Batten Disease Newsletter

ISSUE 30 SPRING/SUMMER 2015

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

Batten Disease Family Association Registered Charity No. 1084908

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The Old Library, 4 Boundary Road, Farnborough, Hants. GU14 6SF

Dear Member

From our Chief Executive


Andrea

Dear members, colleagues and friends,

I am delighted, once again, to introduce an edition of the BDFA newsletter that showcases the outstanding work of our staff, volunteers and supporters in our fight against this devastating set of diseases.

The last six months have seen our staff team grow to include two new fundraising administrators, Gaynor Heeson and Kate Shefford. Both Kate and Gaynor are looking forward to supporting all of you who work so incredibly hard to fundraise for all aspects of our work. On page 3 both talk about their backgrounds and what brought them to working with the BDFA. They would be very happy if you would like to get in touch with your fundraising ideas on **fundraising@bdfa-uk.org.uk** or **07876 682589**.

This is also a great opportunity to introduce Harriet Lunnemann who is our new Support and Advocacy Partner. Harriet is a qualified social worker and brings a wealth of experience to the role and to the families she works with. She also introduces herself on page 3. Over the next couple of months Harriet will be introducing herself to all of you to identify how the BDFA can support you, your family and the professionals around you. To get in touch with Harriet please call our Freephone Helpline **0800 046 9832** or email **support@bdfa-uk.org.uk** She has also updated the support section of the BDFA website and will be regularly adding materials to it and posting on social media. By following us on Facebook and Twitter you can keep up to date with our work and interesting news.

On page 8 you can read about the appointment at Great Ormond Street Hospital of the first UK Batten disease Clinical Nurse Specialist, Katie Hanson. We are extremely proud that the BDFA has funded this post to meet the needs of families living with this disease and to bring our goal of a UK national Batten disease service closer. Over the next few weeks and months Katie will be identifying how she can work with professionals and families across the country to provide the best level of service.

January has also seen the BDFA move into offices for the first time in its history. In 1998 a group of parents set up the BDFA to ensure that no family would face their journey with Batten disease alone. That aim is still central to who we are as an organisation and the move into offices will enable us to grow and continue to deliver essential services and support to families. Our offices are in Farnborough and if you are ever in the area we would be delighted to see you.

Thank you to everyone who completed the survey looking at our work and what the BDFA means to each of you. It is very important to us that the voices of our members and supporters inform what we do, so please get in touch if there is something you feel we do well, there is something we are missing or something we could do better. Every new edition of the BDFA newsletter leaves me very proud of what we as a group can and will achieve.

Best wishes,

Andrea

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

From our Chair


Mike

It has taken many months/years of hard work and perseverance but it really is all coming together at the same time which makes this newsletter the most exciting in many years. Andrea will update you more fully but with new staff members, our new office, the CNS position filled, the BioMarin clinical trials moving ahead and a new fundraising and strategic review under construction we have so many good things finally in place and more in the pipeline.

I would like to touch on our staff and the office. The new office may only be a roof over our heads but it is our home and we should all be very proud that we now have a place we can call home. I know we will all feel the benefits of having our fantastic staff members under one roof. As our charity has grown, managing and staying in control of efficient communication has been a constant concern and I know Andrea and her team will get so much more out of their work by being able to see each other and very importantly feed off each other. This is a significant step forward and one that I'm delighted we have been bold enough to make.

A lot is contained within this newsletter already about Kate, Gaynor, Harriet and Katie, but I would like to welcome them personally and I wish them all the very best in their respective roles. I know that if they show the same commitment to serve our members that our longer term staff members have shown – who consistently go above and beyond – we will all be the better for having them with us. Our team is strengthening all the time and I know Andrea feels we now have a full strength team on the pitch.

Finally, I would like to thank two groups. Firstly, my fellow trustees. I have done this in every newsletter and I make no apology for repeating myself, but your work is greatly appreciated. I know you all have incredibly busy lives and the way you keep finding the time to give to the organisation is a great credit to you and your time management skills. The wonderful thing is that I know you do it because you believe we can eventually beat this disease but in the meantime you really want to make the daily challenges that face affected children, parents, siblings etc easier to deal with.

Secondly, I wish to thank our fundraisers. As I said at the AGM, fundraising has been my real focus over the last five years and without everyone's fundraising efforts we wouldn't be in an office, we wouldn't be able to employ the team we do, and most importantly we wouldn't be able to fund the research we do. You are vital to our ongoing success so I congratulate and encourage you to keep up the amazing work.

Best wishes,

Mike

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

Front cover image: BDFA staff team, copyright Farnborough News & Mail

Welcome to our New Staff Members

HARRIET LUNNEMANN

BDFA Support and Advocacy Partner

Harriet joined the BDFA in November 2014. She is a qualified social worker who has worked with children with profound and multiple learning disabilities as well as children with complex health needs. She has also worked in a school for children with special educational needs and has been a respite carer.

She is currently a foster carer and in her spare time she runs a project for disabled children in Uganda. She can be contacted on **07876 712553** or the Freephone Helpline **0800 046 9832**.

GAYNOR HEESON

Part-time Fundraising Administrator

Gaynor also joined the BDFA in November 2014 following a career break to raise her twin girls. She has a background in administration and event planning as well as 18 years' experience as a PA to senior level management both in charitable organizations and large blue chip companies.

She has travelled extensively and experienced many different cultures and lifestyles and is looking forward to using her past experience to proactively support all our hardworking fundraisers and to encourage anyone who would like to have a go at fundraising in the future.

KATE SHEFFORD

Part-time Fundraising Administrator

Kate joined the BDFA in November 2014 to job-share with Gaynor. Her educational background is scientific and her work life has been mainly in the business world, with roles in HR Management and Business Administration. During a career break to raise her children, she has been involved in fundraising as the school PTA Chair as well as through her children's sporting activities.

She is passionate about most sports and looks forward to using her skills to support all our dedicated fundraisers in their events.

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

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Diary Dates 2015

4 June 2015	Kidz South, Rivermead Leisure Complex, Reading One 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 this event, call 0161 607 8200
9-11 June 2015	British Society For Gene & Cell Therapy Annual Conference, Glasgow
15-16 June 2015	European Education Project Meeting, London
17 June 2015	Batten Disease International Alliance Meeting, London
18 June 2015	PSDL Lab Day at King's College London
20-21 June 2015	Tenth BDFA Annual Walk (venue tbc)
17 September 2015	Kidz Scotland, Royal Highland Exhibition Centre, Edinburgh
17-18 October 2015	BDFA AGM and Annual Conference, Coventry

Support & Advocacy

I am very excited to be in post as the BDFA's first full-time Support & Advocacy Partner. It is important to us at the BDFA to have a holistic approach and to consider all the needs of a child or young person living with Batten disease as well as their families and the professionals who work with them.

We aim to support families from diagnosis right the way through their journey with Batten disease and to ensure that you all have access to the right support and information to enable you and your family to have the best quality of life possible.

Here are just a few ways in which the BDFA can provide support:

- We can give advice and support to all members of the family; over the phone, by email or in person at meetings you feel it would be appropriate for us to attend
- We can provide leaflets and information which you can give to the professionals working with your child so that they can further understand Batten disease
- We can provide educational support and training for schools
- We work with Batten disease specialists and can liaise with them about your situation
- We are also able to provide information on research and trials taking place both in the UK and worldwide
- We can connect families so that they can support one another
- We can connect the professionals working with you with other professionals enabling them to share their experience and knowledge

- We provide workshops for families and workshops for professionals
- We can write letters on your behalf to support grant applications or letters to professionals working with your child
- We can give guidance on specialist equipment and benefits you may be entitled to
- We can also provide small grants to families of children or young people who are living or have lived with Batten disease

We would love to hear from you if you feel we can support you in any of these ways.

