BDFA

BATTEN DISEASE FAMILY ASSOCIATION

BATCure

SPRING 2017

making **the** difference

Contents

Diary Dates 2017

BDFA Education Training Days
Friday 19 May at Linden Lodge, London SW19
Monday 5 June at Royal Blind School, Edinburgh

RVC Lab Open Day with Dr Claire Russell
Royal Veterinary College, London NW1
Thursday 8 June

Batten Disease Awareness Day and BDFA Starlight Ball
Warbrook House, Hook
Friday 9 June

Marathon Walk
Saturday 23 September, London

Batten Disease Professionals Workshop
Crowne Plaza, Stratford-upon-Avon
Friday 24 November (professionals only)

BDFA Family Conference 2017
Crowne Plaza, Stratford-upon-Avon
Friday 24 - Sunday 26 November

NCL 2018
Royal Holloway, University of London, TW20
12 - 16 September 2018 (provisional)

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

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

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

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

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

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

Further information:

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

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

Letter from our CEO

Dear BDFA members, friends and colleagues

With spring nearly here, it seems like only yesterday that we were in Stratford-upon-Avon at the last BDFA Family Conference. Planning is in full swing for our next conference at the end of November 2017 and we look forward to welcoming all of you again. Dave Mitchell, BDFA Trustee and grandparent, has written a piece about the 2016 conference in this edition of the newsletter. We are still compiling the programme for this year, so please let us know if there is any specific session, information or area that you would like us to cover, as this is your conference and your input is vital.

This edition of the newsletter is packed with stories, information and resources including what will become a regular piece from Laura Lee, our Batten CNS at Great Ormond Street Hospital. Laura's role is wholly funded by the BDFA and the fundraising that all of our supporters work so hard at. This enables us to do our work and we are so grateful to each of them. I was told recently that without the BDFA funding for the Batten CNS role, the Compassionate Use Programme for CLN2 disease at Great Ormond Street Hospital would not have been possible. We continue to need your support to enable us to provide these vital support services to families and to fund much needed research both in the UK and worldwide.

We are delighted to announce that to help with our fundraising, we are hosting the first BDFA annual Starlight Ball at the end of Batten Disease Awareness Day on Friday 9 June. This will be a fantastic way to spend time together with friends and provide much needed support for our work so we would be hugely grateful if you are able to come along and to share details of the event with your friends.

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

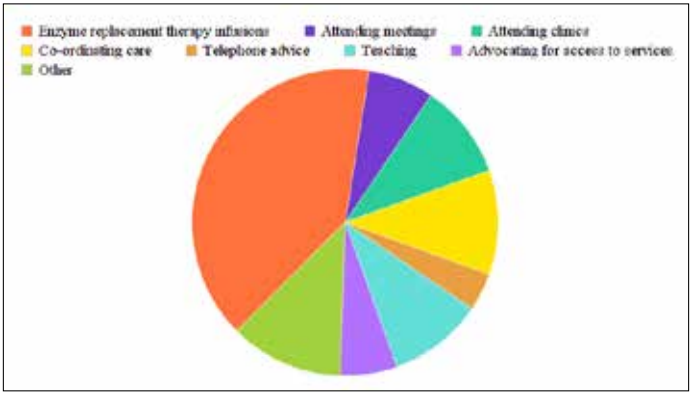
The Starlight Ball will be held on Batten Disease Awareness Day – Friday 9th June – at Warbrook House, Eversley. Tickets are £75 per person. Hotel rooms are available for £42.50 per person. Come and help us raise much needed funds and awareness on this special night.

Warbrook House is set in acres of gardens, greenery, and woodland which will come together to form the perfect backdrop for our special evening. Tickets are available online or please call **01252 416323**.

CLINICAL NURSE SPECIALIST - Laura Lee

As the Batten Disease Clinical Nurse Specialist, I am based at Great Ormond Street Hospital (GOSH) but this unique role is to support children and families living with Batten disease throughout the UK, not just at GOSH. I have already met lots of incredible children, young adults, families and professionals throughout the country.

Understanding the needs of children and families is important to me and this has meant adaptation and flexibility within the role in an effort to provide the most valuable support. The BDFA plays a vital role for families and I have been working closely with them to help define my role within the greater team and ensure I am able to offer the most useful service.



