head.

With each passing day more and more people were donating to the cause and liking our Facebook page, and by the time we reached John O'Groats I had the impression that we had all the momentum in the world.

A huge thank you to anybody who took time out of their day to check in with how we were getting on. Honestly, getting updates on the activity of the Facebook page helped keep the morale of the group high. It was so humbling at the Wooler campsite when a young girl donated her ice cream money to the charity.

And I wasn't the only one sleeping in campsites for two weeks. I had family and friends who were there in all weathers helping me get to the end. My mam and dad, Aunty Sue and Uncle Dave, Mia and Darren, Paul (PER Productions) and Chris (CDB Production Solutions) all deserve just as much recognition as I do.

'Together we will make a difference'

Steve

Newsflash... Steve has just broken the £5k target!





Remembering...

The BDFA is here to support families at all stages of the Batten's 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.

LSD PATIENT
COLLABORATIVE

genzyme
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*If you hear hooves it might
be a zebra..*

The BDFA, as part of the UK LSD Patient Collaborative Group, is delighted to announce that we have been awarded a Patient Advocacy Leadership (PAL) award by the pharmaceutical company Genzyme in their 2013 award round. Out of 43 applications from 23 countries, 8 awards were made.

We have received this funding for a medical student empowerment project based on the outstanding work completed by Lucy Lavery and her team at Bart's Medical School.

"If you hear hooves it may be a zebra..." Medical Student Empowerment Project <https://www.facebook.com/UkLsdPatientOrganisationCollaborative>

The UK LSD Collaborative – a collaborative of six rare disease patient organizations – seeks to improve diagnosis and treatment of rare diseases in the UK by educating and empowering the next generation of health care providers. Together, the organizations of the UK LSD Collaborative represent the Batten, Gaucher, Fabry, MPS, Niemann-Pick, Pompe and Krabbe disease patient communities.

Through their PAL project, the Collaborative will partner with 5th year medical students from the Bart's London School of Medicine and Dentistry, as well as the student-created London Society for Rare Diseases to support the development of rare disease societies across the London Medical Schools. Other activities include the development of a website, toolkit, and speakers' bureau.

A huge BDFA thanks...



All those who took part in the BDFA Annual Walk at Queen Elizabeth Country Park would like to thank Morris Leslie Plant Hire (Woking) and Karen Gaden for the invaluable loan of a flatbed truck and off-road gator. These were fantastic in keeping our walkers well supplied with water and ferrying those with tired feet. Not to mention the distraction it provided for all the children on the walk!

Educational Visit to Scandinavia



In 2012 I was awarded a BDFE Research Grant and I decided to use this to follow up the links established at NCL 2012 to organise a specialist conference at Statped Midt, with Bengt Elmerskog and Per Fosse. Here they outlined the proposed 3-year international education project that Statped and the parent group in Norway have

been working on, an international collaboration on the sharing of ideas and expertise to improve the education of children and young people affected by NCL. Tracy De Bernhardt Dunkin (WESC Foundation) and Glenys Critchley (National Blind Children's Society) also attended. Sarah Holton (RNIB) and Cari Mannion (Royal Blind School) were not able to travel to Norway but are also part of the collaboration. I have been asked to be a member of the project steering group.

The experts from Statped told us about their innovative education programmes, namely 'Sarepta', the computer program that has been developed over many years to help young people with JNCL. Tracy De Bernhardt Dunkin presented on the exciting developments in neuroscience and visual impairment, and we discussed the implications of this for NCL. It was a great opportunity to look at the excellent facilities and services offered at Statped Midt and talk to the staff who work with young people with JNCL.

Next, to Denmark to meet with the Danish family group and specialists there where we talked about the great success of NCL 2012 and how valuable it was to those who attended. Danish families are very keen to meet up with UK families again and to share their experiences. Key issues, such as the right educational support, effective transition to adult life and opportunities to meet other young people and families are important to both groups.

I watched a video of the winter residential visit that the young people with JNCL made to the Norwegian Red Cross Centre. It was wonderful to see them enjoying themselves together in the snow and ice, regardless of their present abilities.

In Kalundborg, Denmark, I met up with Anne Hagedorn Harmann, from the Social Work team, and Janne Trebbien, a teacher at the school for visually impaired pupils. It was interesting to see a contrasting system to our own and we all feel the international NCL education project can provide opportunities to learn from each other. When supporting young people and families affected by such a low incidence condition, professionals can sometimes feel isolated and opportunities to share and develop their expertise are invaluable. There are around 25 young people affected by JNCL in both Norway and Denmark compared to the much larger numbers in the UK but together we can all move our knowledge and understanding forward.