For support or advice, please contact me either by email: **support @bdfa-uk.org.uk** or by phone: **0800 046 9832.**

Harriet Lunneman
BDFA Support & Advocacy Partner

NEW Information Leaflets

We have recently published some new information leaflets which we hope you will find useful.

They are "Financial Support – Benefits & Tax Credits", "Equipment" and "Supporting the educational needs of children who have a diagnosis of NCL (Batten disease)."

They are all available to download from our website or can be obtained in hard copy by calling our office on **01252 416323**.

BDFA Small Grant Scheme

Marshall's P-pod bought using a BDFA Small Grant

The BDFA appreciates that families are often faced with financial challenges when caring for someone with Batten disease. Whilst there may be various options available, the BDFA aims to provide assistance to help families cope with these circumstances and therefore operates a "Small Grants to Individuals Scheme". This is designed – with a streamlined application process – to be able to provide a rapid practical response to difficult situations.

The BDFA can offer a payment of up to £500 to families of children or young people with a diagnosis of an NCL. The grant can be used in many different ways.

Families can often find themselves washing clothes and bed sheets far more than is usual and this can take its toll on the household washing machine. The grant might be used to replace a washing machine or for other household appliances such as ovens and tumble dryers.

Living with a diagnosis of NCL can also mean unplanned stays in hospital for your child or young person. You can apply to the Small Grant Scheme for help with the associated transport costs, or for unusually high heating or water bills.

Children or young people with a diagnosis of NCL often have access to a stimulating sensory environment and specialised equipment at school or in respite or day care, but very few have these facilities at home. Our funding can help provide sensory equipment at home.

We are also able to help fund other much needed equipment which may not be available from your local health or social services authorities. To discuss a possible application for a small grant, please contact Harriet on **07879 712553** or **0800 046 9832** or by email: **support@bdfa-uk.org.uk**

BDFA Office Launch

On Friday 27th February, to coincide with Rare Disease Day, we held our new office launch and official opening in the company of the deputy Mayor for Rushmoor, Councillor Martin Tennant and MP for Aldershot, Sir Gerald Howarth. This was the culmination of many months' planning and negotiation to find suitable premises.

Our success as a charity and growth of the staff team means that office space has become necessary to successfully and efficiently work together to achieve the Charity's aims. Our new home at The Old Library in Farnborough is proving just the place to do this.

Our event was featured in the local press and we were joined by local businesses and volunteering coordinators, as well as Trustees, some of our Battens families, scientists and volunteers. Our guests were given information on our rare disease charity to take away with them and had drinks and a buffet lunch with us as well.

Sir Gerald Howarth MP welcomed us to the area in a speech and was particularly interested in our charity as he had a family connection with a rare disease in the days when far less support was available to families.

With our new office base, we hope to spread our message and raise awareness and fundraising interest in the local community as well as continuing to support Battens families across the whole of the country.

Lucy Roose
Office Manager

Supporting the Educational Needs of Children who have a Diagnosis of NCL (Batten disease)


Barbara Cole

The BDFA's Education leaflet "Supporting the educational needs of children who have a diagnosis of NCL (Batten disease)" was launched in October 2014 at Linden Lodge School as part of an information and support day for professionals. Many families affected by the various forms of Neuronal Ceroid Lipofuscinoses (NCLs) will be faced with numerous important decisions about education for their child. The BDFA aims to help them get the best possible provision, as well as advising and supporting the professionals working with them.

We are able to provide unique support through our BDFA Education Advisor, a qualified teacher of the visually impaired who has worked with many children and young people diagnosed with the various forms of NCL.

The leaflet offers information about support available for individuals who have Special Educational Needs due to a diagnosis of NCL as well as providing general advice for families who wish to ensure that their child receives the best education possible. The document can be downloaded from the BDFA website and the 8-page leaflet is available on request from the BDFA office. We recommend that families share this document with all the professionals from education, health, social care and other services working with them.

The BDFA recognises that the different forms of the disease processes impact on children and young people in varying ways and at different stages of their life. Each child and family's journey with NCL will differ in some ways due to many varying factors (e.g. the specific genetic diagnosis, age of onset).

The leaflet gives details of the recent changes in SEN law and of the assessment process leading to the drawing up of an Education, Health and Social Care Plan that describes the child's education, health and social care needs and the provision needed to meet those needs. The EHC Plan replaces the Statement of Special Educational Needs. Some children and young people already have a Statement of SEN and the leaflet describes the process by which the Statement will be converted to an EHC Plan before 2017. It also gives details of the support that is available for parents and carers.

Issues around school placement are also covered in the leaflet. Mainstream and special school placements are discussed together with education at home. The importance of a well-supported transition process when there needs to be a change of placement is highlighted. There are sections on the importance of visual impairment support, therapy input, specialist equipment and support for maintaining communication skills. The specific educational issues arising from each form of the disease are also discussed.

The leaflet will be revised and updated regularly and further leaflets are planned focusing on useful strategies, equipment and additional sources of support. The BDFA Education Advisor can be approached by any party seeking advice, information or assistance with the educational needs of any child or young person diagnosed with NCL.

Contact us on **0800 046 9832** or email: **education@bdfa-uk.org.uk** or **support@bdfa-uk.org.uk**

Barbara Cole
BDFA Education Advisor

Advice and information can also be obtained from the following organisations:

BlindChildrenUK (www.blindchildrenuk.org) offers expert independent advice to help make sure that blind children get the best from their education. They have a range of services including education support and access technology and can be contacted on **0800 781 1444** or **services@blindchildrenuk.org**

SOS!SEN (The independent helpline for special educational needs) offers a free, friendly, independent and confidential telephone helpline for parents and others looking for information and advice on special educational needs. They have a term-time helpline **(0208 538 3731)** and their website is **www.sossen.org.uk**

IPSEA (Independent Parental Special Education Advice) has trained volunteers who give free legally-based independent advice and support in England to help get the right education for children and young people with special educational needs/disability. Their website is **www.ipsea.org.uk** where details of their services and ways to contact them can be found.

THE ROYAL BLIND SCHOOL

Cari Mannion, Depute Headteacher of the Royal Blind School writes…

The Royal Blind School is based in the heart of Morningside in south west Edinburgh. Links to the railway station, bus station, airport and the city bypass are very good. Morningside is a thriving community with a wide range of facilities such as a library, supermarkets, bookshops, independent retailers, banks and, of course, coffee shops. The School is a grant aided independent specialist school for children and young people with a significant visual impairment or with a visual impairment and additional support needs. The needs of learners may vary from those on the autistic spectrum to those with complex needs. The School prides itself on its role in supporting children and young people with JNCL.

Over the past few years the School and the BDFA have forged links to ensure both parents and children are given the attention and support they deserve. As with any diagnosis that has a major impact on families, *Getting It Right for Every Child* is vital. We also work with the Safe, Healthy, Achieving, Nurtured, Active Respected, Responsible and Included indicators. These ensure that well-being in its widest sense is at the forefront of our

thinking when planning for our pupils. As a staff we all believe that learning is a right for all pupils and that learning should be an enjoyable experience!