In my short time in this role I have been working with a number of families and the pie chart shows how my time is spent during an average week.

Numerous professionals have asked me to provide education and support to help them understand the condition and challenges faced by you as families. Their enthusiasm to learn more about the disease to provide the best possible care for your children has been clear.

However, a common frustration for families looking after a child with Batten disease is not feeling heard by professionals, or problems getting through to the right people who understand Batten disease. I endeavour to make the communication between you and healthcare professionals as easy as possible by bridging that gap. This could be as simple as a telephone discussion, a letter of support, or for more complex situations, bringing professionals together via a multidisciplinary team meeting. If I cannot provide the information or service you need, I will do everything possible to help you access the people who can.

"The BDFA plays a vital role for families and I have been working closely with them to help define my role within the greater team and ensure I am able to offer the most useful service."

Whilst there are many ways I hope to support you, there may be other things I have not thought of. I am relying on you; the children, families, carers and professionals in the world of Batten disease to tell me what's important to you. I have created an anonymous SurveyMonkey which I urge you to complete to feed back your ideas:
www.surveymonkey.co.uk/r/KJFNC3H

My contact details are below. Please note that I work clinically every Tuesday and Wednesday so at these times please email or leave a voicemail and I will get back to you as soon as I can.

Great Ormond Street Hospital for Children NHS Foundation Trust

Laura Lee
020 7829 8721 / Laura.lee@gosh.nhs.uk

We found out Ryan's diagnosis in November 2013, a few days after his eighth birthday. That is a day we will never forget. In the months leading up to it, we had attended several appointments at different hospitals to find the reason behind his rapidly deteriorating vision. When we were asked to go to Moorfields Hospital without Ryan, we knew something was wrong.

It's hard to describe the feeling of being told that your child has Juvenile Batten disease. To be told that your child will lose every ability. To be told that your child will have a short life. There was such an overwhelming sense of sadness, grief, panic and a whole host of other emotions. We were lucky that day to have two family members with us, as we cannot even recall how we got home. It was just a numb blur. Similar can be said for the following days and weeks too. It was too painful to comprehend.

We wanted to keep everything normal for Ryan's sake, as he had gone through so much already. I guess you could say that we were living in denial for a while, just pretending that this wasn't happening. At the time, that was what felt right for us. Telling our closest family members was painful, so we just told a handful of people.

A few months down the line, when we were at Ryan's school trying to get all the help in place, we realised that we didn't need to be alone. They were so supportive and encouraging and it prompted us to realise that Ryan needs us to be strong… and in order to be strong, we needed to come to terms with his diagnosis. We went home and opened the brown envelope that had been handed to us that day at the hospital. It gave us an insight into what we could expect about this disease, and also information about the BDFA.

"They were so supportive and encouraging and it prompted us to realise that Ryan needs us to be strong... and in order to be strong, we needed to come to terms with his diagnosis."

Although we were slightly hesitant at first, we made contact with the BDFA and from then on, our world has been a little bit brighter. We have been able to meet with different doctors and specialists and have support in place with the school and local services. It has been a difficult process to go through, but necessary. We can't say that we have come to terms with it, because we don't feel that is ever possible, but we would say that with the help and support from all of those around us, it has made the process a little easier to bear.

We attended our first BDFA Conference in November 2016, and met lots of lovely parents, all facing the same journey as us. For us, it was a wonderful experience – feeling totally welcome and at ease in their company. Being able to openly share stories and life events was comforting, as we no longer feel alone.

At present, we feel empowered to raise awareness and throw ourselves into fundraising, hoping that it will go some way to finding a cure. We aim to live each day to the full with the ups and downs. Some days are easier than others, but ultimately, we know that support is always there.

Leanne Buggins

CLEARVISION CHILDREN'S LIBRARY

Could your child or teenager benefit from the ClearVision library? ClearVision is a charity which runs a postal lending library of children's books. All books are dual format, with print and braille (or print and Moon) together on the page so young people can read with sighted family, teachers and friends. It's a great way to enjoy the comfort and pleasure of sharing stories, as well as supporting your child's literacy, motivation and interests.