On to Oslo and a meeting with Trine Paus (chairperson of the Norwegian family group) and Svein Rokne, who are also involved with the work at Statped Midt. This close co-operation, so evident all over Scandinavia, is of vital importance and really adds to the support on offer to children and families. Plans were made for individual families to be 'linked up' and for reciprocal visits by groups of families.

All the young people in Norway and Denmark with Batten disease have the juvenile form. There have been a very small number of children with the Late Infantile form but there are none at present. Although the international project will focus on JNCL, there is also a clear intention to include other forms of NCL as well as learning from other neurodegenerative diseases.

My visits showed that there are many different ways that services for young people and families affected by NCL can be organised and funded. However, wherever I went the dedication and commitment of the professionals and families I met shone through, together with their determination to make sure that our young people have the very best support.

Barbara Cole
BDFE Education Advisor

Barbara's trip was made possible under the BDFE small grant awards made to our Advisors in recognition of their work at NCL 2012. Full reports of all these awards can be found on our website.

BDSRA Director's visit to the UK



It is very important to us in the BDFE, that we work collaboratively with our friends and colleagues in the Batten disease community across the world. Building on our

long history of working closely with our US colleagues, it was with great pleasure that we were able to welcome the BDSRA Director Margie Frazier to London where she took time out of her holiday with family to meet with us and members of the Batten disease scientific and clinical community at UCL.

Attendees included Andrea West (BDFE CEO) Margie Frazier (BDSRA Director), Pauline Docherty (BDFE Chair of Trustees), Heather Band (BDFE Research Officer), Nikki Radcliffe (BDFE Fundraiser), Dr Sara Mole (UCL), Dr Claire Russell (RVC), Dr Ruth Williams (Consultant Paediatric Neurologist), Dr Brenda Williams (Kings College), Professor Jon Cooper (Kings College), Barbara Cole (BDFE Educational Advisor) and Nick Lench (Director, NE Thames Regional Genetics Service).

We all know how important it is for us to share resources and work in the field of research as well as across all the areas that affect the lives of children and young people living with Batten disease. This enables us to make best use of our scant resources and allows us to share good practice across important areas such as social care and education.

Margie writes: "While visiting family in the UK, it was great to see my other family, our close collaborators in Batten research. Sara Mole and Andrea West organized a fantastic lunch at University College London where we greeted those who work so hard every day for Batten families around the world."

Building on our strong research funding collaboration and close working relationships we look forward to a future of working together to make a difference to families living with this disease and make our aims of treatments and ultimately a cure, a reality.

Andrea West, BDFE CEO

BioMarin Clinical Development Programme for CLN2 Late Infantile Batten disease

BIOMARIN

PRESS RELEASE:

BioMarin Pharmaceutical Inc. announced on Sept. 23 2013 that it has dosed the first patient in the Phase 1/2 trial for BMN 190, a recombinant human tripeptidyl peptidase 1 (rhTPP1) for the treatment of patients with neuronal ceroid lipofuscinosis type 2 (NCL-2), a form of Batten disease. This is the first time that a patient with Batten disease has been treated with an enzyme replacement therapy in a clinical trial setting.

“This program is representative of the company’s core competency of developing life-altering enzyme replacement therapies for serious unmet medical needs,” said Hank Fuchs, M.D., Executive Vice President and Chief Medical Officer of BioMarin. “We are inspired and motivated by the patient and physician community and encouraged by the pharmacological activity demonstrated in preclinical models. We hope to leverage our expertise in enzyme replacement therapy development to deliver a viable treatment option to patients with this form of Batten disease.”

“The completion of dosing of the first patient in the trial of BMN190 marks the beginning of an important journey. This neurodegenerative disease of childhood is devastating for patients and families. While we hope to make a large difference in their outcome, we are moved and grateful for the support of affected families worldwide,” said Angela Schulz, M.D. Ph.D., Children’s Hospital, University Medical Center Hamburg-Eppendorf.

“Clinical trials are the only means to ascertain whether the promise of this new therapeutic approach will be fulfilled. Without the families’ selfless dedication to the important principles of science, this would not be possible.”

The Phase 1/2 study is an open-label, dose-escalation study in patients with NCL-2. The primary objectives are to evaluate the safety and tolerability of BMN 190 and to evaluate effectiveness using an NCL-2-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 NCL-2 natural history after 48 weeks of treatment and to characterize pharmacokinetics and immunogenicity. The study will enroll approximately 22 subjects at up to ten clinical sites for a treatment duration of 48 weeks.

BioMarin have indicated to the BDFA that they anticipate the Evelina Children’s Hospital in London will be the second clinical site, with patients enrolled here by the end of 2013 or early in 2014.

We must reiterate that this trial is only relevant to those affected by CLN2 late infantile disease and who meet the eligibility criteria. It will not be applicable to other forms of NCL, such as CLN5 or CLN8.