The Scottish Curriculum for Excellence is inclusive and covers a range of experiences and outcomes from Early Years to the end of the 3rd Year of secondary education. We provide the full range of subjects including P.E., Outdoor Education, drama, music, computing and CDT. In our grounds we have a sensory garden, swings (popular with all ages!) and a multi-use games area. There is also a hydrotherapy pool and a soft play area and in the Spring we will have a traverse wall too. Classrooms are fitted with state of the art tracking hoists.

When a child diagnosed with JNCL joins our school, we work closely with parents and staff from the previous school to ensure a smooth transition. An individualised education programme is designed which is tailored to meet the needs of that child. There is a focus on independent living skills and communication e.g. braille and assistive technology. We work particularly on expressive language, motor skills and memory. From the 4th to 6th Year, pupils are in the Senior Phase and can opt to choose a range of subjects from the whole curriculum that are of particular interest to them as well as continuing to develop their independence skills as far as possible.

All pupils with JNCL who have attended or are attending the School join a class suited to their age, stage and ability. All classes are small with 4-6 pupils and a high staff/pupil ratio. We have Habilitation Specialists as well as a nursing team and a range of therapies including speech and language therapy, occupational therapy and physiotherapy.

The School is involved in an exciting European Erasmus+ project with several partners - from Norway, Germany, England, Finland and the BDFA as well. The project is looking to establish a knowledge base about all approaches to learning for children and young people with JNCL to ensure the highest quality of education and educational practice across Europe. The group will report during 2017.

The School has an annual Friends and Families Day as well as Parents' Consultation Day and a Parents' Consultative Group. We welcome parents into the School whenever they feel the need to visit. We also have a parents' flat for those who have to travel some distance. Throughout their child's time at the School we strive to include parents in decisions about their child and to support them. A key aspect of this is that we recognise we are working with and for that child, not JNCL. And the best news we hear any pupil tell their parents is 'I love being at The Royal Blind School!'

For further information about The Royal Blind School, please visit **www.royalblind.org/education** or telephone **0131 446 3120**. Alternatively please contact our Education Advisor, Barbara Cole (**education@bdfa-uk.org.uk**) who will be able to advise on your individual situation.

Clinical Nurse Specialist at Great Ormond Street Hospital

"Here at Great Ormond Street Children's Hospital we are delighted to be working so closely with the Batten Disease Family Association to realise the first Clinical Nurse Specialist post dedicated to Batten disease.

It is a unique experience to be developing this new role and supporting the multi-professional teams in providing a holistic care pathway for these children and their families.

The ambition to use the post to have a national reach to centres where children receive much of their health care sets this position apart from other nurse specialist posts.

We are enthusiastic and keen to develop the post to best meet the needs of families ahead of working in some of the traditional ways that may have been employed previously.

We look forward to updating you after the post holder has commenced in early March."

Siobhan Lalor-McTague
Head of Nursing Neurosciences
Great Ormond Street Hospital

Harriet Lunnemann welcomes Katie Hanson (left) to the BDFA offices

Hello, my name is Katie Hanson and I start work as the Batten Disease Clinical Nurse Specialist in March 2015. I grew up in Yorkshire and attended Harrogate Grammar School. I had decided in the sixth form that I wanted to be a paediatric nurse.

Before I attended university to study to be a nurse I took a gap year, travelling and working in Australia. I have several hobbies in my spare time which include horse riding, playing the piano, musicals and long walks.

After completing my honours degree in Children's Nursing at Leeds University, I was given the opportunity to work on the Neurosciences ward at Leeds General Infirmary where I stayed for nearly four years. I was keen to increase my knowledge within the speciality of neurology and neurosurgery, so I moved to London and was privileged enough to get a job as a senior staff nurse on Koala Ward at Great Ormond Street Hospital, which is also a Neurosciences ward.

My aim for this new role as the Batten disease Clinical Nurse Specialist, will be to create a new service that children living with Batten disease and their entire family can gain from.

I will be helping to educate professionals and the wider community about this rare disease, supporting families from the beginning of the diagnosis and throughout their journey and also finding new ways to communicate with families about the different needs of their child, and how we can do this effectively as a team of healthcare professionals.

As the first Batten disease nurse, I will be reaching out to the families and those affected by Batten disease to find out what they identify as their specialist nursing needs and what vision they have for my new role.

I feel so honoured to be appointed in this role and I am excited for the opportunity and challenges that lie ahead. I look forward to meeting you all once I start in post and also to working alongside the fantastic teams that support Batten disease across the country.

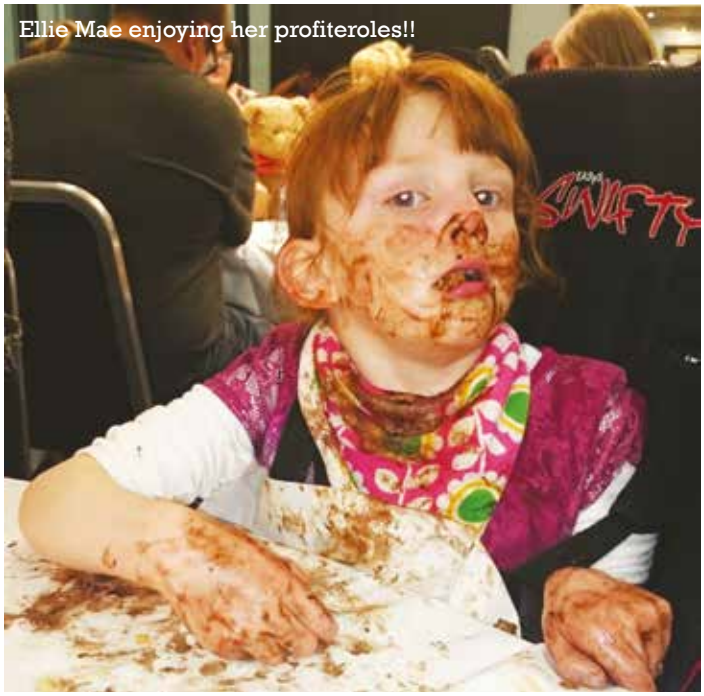
Katie Hanson
Batten Disease Clinical Nurse Specialist
Great Ormond Street Hospital

BDFA Family Conference, November 2014

Last November we had the opportunity to attend the BDFA family conference. It was our first time as our childrens' diagnosis was still fairly new. As soon as I heard about the conference, I was determined to go and meet other families living with Battens and also hopefully learn more about the disease and how to look after the children as their needs change in the future.

It was nerve-wracking walking into the conference on the first day as in some ways it was a room full of strangers but Andrea came straight up to welcome us and introduce herself which put us at ease. Soon we recognised families from Facebook and it was fantastic to meet the children we felt we already knew.

The conference had different talks and seminars and we could choose the ones most relevant to us. I found the talks on current research and those with practical ideas for caring for the children the most helpful.



Ellie Mae enjoying her profiteroles!!

There were lots of experts present including Dr Sara Mole, Professor Jon Cooper and Barbara Cole, and there were great opportunities to hear them speak and / or ask questions. There were also seminars run by charities such as Seeability who run Heather House.

Meanwhile we could settle the children into their activities which were run by Lizzie Spyrou and a team of volunteers from Heather House. There was 1:1 support for both our children and many exciting things for them to do.

Ellie Mae got stuck in straight away, getting messy and creative. She also loved the drums and shakers in the music session. Caleb found a room devoted to DVDs including lots of his favourites so he was in his element. He especially enjoyed meeting the dog from the Pets As Therapy charity. The centre itself was accessible with great facilities making it stress free for us parents!