ClearVision's collection includes fiction and non-fiction picture books covering a huge variety of authors and interests from facts on the Great Barrier Reef or Henry VIII to stories about hippos or Peppa Pig. Books cover a wide range of reading levels, including those suitable for older readers who are just starting braille, and short chapter books for more confident readers in paper braille with a print copy for adults to read alongside.

There are also age-appropriate books for older teenagers with a lower reading age. In addition to braille, there is a small collection of simple books in Moon, a touch-reading system similar to braille where the letters look more like print.

Some young people with Battens find Moon easier than braille because the letters are more like the print they remember; and some move on to it if braille becomes difficult.

ClearVision also has tactile books for schools and Visual Impairment/Sensory Support Services to borrow. These are mainly handmade and the illustrations are designed to be 'read' by touch, with different textures to investigate.

"It's a great way to enjoy the comfort and pleasure of sharing stories, as well as supporting your child's literacy, motivation and interests."

They're great for helping young people exercise their touch skills whilst exploring the pictures. They're loved by large print readers, braillists and children who don't use either. Photos can be found on ClearVision's Facebook page and website.

If you think your child or teenager may benefit from tactile books, please ask their school or Sensory Support Service to contact us.

Our library is entirely free of charge for families. We send out six books initially, and change three at a time, using the Articles for the Blind freepost service, so there are always some at home to read. There are no late fees, and books can be kept for up to six months.

Please contact ClearVision with any questions or complete the membership form on our website to join the library.

Email: **info@clearvisionproject.org**
Tel: **0208 789 9575**
www.clearvisionproject.org
www.facebook.com/clearvisionproject
Twitter: **ClearVisionOrg**

ClearVision Books

Erasmus & EU Funded Project on Juvenile Batten disease and Education

The European Union-funded Education project is now in its third year and continues to bring together many people working with children and young people with CLN3 disease (Juvenile Batten disease) from Norway, Germany, Finland, Denmark, Scotland, England and the United States. The BDFA has been actively involved throughout the project and has worked with other patient associations, specialist Batten disease services and specialist schools for the visually impaired.

The third meeting of the working/steering committee was held in June 2016 at the Royal Blind School, Edinburgh and the fourth meeting was held at the School for the Blind in Hamburg in December.

The previous year's surveys and interviews involving parents and professionals have been analysed and summarised and the results will be disseminated at meetings in each participating country between March and September this year. The interviews and surveys from the UK and the other participating countries have provided a great deal of information about the experiences of children, young people, their families and the professionals working with them.

Work has been continuing on developing educational tools to support the learning of children and young people with CLN3 disease (Juvenile Batten disease) including work on communication, literacy, behaviour issues and transitions. Participants have been writing chapters for the project textbook that will be available later this year in print and online. The BDFA and the BDSRA are working on the "Family" chapter along with contributions from the family organisations in Germany, Denmark and Norway.

The BDFA are delivering three dissemination events in the UK at New College Worcester on 22 March, at Linden Lodge School, London on 19 May and at the Royal Blind School, Edinburgh on 5 June. The project findings will be presented and there will be sessions on a range of issues such as communication, memory difficulties and challenges with behaviour. These events are for parents and professionals, including teachers, learning support assistants, therapists and SEN officers. Please let Harriet Lunnemann know if you would like to attend (**support@bdfa-uk.org.uk** or **0800 046 9832**).

The BDFA is also a key stakeholder in the MIND project (Music in Neurodegenerative Disease) together with the University of Roehampton, the Amber Trust and Chiltern Music Therapy. This project is looking at the importance of music and music therapy to children and young people affected by Batten disease and on how music can support learning, communication and enjoyment.

Professor Adam Ockelford and Rebecca Atkinson led a session on the MIND project at the BDFA's 2016 Family Conference. The results of the questions on music and music therapy in the project surveys will also be presented at the dissemination events together with updates on the MIND music project.

BATTEN DISEASE FAMILY ASSOCIATION
Together we WILL make a difference
Registered Charity No. 1084908

Helping us to support families

In a difficult and changing environment it is a challenge for many charities now to continue to deliver much needed services to the people who rely upon them. The BDFA is no different, and with a small dedicated team we are always looking at ways in which we can ensure that the vital services for families living with this devastating diagnosis are here when they need them.