Please contact Andrea West if there is anything you would like to discuss about this.

andreawest@bdfa-uk.org.uk 01252 416110

PSDL Lab Open Day, Institute of Psychiatry, King’s College London, June 2013



Ellen Bletsoe

Each year the Paediatric Storage Disorders Lab (PSDL) opens its doors and with the BDFA invites family members, friends, carers and health professionals to come and see what we do.

The main aim of the day is to get everyone together, to demystify the science we’re doing and give an update of the progress we’ve made so far.

We would like to think this helps all those who come but it is also very important for everyone who works in the lab. The chance to talk with people whose everyday lives are touched by Batten disease is invaluable.

We started with talks from all the lab members on their individual projects and then toured the labs. We even had our visitors mounting brain sections on slides, something that’s not as easy as it looks!

The whole day was designed to maximise the time spent together, to promote interactions between scientists and families, to talk and ask questions.

We all enjoyed it very much and look forward to welcoming anyone who is interested in Batten disease at next year’s PSDL Open Day.

Professor Jon Cooper

Dr. Sara Mole hosts the first UCL-BDFA Lab Day at the MRC Laboratory for Molecular Cell Biology (LMCB)

The concept for my lab day was a little different; we wanted to show families what we do at the LMCB, and also to bring together scientists who work on different aspects of Batten disease at UCL. So, thirteen of my colleagues were persuaded to give up their Saturday to come and talk about their work at UCL.

Support staff at the LMCB were extremely helpful in the week before, covering health and safety responsibilities, ordering poster boards and lunch, arranging the venue and even offering to help with the lab demonstrations.

The day started with an introduction from Heather Band, BDFA Research Officer about the BDFA and how they support and fund research. This was followed by four presentations covering topics from disease to diagnosis, including how new genes are identified. We then had a deeply moving talk from Rachel Thompson about the rollercoaster journey that has been their family's experience of Batten disease over the last few years.

Over lunch there was time to network, followed by the afternoon session – diagnosis to therapy, thinking about this in relation to gene therapy for the eye, (a BDFA funded project), the body, ways of finding new drugs and using yeast as a model organism. There were 6 posters, some from recent scientific conferences, others written specially for the day.

To finish, we provided demonstrations in four different labs so visitors had an opportunity to don a lab coat and try their hand at electron microscopy, tissue culture, high-throughput sequencing watching “the robot” make a colourful Union Jack pattern, using a microscope to look at cells growing in culture and manipulating yeast.

“I found it very informative and interesting. The Lab tours were the highlight for me. It's been good to meet the scientists and see first hand the work they are doing. I would like to thank you all for making it such an enjoyable day.”

The UCL scientists all enjoyed and appreciated the day, finding it very inspiring and giving them a chance to chat together as well – there were even some new collaborative experiments discussed which was not the aim of the day but an unexpected and fantastic outcome!

“Really enjoyed the day. Think it switched on a science gene in my child who had lots of questions on the way home and was really fascinated by EM and yeast! Families, as usual, were amazing.”

So all in all, a day that exceeded expectations, certainly very worthwhile, and very much appreciated by the UCL scientists.

Dr Sara Mole



Rose Heales, Ben Marlow and Alice Crowley





Jordan

Our Path to Batten Disease

We very rarely comment,
We don't know what to say.
We find it hard to communicate
How we feel each day.
This Batten disease has floored us,
It's hung us out to dry,
It's hit us like a thunderbolt,
"Why", we cry, "oh why?"

It hit us like a thunderbolt –
That isn't strictly true.
For oh so long inside our hearts
There was a fear that grew.
He lost his sight. "Oh well", we said,
"We'll make another plan."
Lots of people lose their sight,
We will help him all we can".

The seizures were the next to come,
So helpless did we feel.
"But we can deal with this", we said,
"We're strong, it's no big deal".
But then there came the tantrums –
Little things would make him mad.
We knew he couldn't help it,
We knew he wasn't bad.

The "professionals" they thought differently –
"Your parental skills are weak".
They judged, criticised all the time,
With meetings every week.
But deep inside alarm bells
Were tinkling away.
"There's something more that's going on"
Our inner voice would say.

Then we got the diagnosis
That no-one wants to hear.
"It's Batten disease", they told us.
Our hearts were filled with fear.
We have had to make decisions
Which have caused our hearts to break.
But we'll always do our best for him,
Whatever that may take.

Our lives will never be the same.
It's different now, you see.
But our love for him will never change
Wherever he may be.
We didn't choose this journey,
But travel it we must.
We know now that we're not alone,
There are others we can trust.
Those who understand
And know the pathway that we walk.
All at different stages
But there to listen or to talk.

We cannot plan a future
We can only live each day.
Some days are good, some days are bad,
Some are just okay.
But there is one thing that this disease
Can never take away –
The love we'll always have for him,
Forever and a day.

DEBBIE NORRIS