There was plenty of time to talk to other families and BDFA staff, and to share experiences and learn new ways of coping, many of which have been useful since. There were stands from different charities with useful information and time to ask them questions.

In the evening we had a meal and quiz at the hotel, which allowed us to get to know more people and share some laughs. The kids enjoyed time with their new friends.

We had a great time and even though it was a big step and in some ways scary, I am so glad we went. It helped us to feel we are not alone, but have a team supporting us as we try and look after the children and make sure they have the best possible quality of life.

It was humbling to meet people who give so much of their lives to those affected by Battens, either in seeking to find a treatment or by supporting the families.

Sometimes the journey we are on feels overwhelming and impossible, but making friends with others at different stages of the journey has been an immeasurable help. We are grateful to the BDFA for organising the conference and to everyone who contributed.

Lynsey Brownnutt
parent

Family Conference 2014

Family Conference 2014

Family Story

In March 2009 Harry was born – perfect. When Harry had his Day 2 check at the hospital they noticed a dimple at the bottom of his back. We were taken aside and told to prepare ourselves as Harry might have Spina Bifida Occulta. This was a big shock and you automatically think the worst.

Between that day and when Harry was about 18 months old we attended quite a few hospital appointments. Harry was weak on his right side but we were told this shouldn't cause any major problems. At 21 months, and much to our surprise, a few days before Christmas Harry got his balance and took his first steps. This was the best Christmas present imaginable.

Everybody was thrilled and he was signed off from the hospitals with the explanation he may have some delays but there should be no serious issues. Harry started nursery and went from strength to strength. He was a little monkey and would climb whenever given the opportunity, whether it be over stair gates, fences or falling off the edges of chairs. He was always on the go and getting into mischief and was extremely cheeky. If he was doing something he knew he shouldn't, you could always tell as you could hear a cheeky giggle coming from him.

When Harry got to about three and a half years old, we noticed he was unable to do some of the things his peers could, and as the time was passing the gap was increasing. He was still not a strong walker but we thought that was due to late walking and his fine motor skills were quite poor as well. His speech was not fluent and he could only say 3-5 words.

We attended a paediatrician appointment and Harry was diagnosed with global developmental delay and echolalia. We were told that as he was delayed in several areas an underlying condition could be the cause. We were relieved when genetic testing came back clear.

Just after Harry's fourth birthday it was suggested that Harry's eyes were checked. He was prescribed glasses for long-sightedness which gave us hope that things would improve.

Just before the summer holidays I was called in by Harry's nursery teacher. She expressed her concern that Harry was struggling to recognise his peers - even his best friend standing in front of him. Over the summer holidays Harry's sight deteriorated significantly. He had to feel people's hands to recognise them; even his Nan who he had seen every day since birth. He also spent a lot of time staring into space.

When Harry returned to school in September he was in Year R and my stomach sank at the door as I knew this wasn't the right environment for him. We all found this very difficult. His older brother Joshua was asked to help Harry in school a lot as he was the only one who could look after him at lunch time and calm him when he got distressed.

Harry then saw an ophthalmologist who found he had tunnel vision, particularly in the right eye. This came as a relief as it might explain why he had difficulties in various areas and we could put things in place to help him. He was referred to an

eye specialist at Great Ormond Street to see if they could help further.

A few weeks before Harry's fifth birthday Great Ormond Street issued him with a visual impairment certificate. Extra tests showed he had retinal dystrophy and we thought we had our answer to the problems. We believed this was our key to helping Harry and, with no appointments for a few months, thought we could start his Statement process and get him the support he needed. Little did we know that the following month would bring a letter that would change our lives forever.

On opening a letter from Great Ormond Street on Easter weekend 2014, one sentence stood out: "We believe Harry could have a Batten type disease". The first thing we did was google this, while our beautiful children sat on the floor in front of us and I'll just say that the results that came up on the screen were not very nice. Heart wrenching.

The next couple of months were a roller coaster of emotions as tests were rushed through. Harry had an EEG and to our horror we discovered he was having constant epileptic seizures although his body wasn't physically showing it. He was immediately put on medication which helped straight away.

On 30th July 2014 we had a message from Great Ormond Street asking to see us without Harry the next day – we instantly knew this was it and they were going to tell us what was causing these problems.

I can remember the day as if it was yesterday. They had found a variant of Batten disease and Harry is one of three children in the UK to have it. The feeling of sitting across from a consultant

telling us to go and enjoy our child while we can will never leave me. Life was never going to be the same from that moment on.

We now know how precious time is. Making memories and having fun is a lot more important than the trivial worries we used to have. This year we are concentrating on taking Harry and our other son Joshua on a few holidays while Harry is still quite well.

We know the journey that we face ahead and we plan to make it as fun and enjoyable as possible. We will make the most of every day as it comes. There will be bad days and there WILL be more GOOD Days. Words cannot come close to how we feel.

We love our boys to the moon and back.

Graham, Laura, Joshua & Harry Rolph-Wills x x x x

Boris the Bear

You can follow the adventures of Boris the Bear on Laura's Facebook page as his adventures raise awareness of Batten disease.

Support Resources

The Daisy Garland

The Daisy Garland is a family-run, national registered children's charity set up in memory of our daughter Daisy who tragically died in her sleep from SUDEP (sudden unexpected death in epilepsy patients) when she was 6 years old.

We provide active help and support for children suffering from difficult to control epilepsy and their families/carers. We do this in a number of ways:

- **Providing grants for night-time breathing (SATs/ Epilepsy) monitors for use in the home.** These monitors help prevent SUDEP by alerting parents/ carers to night-time seizures. Families are invited to contact karen.daisygarland.org.uk in the first instance to check eligibility and request an application form.

- **Funding Daisy Garland Ketogenic Dietitians**. The ketogenic diet is a specially formulated diet used to treat difficult to control epilepsy. It is high in fat and low in protein and carbohydrate and is formulated to sustain the state of ketosis within the body.

  Ketosis occurs when the body principally burns fat instead of the more common energy source, carbohydrate. Ketones become concentrated in the blood and inhibit seizures. To date we have funded 11 specialist trained Daisy Garland Ketogenic Dietitians who are based in NHS hospitals countrywide.

  We also provide complimentary Daisy Garland Ketogenic Welcome Boxes containing useful equipment to get new ketogenic parents/carers off on a positive start as well as free button badges highlighting that a child is on the ketogenic diet.

- **Providing useful information and support** to families, schools, epilepsy nurses, hospices and the medical profession. 'The Daisy Garland' Facebook page is a wonderful resource for parents wanting epilepsy information, tips and advice.

For further information about The Daisy Garland, please visit our website: **www.thedaisygarland.org.uk**, call us on **01803 847999** or follow us on Facebook and Twitter.

willow

special days for seriously ill young adults

Willow is the only national charity working with seriously ill 16-40 year olds to fulfil uplifting and unforgettable Special Days.

Every day deserves to be treated as precious, but the pressures of diagnosis, treatment and recovery from illness can have a devastating effect on work and family life. Willow aims to redress the balance by providing unique and unforgettable Special Days tailored to an individual's needs. We make every effort to ensure a stress-free, seamless experience which not only meets but exceeds expectations.

For some young adults a Special Day is their last chance to fulfil a dream; for others it simply provides the opportunity to bring some normality back into their lives or mark the end of treatment and a new beginning. But for all, a Special Day creates treasured memories at the most difficult of times.

To date, Willow has fulfilled more than 11,000 Special Days. Some of the most popular requests include UK city breaks, theatre trips, sporting events such as attending Premier League football matches, visits to holiday parks and family days to theme parks.