We also know, because families tell us, that we could do more. Many of you know that our Support and Advocacy Team consists of Harriet Lunnemann (qualified social worker) and Barbara Cole (volunteer Education Advisor). This team now has much more work than it can do alone and whilst we have always worked closely with other organisations to make sure that families receive support, our plans now include another support and advocacy worker. This new team member would be based in the Midlands to more easily meet the needs of families there and in the North of England and Scotland.

To enable us to make this commitment we need to know that we have a guaranteed source of funding to ensure that the service can continue. Families tell us that when they first receive the devastating diagnosis, people they know always ask them "What can I do?" Asking them to support the BDFA through our regular giving programme would enable us to make sure that we are here when families need our support and also to make sure that the professionals around them are supported with the Batten knowledge and expertise that they need.

Less than the price of a cup of coffee, £2 per month for example, would enable us to:

- Run the BDFA Freephone Helpline for families and professionals for a month.
- Provide a family folder for a newly diagnosed family.
- Enable a school visit by our education advisor to support a child in the classroom.

Visit **www.bdfa-uk.org.uk/donate** for more information

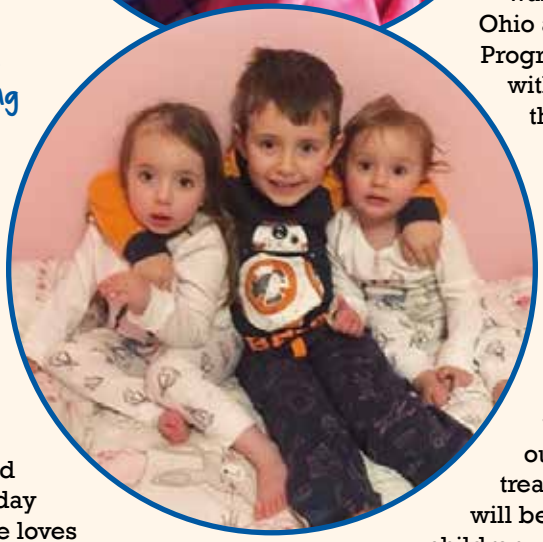
Compassionate Use Programme

Our hearts broke into a million pieces when we were told our beautiful little girl Nicole had Batten disease. We had the answer to the puzzle but it was more devastating than we could ever have imagined. The day we found out saw us plunged into an unbearable place – no cure, no treatment, nothing. So were we just meant to go home and watch our little girl decline?

Then a short while after, we were given a lifeline; hope, when we were offered a place on the Compassionate Use Programme at Great Ormond Street Hospital.

We know this is not a cure, but it is hope, and it is this hope that is giving us the strength we need.

We travel every fortnight from Newcastle to Kings Cross then walk along to the hospital.



Laura, the Batten disease Clinical Nurse Specialist, and all of the nurses are just incredible. They are all not only amazing professionals, but they are supportive and caring. They even surprised Nicole with birthday cards and presents on our first visit after her birthday (a little birdy must have told them Nicole loves horses because one of the presents was a gorgeous cuddly pony!). The staff all know Nicole loves her films with Disney princesses, Toy Story and Cars, and when they come in to do her observations they always ask "so what are we watching today, Nicole?" Everything seems a lot brighter when you watch a Disney Princess film... and we certainly watch a good few when we are there!

In December, we were given the news that our baby girl, Jessica also has the condition. We were completely floored. We were so sure Jessica would be okay and we were even thinking, "Whatever Jessica does in her life, she won't be doing it just for herself, she will be doing it for her sister too".

Now we had a new focus, we needed to get the same treatment for Jessica.

The Compassionate Use Programme was not an option (all places had gone but even if there were spaces, Jessica was too young) but we were aware of Ohio and Hamburg already having a Sibling Programme established, so we made contact with doctors in Hamburg so we were on their radar. We were desperate for help and we reached out saying just that.

We maintained regular communication with America and Hamburg and found out that Hamburg was in the process of amending the protocol to remove the lower age limit, which immediately meant Jessica was eligible. We would travel anywhere in the world if it meant our children being given access to treatment. We will be proud to know Jessica will be providing vital data on treatments to children under two years of age as this will be the first time it has been done.