Applying is simple. Check the eligibility criteria on our website and fill out your application online, or you can download a form to fill in and send back. You will need to ask a healthcare professional to complete a short section of the form. This can be a nurse, GP, registrar or consultant you see regularly and is familiar with your condition. Don't worry if you are not sure what you want to do just yet. Once your application is approved one of our Special Day Makers will be in touch to discuss your day with you.

Please visit our website for more information: **www.willowfoundation.org.uk** or call us on **01707 259777**. You can also find Willow on Facebook: **www.facebook.com/ WillowFoundation** and on Twitter: **@Willow_Fdn**

Support Resources

Based on the shores of Lake Bassenthwaite, The Lake District Calvert Trust is a residential outdoor centre with over 35 years of experience in delivering challenging outdoor adventure holidays for people with disabilities. They offer exciting residential breaks for schools, groups, families and individuals with their friends and carers, which are accessible to people of all ages including those with the most complex needs for which most outdoor centres cannot cater.

Founded in 1978, Calvert Trust is the inspiration of John Fryer-Spedding, whose vision was to enable people with disabilities to benefit from outdoor activities in the countryside. The Lake District Calvert Trust now employs 35 permanent staff and welcomes over 3,000 people every year to their specialist facilities.

Their experienced staff and adapted equipment allows guests to really experience a range of activities including (to name a small selection) sailing, climbing, abseiling, horse riding and bushcraft. Safety of guests is paramount and their instructors hold all the relevant national governing body awards, with the centre fully licenced by the Adventure Activities Licensing Authority (AALA).

Catering for up to 60 guests per week, rooms are specifically designed to enable the care of those with profound and complex disabilities and a full range of additional specialist equipment is available on request. The team of chefs provide all guests with 3 meals per day, which are freshly prepared on-site using local ingredients and can be adapted to accommodate any special dietary requirements.

For non-residents, day visitors are welcome at their state-of-the-art Water Centre, which includes a hydrotherapy pool, multi-sensory room, poolside sauna and any mechanical poolside lifts that may be required.

The Calvert Trust has been built upon the spirit of its visitors and continues to concentrate on challenging disability through outdoor adventure, helping its visitors find out "it's what you can do that counts".

For all further information please visit **www.calvert-trust.org.uk/lakedistrict** or call **017687 72255** to find out what they can do for you in the beautiful surroundings of the Lake District.

The Maypole Project provides a lifeline of support for children with complex medical needs and their family.

We are based in Green Street Green, Kent and every year we support over 200 children with life threatening, life limiting illnesses and their families across South East London and Kent. We offer a free support Helpline: **01689 889889**.

When a child is diagnosed with a chronic illness or disability, each member of the family will react differently. Many feelings can surface including fear, sadness and anger.

The child's illness can often cause much confusion and stress within the family and it can be difficult to talk to other family members because they may also be hurting. This is where the Maypole Project can step in and help support families either through our Helpline or face to face.

The Maypole Project provides unique emotional support that is ongoing, flexible, responsive, professional and confidential. Each family is supported by one of our key workers who is specifically trained and supervised to offer emotional support ranging from befriending to counselling.

Our overall aim is to provide a FREE and unique support package for families with children with complex medical needs. This includes counselling, art and craft groups, respite days, inclusive sports and befriending.

The support we offer is ongoing, so families are able to contact us and engage with our support team if and when they need to.

Please visit our website for further information at:
www.themaypoleproject.co.uk or email us at:
info@themaypoleproject.co.uk.

We can also be contacted by phone on 01689 851596 or follow us on Facebook or Twitter.

The Amber Trust
Music for Blind Children

The Amber Trust

The Amber Trust was set up 20 years ago to help blind and partially sighted children, including those with additional disabilities, in their pursuit of music at all levels. We currently support a number of children with Batten disease – often through the provision of music therapy. However, Amber also funds music lessons, the purchase of instruments, concert tickets and travel to musical activities which may be of interest to your child.

Music is not only fun; it can also help children to share their thoughts and feelings with others – particularly when using words is difficult or impossible – and to understand what is happening and who is around in the absence of vision.

As well as working directly to support the families of children with a visual impairment, over the years the Amber Trust has spearheaded research into how blind and partially sighted children learn and respond to music, and the role it can play in their wider development and well-being. As a result, there are a number of freely available music resources created as part of the 'Sounds of Intent' project, which will be of interest to the parents of children with Batten disease and the professionals who work with them.

You can find out more at www.soundsofintent.org. This website sets out a framework of musical development for all children with learning difficulties, and has downloadable songs and other musical activities that you can use with your child. You don't need any musical training!

In partnership with the BDFA, Amber is setting up a new project at the University of Roehampton which will explore for the first time the place of music in the lives of children with Batten disease. Using the Sounds of Intent framework, it will track children's musical engagement over time. The thinking behind the project, which is based on previous, anecdotal reports, is that musical skills and interests may well continue after other abilities – such as speaking – are lost.

Beyond this, the plan is to create some new resources aimed specifically at children with neurodegenerative diseases such as Batten's, which can be used throughout their lives, and in particular to support communication and understanding in the later stages of the disease.

If you'd like to be part of the project, do please get in touch with me.

Professor Adam Ockelford
Director

Applied Music Research Centre
Queens Building
University of Roehampton
Roehampton Lane
London SW15 5SL

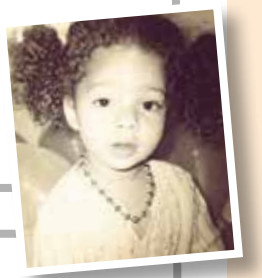
Tel: 07818 456472
Email: a.ockelford@roehampton.ac.uk

To remember Sacha who we lost 18 months ago

Sacha's Poem

A brave little girl has flown up to the sky,
Now being looked after by the angels on high.
She fought a strong battle and each night we'd pray,
But as much as we wanted, we could not make her stay.
Loved by many, adored by more,
With God's new angel, one thing is for sure –
Her spirit will always remain in our heart,
For Sacha Skinner – will never truly part.

Maureen Skinner



In memory of

Jane Pope

9th April 1954 - 30th June 2014

Don't let her become a picture on the wall,
Only a distant memory to mind you might call.
Don't turn your face from the hurt or the indescribable pain.
Look it in the eye, as she did, and from it don't refrain.

Mum is my gravity.
She runs through my veins.
She is my music, my soul.
She never complained.
She is my hands, my fingers, the shoes on my feet,
The rhythm of my heart
And it's unrelenting beat.
Don't let her become only words on a page,
Forgotten anecdotes that disappear with age.

Drink her in your tea,
Dance for her feet,
Sing her favourite songs,
See her in the eyes of the people you will meet.
Do as she did and give a stranger a lift,
Clap for her hands,
Never forget life is a precious gift.
Mum is my stop. My go.
She is my beginning. My end.
She wasn't just my mum,
She was my best friend.

Don't ask why.
There is no answer.
Take each day as it comes.
Her life was not a disaster.

She taught me to love,
To laugh and to cry.
She taught me to have fun; grab life with both hands.
And for this reason
She will never die.

Jane Pope was diagnosed with Multiple Sclerosis in 2002 on her 50th birthday. She was later re-diagnosed with Adult NCL. Her story can be found on our website. This poem was written by her daughter, Emma.

Remembering...

The BDFA is here to support families at all stages of the Battens 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.**

From our Scientific Officer

In February I attended the Unusual Suspects meeting at the Royal School of Medicine in London organised by the students4rarediseases group. It is part of their program to encourage medical students to learn more about rare diseases. It was a very interesting afternoon, with talks from scientists, clinicians and a parent's view from Daniel Lewi of the CATS Foundation (Cure & Action for Tay-Sachs), a lysosomal storage disease. It is a very important initiative, as raising awareness of rare diseases to the next generation of medical professionals is of vital importance and one of the BDFA's strategic aims.

The BDFA has funded a new Research Assistant at the Royal Veterinary College (RVC), Gini Brickell (opposite) and our collaborative PhD studentship programme continues to go from strength to strength. Congratulations to Davide and Marianna from Dr Sara Mole's lab, who have both recently been awarded their doctorates. Dr Mole also co-supervises Sophia kleine Holthaus, who is working on "Gene Therapy to treat Visual Failure in JNCL" based with Professor Robin Ali's group at the Institute of Ophthalmology. Sophia is currently writing up her thesis and will continue and extend her project this year.

On page 20 we have a full report from the BDFA-funded project with Professor Hofmann in the US, on her preclinical work for CLN1 and a report from Dr Greg Anderson on the plans to complete this project at King's College, London.

The funds that the BDFA has provided have enabled high quality vital work to be completed and we are aiming to build on our success in 2015. In addition to the projects highlighted we are collaborating with SPARKS on a research call inviting researchers to submit project proposals for the rarer NCLs. These will be reviewed and the chosen project awarded in the Autumn.

I am delighted to introduce Dr Brenda Williams as our Scientific Advisor (pictured below right with Heather Band, BDFA Scientific Officer).

Brenda is a Senior Lecturer in the Department of Basic and Clinical Neuroscience at King's College London. Her research background is in understanding how brain cells develop and communicate with each other. Brenda first became aware of Batten disease and the work of the BDFA through her colleague Professor Jonathan Cooper. Since that time Brenda and Jon have collaborated, with support from the BDFA, to address questions like: how does Batten disease affect the functioning of the different brain cell types? How does this affect cell communication and especially the health of nerve cells?

Brenda's other passion is education and her major focus at present is to expand neuroscience education at King's College. However, having been inspired by the dedication of the BDFA, together with the enthusiasm, warmth and thirst for knowledge of the many families she has met through open days and conferences, wishes to maintain her links with the BDFA through her role as Scientific Advisor.

S4RD Committee: Emma Keohane, Debra Fine, Lucy Mckay, Hannah Grant, Vicki Ward

S4RD

The students4rarediseases (S4RD) group aims to bring medical students into contact with both patients and clinicians in order to educate them on rare diseases.

By educating future doctors they aim to increase early recognition and diagnosis of rare diseases. They support and provide resources to medical students to enable them to set up their own groups to hold events to learn about rare diseases.

Their main aim is to create a network to pool resources and information about rare diseases. Students4rarediseases is currently a collaboration across five London medical schools although there are plans to form a nationwide group. If you would like more information or to get involved, further details can be found at www.students4rarediseases.org.

If you have a family member or friend currently at or applying for medical school please contact Heather Band, BDFA Scientific Officer, heatherband@bdfa-uk.org.uk for further information.

"if you hear hooves don't rule out zebra"

Drug discovery for CLN2 disease

I learnt about Batten disease whilst studying for an MSc in Neuroscience at King's College London. I was surprised that I had never heard of NCL considering how devastating it is, not only in scientific terms, but also for the individuals affected by the disease and their families.

I was keen to explore this area of research and my dissertation project was based with Dr Claire Russell at the Royal Veterinary College (RVC), where I became familiar with using zebrafish as a model for NCL. I have now graduated and, thanks to funding from donations to the BDFA, for the next 6 months I will be exploring drug discovery for CLN2 disease using the RVC DanioVision technology. This equipment was provided entirely by donations from the Freeman family in memory of their daughter Katie to help further research into finding potential therapeutic drugs for CLN2 disease. DanioVision enables us to record live videos of zebrafish and analyse many aspects of their movement.

The *CLN2*^{sa0011}-/- mutant zebrafish model is representative of the human CLN2 disease and displays typical motor defects including seizure-like activity and reduced locomotion. Using DanioVision we can examine the effects of different drugs on these motor behaviours to determine whether there has been any change or improvement. If any drugs appear promising we will be able to look into them in more detail. This research is exciting as it allows us to look at a number of different drug compounds in a relatively short space of time and gives us the opportunity to keep the momentum for CLN2 disease research at the RVC.

Gini Brickell

BDFA funded students at UCL are awarded doctorates

Congratulations to Davide Marotta (pictured below) and to Marianna Vieira who have both successfully completed their studies with Dr Sara Mole at the Laboratory for Molecular Cell Biology, University College, London.

Both students were funded by a joint initiative by the BDFA, UCL (Impact Award scheme) and NCL-Stiftung, Germany. It is one of the BDFA's key strategic aims to support young researchers, and six students from UCL, King's and the Royal Veterinary College, London have been awarded BDFA grants and obtained PhDs in NCL research since 2008. Further details on all these projects can be found on the research pages of the BDFA website.

Dr Sara Mole (UCL), Davide Marotta, Heather Band (BDFA)

NCL Resource – A gateway for Batten disease

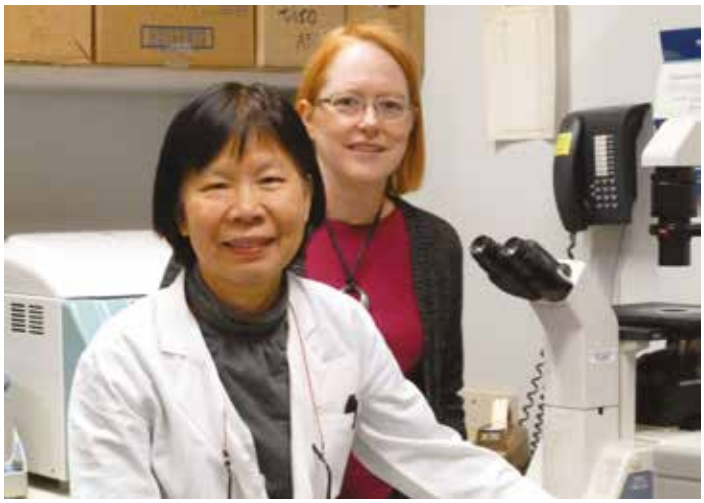
This website (maintained by Dr Sara Mole, UCL, London) serves as a global gateway for all those who have an interest in or are affected by Batten disease and for anyone who wishes to find out more.

Information can be accessed via four main routes – Clinicians, Families, Researchers and Professional Support. An additional fifth route – Research Consortia – serves to meet research needs and currently acts as a focus for collaborative efforts to identify the remaining NCL genes. The BDFA is currently working with Dr Mole to look at ways to support and develop this excellent vital resource.

Breaking News…

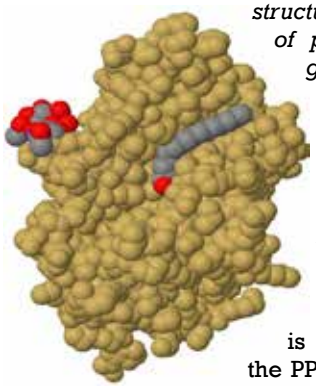
The research bid submitted by the consortium led by Dr Sara Mole, UCL in which the BDFA is taking a leading role has passed the first Stage for EU research funding. Now the hard work starts as we prepare for second stage submission in April.