After many conversations and emails, we are delighted to say that we have been accepted in Hamburg for both Jessica and Nicole to receive treatment together at the German site... and so our next adventure will begin.

Gail Rich

Gene Therapy Development for CLN3

In January 2017, Abeona Therapeutics announced that they had received orphan drug designation by the European Medicines Agency for their ABO-201 Gene Therapy program for CLN3 (Juvenile Batten disease).

This is a great and much needed step forward as orphan drug designation by the European Medicines Agency provides incentives for companies to conduct research into rare diseases and we are hoping that accessible clinical trials will follow. To qualify for orphan designation, a medicine must meet the following criteria:

- it must be intended for the treatment, prevention or diagnosis of a disease that is life-threatening or chronically debilitating;
- the prevalence of the condition in the EU must not be more than 5 in 10,000 or it must be unlikely that marketing of the medicine would generate sufficient returns to justify the investment needed for its development;
- no satisfactory method of diagnosis, prevention or treatment of the condition concerned can be authorized, or, if such a method exists, the medicine must be of significant benefit to those affected by the condition.

The full press release from Abeona can be found by following this link: **http://m.marketwired.com/press-release/-2185873.htm**

For all of our families with children and young people with CLN3 (Juvenile Batten disease), we remain in regular contact with Abeona Therapeutics and have asked for clarification on whether families from outside the United States can access their forthcoming clinical trial. We have received the following information from them and we have asked to be kept updated on any changes or new information as it becomes available. This also includes information and timing concerning a site for the trial in Europe.

"At this time Abeona Therapeutics are focused on the necessary studies and efforts needed to enable regulatory allowance by the FDA here in the US. With that, we are working with the clinicians at University of Rochester – the selected initial site – to determine the design for the study and the criteria for participation. We are asking those interested in the study to place their information into the University of Rochester's Institutional Review Board approved online contact registry."
https://redcap.urmc.rochester.edu/redcap/surveys

The University of Rochester Batten Center has further information at the following link: **https://www.urmc.rochester.edu/neurology/batten-disease-center.aspx**

CellCept for the Treatment of Juvenile Neuronal Ceroid Lipofuscinoses (JUMP)

The primary objective of this trial was to establish the safety and tolerability of short-term administration of mycophenolate (Cellcept). Dr Erika Augustine (University of Rochester Medical Center) presented results of the study: *Developing therapies for individuals with CLN3 disease – results from a phase 2, double-blind, crossover study of mycophenolate* at NCL2016 held in October in Boston, USA.

Nineteen people were enrolled into a double-blind crossover study. This meant that they were given 8 weeks of the drug, mycophenolate, followed by 8 weeks of placebo, or vice versa. The most common adverse events (often referred to as side effects) in participants on mycophenolate were vomiting, diarrhoea, and cough. These symptoms were mild in severity.

This study has now been completed. Overall, mycophenolate was well tolerated in the short-term by those taking part in the study. Evaluation of long-term mycophenolate exposure is needed to evaluate whether mycophenolate is beneficial in people with CLN3 disease.

A useful source of information about clinical trials is the National Institutes of Health (NIH) website. Visit **www.clinicaltrials.gov** and then search for Batten disease or Neuronal Ceroid Lipofuscinoses (NCL).

In November I had the privilege of attending the BDFA family conference at the Crowne Plaza Hotel in Stratford-upon-Avon.

I arrived at the recently refurbished hotel in the late afternoon and the ever-efficient BDFA staff, under the guidance of Andrea, was on hand to make me feel welcome and make sure that I settled in easily.

The venue certainly had the 'wow' factor and all weekend nothing seemed to be too much trouble for the BDFA team or the hotel staff.

There was certainly a mix of conference attendees – grandparents, parents, siblings, friends old and new, and most importantly the children affected by this terrible disease. There were also parents of children who have passed away due to the disease, who were able – however hard it was for them – to pass on their experiences to parents of children recently diagnosed with the disease.

Over the weekend 'The A Team' catered for everyone. The adults attended various informative workshops and the children had fully trained carers to help them with their numerous activities. I visited the children's area and was surprised to see a 'mini farm' with animals for the children to pet. All the children were having a fantastic time under the guidance of the BDFA's Harriet Lunnemann.