Progress in preclinical study of enzyme replacement therapy for the CLN1 form of Batten disease



Drs Jui-Yun (Laura) Lu and Sandra Hofmann, University of Texas Southwestern Medical Center received BDFA support for their work on CLN1/PPT1. Dr Lingling Wang (not shown) performed all of the mouse injections for the study described in the article.

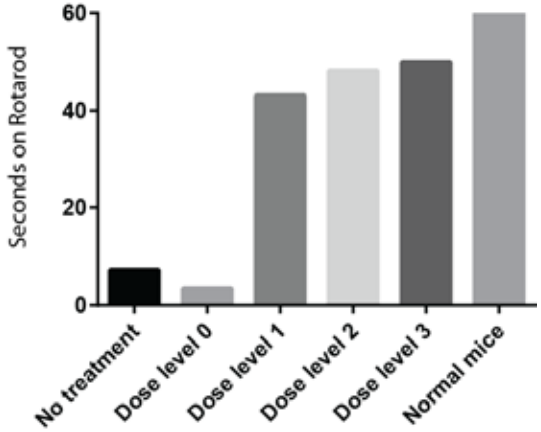
The CLN1 form of Batten disease is caused by build-up of material in the brain due to lack of an enzyme called palmitoyl-protein thioesterase, or PPT. The normal function of PPT is to break a chemical bond between naturally-occurring linkages between fatty acids and amino acids in proteins (the amino acid cysteine, in particular). The enzyme normally functions in a compartment within each cell (the lysosome) where degradation and recycling of cellular material normally occurs. Lack of the enzyme PPT is harmful to neurons and causes the serious symptoms of CLN1, infantile Batten disease. Some older children with Batten disease (CLN1-late infantile and CLN1-juvenile) also have PPT deficiency with a later onset due to mutations that provide a small amount (2-4%) of normal enzyme function. If PPT could be supplied back to the brain, it is possible that the loss of neurons could be stopped although probably not reversed.



Atomic model of the PPT enzyme. The structure is derived from X-ray analysis of pure crystalline PPT protein. The grey atoms in the centre are the fatty acid that PPT has just removed from a protein. The grey and red "knob" on the left side is the mannose 6-phosphate sugar that is recognized by cells and allows PPT to be taken into the cell and the lysosome.

A mouse model was created that is genetically engineered to lack the PPT enzyme. The mice become ill at around 5 months of age and survive for 7 to 8 months. The normal lifespan of a mouse is about 2-3 years. The mouse model has been useful for testing therapies for Batten disease caused by lack of PPT.

The project funded by the BDFA sought to determine the effect of administering PPT directly to the central nervous system of PPT-deficient mice by injection into the cerebrospinal fluid, as in a spinal tap. About 80 mice were used for the experiment. Mice received no injections, injection of a solution of artificial cerebrospinal fluid, or the same fluid with three different dose levels of PPT. For technical reasons only one set of injections was performed at around 6 weeks of age, which is early adulthood for a mouse. Injected mice were subjected to motor tests (see chart below) throughout their lifespan. Data gathered so far indicates that injected mice live longer, perform better on motor tests and have improvements in brain and spinal cord pathology.



Mice treated with PPT injected into the cerebrospinal fluid perform better in a test of motor function. The ability of mice to stay on a rotation rod is recorded. Each test is ended at 60 seconds and normal mice complete the test easily. Mice lacking PPT can only stay on the rod for about five seconds when they are seven months old. Mice treated with PPT perform better on the test, and the higher the dose of PPT they are given the better the performance.

Currently there is a clinical trial for children with CLN2 Batten disease in which the deficient enzyme TPP-1 (BMN-190) is being administered to the cerebrospinal fluid of the brain. The clinical trial was based largely upon preclinical mouse studies involving the CLN2 enzyme, much like the current study of CLN1.

Results of the current study funded by the BDFA on CLN1/PPT support using the same approach for CLN1 disease, particularly if BMN-190 shows good results for late infantile NCL. Future experiments would need to prove the safety of the PPT1 enzyme in at least two species, which is a requirement for regulatory agencies, and more detailed efficacy studies for the final therapeutic product will also need to be done. Because the final potential treatment given to patients will need to be tested in a variety of ways to ensure safety and effectiveness, these studies are best done in the context of a pharmaceutical company drug development program. The results of the study funded by the BDFA will make infantile Batten disease/CLN1 an attractive opportunity for drug development.

Sandra L. Hofmann, M.D., Ph.D.
Professor in Internal Medicine
University of Texas Southwestern Medical Center, USA

Cell based systems for drug discovery in JNCL

Professor Jon Cooper, Dr Brenda Williams and Dr Greg Anderson of King's College London have been awarded funds for a six month extension for their work on cell based systems for drug discovery in CLN3, JNCL.

There have been some delays in providing resources for the project by King's and so the College has agreed additional funds to supplement the current research grants awarded in 2013-14 by the BDFA, BDSRA (USA) & NCL-Stiftung (Germany).

The aim of this research is to generate a human neural stem cell line that carries the CLN3, JNCL mutation (mistake in the CLN3 gene) in order to generate different brain cell types. The King's team has completed the first steps to reproduce the mutation, having introduced it into cells, and is now looking for the specific cells in which this has happened successfully.

Neural stem cells are important as they are the founder cells of the central nervous system (brain and spinal cord). They make all the nerve cells (neurons) and all the support cells (astrocytes and oligodendrocytes) found in the adult brain.

They may be able to repair or replace damaged brain cells, or may be used to make different brain cell-types in a dish in order to study their function. The cell lines which may have the mutation that the team is aiming to produce could then be very useful to use for a drugs screen.

If you would like to find out more about this project and other NCL research, please come to the PSDL lab day on 18th June 2015. To book a place, please contact Lucy Roose, BDFA Office Manager by emailing admin@bdfa-uk.org.uk.

Research Studies and Clinical Trials for CLN2

There are currently two major clinical research centres seeking to enrol patients, who meet the eligibility criteria, to participate in ongoing clinical studies.

- The department of Genetic Medicine at Weill Cornell Medical College in New York City is conducting a CLN2 gene therapy study.
- The University of Rochester Medical Center (UMRC) in the 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 Late Infantile NCL who meet the eligibility criteria would be enrolled in the study for 18 months. The study aims to determine whether gene transfer surgery, in which an experimental drug call AAVrh.10.cuhCLN2 is administered to the brain, can be achieved safely and whether the procedure will slow down or halt the progression of the disease.

There is no compensation for participating in this study. The costs of travel and accommodation are not covered. There are no costs associated with the tests and procedures that are conducted under the scope of the research study. For more information please contact Denesy, Research Manager, at dem2026@med.cornell.edu. Please search for "Safety Study of a Gene Transfer Vector (Rh.10) for Children with Late Infantile NCL" on the www.clinicaltrials.gov website.

Weill Cornell Medical College

CellCept for Treatment of Juvenile Neuronal Ceroid Lipofuscinosis (JUMP)

The clinical trial at the University of Rochester Medical Center (UMRC) is to learn if mycophenolate (CellCept) is safe and well tolerated in children with JNCL. The JUMP study focuses on evaluating CellCept and its effects on the symptoms of JNCL. It must be stressed that this trial is only open to US patients who meet the eligibility criteria. The trial is ongoing and to date 19 patients have been enroled with 17 having completed the study. URMC are continuing to recruit and enroll the remaining patients and although no data is currently available to be released they hope to have some preliminary results by late 2015. There is more information regarding the trial itself on the website provided by the U.S. National Institutes of Health, www.clinicaltrials.gov.

UNIVERSITY of ROCHESTER MEDICAL CENTER

Heather Band
BDFA Scientific Officer

Call for Research Proposals

Sparks, on behalf of the Batten Disease Family Association (BDFA), would like to invite grant applications for medical and scientific research into Batten disease.

Applications are invited for high quality research into the causes and treatments for the following forms of Neuronal Ceroid Lipofuscinoses, NCL (Batten disease) CLN1, CLN5, CLN6, CLN7 and CLN8. Collaborative applications between the UK and other international research groups are particularly encouraged. All applications will undergo competitive scientific peer-review. We will consider applications from overseas but please contact us for more information.

Guidance for Applicants:
Applications are welcome for project grants of up to 1 year for a maximum award of £50,000.

Grants can cover any combination of salaries, equipment and consumables as long as all costs are fully justified within the application. Please note we will not cover the salaries of the principal applicant or any tenured staff members.

Grants are only made for research projects where the principal applicant has a contract at the host university that extends beyond the life of the proposed project. Researchers who are making a substantial intellectual contribution to the project and require personal support from the grant may apply as co-applicants.

The deadline for submission of full applications is 5.00pm Tuesday 5th May 2015. Grants will be awarded in October 2015.

Please contact Dr Virginie Bros-Facer (medical.team@sparks.org.uk) for an application form and further details.

Peer Review:
All complete applications are assessed by full peer review, firstly by independent external referees and then by Sparks Medical Advisory Committee. Sparks is a member of the Association of Medical Research Charities (AMRC) – a membership organisation of the leading medical and health research charities in the UK. www.amrc.org.uk. The final award decision will then be made by the BDFA Board of Trustees and approved by Sparks.

Sparks holds an AMRC Certificate of Best Practice in Medical Research and Health.

Research Peer Review. This signifies that Sparks is demonstrating best practice in Medical and Health Research peer review as audited by AMRC in 2011.

Raise Funds for Research?

We have places in a whole series of events over the next 9 months ranging from road runs, muddy runs, cycling and long walks. A lot of the events have distance options so please contact us to find out more or visit **www.bdfa-uk.org.uk/challenge-yourself-for-bdfa** for further details.

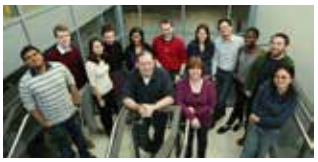
All money raised will be used to fund research into the NCLs in conjunction with Sparks, the children's medical research charity. Please note, there are minimum fundraising targets for each event.

If you think you are up for a challenge please contact Kate or Gaynor on **07876 682589** or email: **fundraising@bdfa-uk.org.uk.**

You can now also find us on Facebook at BDFA Fundraising.

So dust off those trainers, get those wheels oiled and raise some money for essential research into the causes of Batten disease. Together, we WILL make a difference.

PSDL Lab Open Day
Thursday 18th June 2015



http://tinyurl.com/newpsdl

James Black Centre, Denmark Hill, London 10.00-16.00

BIOMARIN

Californian pharmaceutical company BioMarin issued a press release in January providing preliminary data from the Phase 1/2 BMN190 trial in CLN2 (Late Infantile) Batten disease as follows:

BioMarin Pharmaceutical Inc. (Nasdaq:BMRN) today announced interim results from its Phase 1/2 pivotal study for BMN 190 or cerliponase alfa, a recombinant human tripeptidyl peptidase 1 (rhTPP1), to treat of patients with late infantile CLN2 disease, a form of Batten disease. Interim data indicates that in all nine of the BMN 190 patients who have been followed for at least six months and up to 15 months, the treatment appears to show stabilization of the disease compared to the natural history based on a standardized measure of motor and language function.

The primary end point of the study is a standardized mobility and language score using a CLN2 specific rating scale. The scale separately measures performance of mobility and language with normal function in each being a score of three and no function being a score of zero. The highest score possible is six.

According to data from a natural history study of the disease, patients generally lose one point every six months and generally lose most language and mobility functioning over a two to four year period in this rapidly progressive disease. In the nine BMN 190 patients treated for more than six months and up to 15 months, six patients showed no net change in their CLN2 rating scale score, while the other three showed a decline of one point.

In addition, seven of the nine patients in the BMN 190 study for more than six months were matched to between one and 12 individuals from the natural history data set according to baseline age and disease severity. (For two of the BMN 190 patients, there is no matched patient in the natural history database based on age and disease severity.) All seven BMN 190 patients with six months of treatment and at least one control match had better walk/talk scores as of their last evaluation than their natural history counterparts.

Additional detailed information on the interim preliminary results from the nine patients who have been on BMN 190 for more than six months and their matched natural history counterparts can be found at http://www.bmrn.com/pdf/JPMPresentation011215.pdf.

"This trial represents the essence of BioMarin's commitment to patients with fatal rare diseases and no treatment options. This initial look at the data is encouraging, and this therapy may make a meaningful difference for children with this form of Batten disease. We look forward to working with the regulatory authorities to determine if this single study will support regulatory approval as quickly as possible," said Jean-Jacques Bienaimé, Chief Executive Officer of BioMarin.

"This interim data represents an important step on a journey to develop a treatment for CLN2 disorder that may be able to slow the course of this fatal disease," said Angela Schulz, M.D. Ph.D., Department of Paediatrics, University Medical Center Hamburg-Eppendorf. "We appreciate the commitment of the children and their families who are participating in this study."

"We welcome BioMarin's update on nine of the 24 patients participating in this clinical trial and appreciate the efforts they are making in developing a treatment for this deadly ultra-rare disease," said Andrea West, Chief Executive, Batten Disease Family Association.

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

This is welcome news and the BDFA will continue to work closely with BioMarin, regulatory bodies and partner patient organisations to ensure the best support for those families on the trial and the best outcome for the future. If you would like any further information about the trial please contact Andrea West on **01252 416110** or **andreawest@bdfa-uk.org.uk**.

CLN2 Burden of Disease Project

To enable us to influence government and policy makers to deliver quality services and treatments to families living with a diagnosis of Batten disease we need to have good research evidence to present. For families living with CLN2 (Late Infantile Batten disease) we are working with BioMarin to deliver a Burden of Disease Project which will provide the evidence we need to approach governments both in the UK and Europe to press for the need for change.

The first stage is a focus group to get the views of families with a CLN2 diagnosis. We are looking for families from all stages of the journey from diagnosis and also those families who are bereaved. The focus group will be held on Saturday 25th April in Sheffield (location to be confirmed). All your travel and accommodation expenses will be paid (including a Friday night stay if this makes travel easier) and you will receive a small honorarium for your time. We have also organised a childcare programme with qualified staff for both affected children and siblings. Unfortunately, because of the conflict of interest those families currently enrolled on the BMN190 trial are unable to take part. However, anyone who has a direct caring role for a child with CLN2 (Late Infantile Batten disease) including siblings and grandparents would be very welcome.

This is your opportunity to share your invaluable experience and stories of living with this devastating disease and we would be very grateful if you would consider attending as it is only with research like this that we can hope to influence government to meet the needs of families today and in the future.

If you would like more information or to discuss this further please contact Harriet Lunnemann on **support@bdfa-uk.org.uk** or **0800 046 9832.**

BDFA

BATTEN DISEASE FAMILY ASSOCIATION

Save the Dates

20-21 June 2015

Tenth BDFA Annual Walk

(venue tbc)

17-18 October 2015

BDFA Family Conference

at the Village Hotel
Coventry

Sponsored by the Village Hotel, Coventry

For more information
please call 01252 416323 or
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