One of the many highlights of the weekend was the opportunity to meet with the dedicated researchers and representatives of BioMarin and Abeona (two pharmaceutical companies making massive strides in combating this disease). Research documents can be daunting to the average person, but to actively talk to these researchers helps to make things much clearer. It is enlightening to know that money that you have raised by your own fundraising efforts is being used wisely and in the right areas.

Another highlight was the meal on the Saturday night. The hotel certainly made the event special. Part of the evening was the BDFA Quiz which was extremely competitive (yes, really). I had the 'honour' of being on the winning team, along with Paul, Jayne and several others. It was certainly memorable!

On Sunday all the children went on a canal trip in a specially adapted narrow boat. Their faces were beaming when they came back and they all had a fantastic time. As we were in Stratford, there was also a theatre production, complete with Father Christmas. This was very interactive and again the children all had a marvellous time.

The weekend went too quickly. I was so grateful to the BDFA staff team for making sure that everyone felt welcome while they all worked so hard over the weekend from early morning to late evening. I would also like to give special mention to Paul Robinson, the BDFA's volunteer photographer. He was always on hand to take photographs of all the events and children and even printing out the photos late at night ready for display the next day (see overleaf).

On behalf of the BDFA Board of Trustees, I would like to express gratitude to Andrea and her team for their hard work and dedication before and during the conference to make it such a success, as well as my gratitude to all the young people and their extended families for making such an immense contribution over the weekend.

CROWNE PLAZA HOTELS & RESORTS

Dave Mitchell
Grandparent and BDFA Trustee


BATTEN'S
BDFA
SCANIA


Proud to support
the BDFA
PER
PRODUCTIONS

Remembering...

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

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

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

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

In memory of my beautiful, spirited daughter

Joanna

20th July 1984 ~ 21st April 2008

and my handsome, smiley

James

28th January 1990 ~ 16th July 2016

Friend, please don't mourn for me
I'm still here, though you don't see.
I'm right by your side each night and day
And within your heart I long to stay.

My body is gone but I'm always near.
I'm everything you feel, see or hear.
My spirit is free, but I'll never depart
As long as you keep me alive in your heart.

I'll never wander out of your sight –
I'm the brightest star on a summer night.
I'll never be beyond your reach
I'm the warm moist sand when you're at the beach.

I'm the colourful leaves when Autumn's around
And the pure white snow that blankets the ground.
I'm the beautiful flowers of which you're so fond,
The clear cool water in a quiet pond.
.
I'm the first bright blossom you'll see in the spring,
The first warm raindrop that April will bring.
I'm the first ray of light when the sun starts to shine
And you'll see that the face in the moon is mine.

When you start thinking there's no one to love you,
You can talk to me through the air around you.
I'll whisper my answer through the leaves on the trees,
And you'll feel my presence in the soft summer breeze.

I'm the hot salty tears that flow when you weep
And the beautiful dreams that come while you sleep.
I'm the smile you see on a baby's face.
Just look for me, friend, I'm every place!

Marshall Starkahn

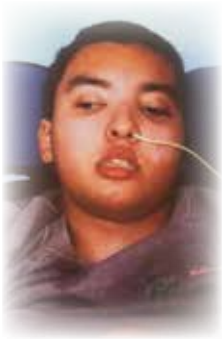
11th November 2008 ~ 29th October 2016

Hello. My name is Marshall and my eighth birthday was on the day of my funeral. I had an amazing send off, with so many people in superhero costumes but everyone said I was the real superhero. I was loved so much. Still am and always will be. And not because I had the most beautiful face and big hazel eyes. But because I was kind and loving and accepted everything that happened to me over the last three years without tears or complaint. I embraced every moment for as long as I was able. Nana told me all the time that I sent out rays of love like the sun sends out rays of light. I faced every day with hope and optimism. Batten disease robbed me of my childhood but my family tell people 'Trev the Rev' got it so right when he said: "Marshall's acceptance, apparently unafraid, prepared and ready, says a lot for the way we grown-ups complicate life when a young boy teaches us how to face both life and death". My family fought hard to keep me free from the evil symptoms of Batten disease and thanks to them I rarely suffered from seizures or pain. And because I hated being anywhere but home, when a chest infection made me very ill I was kept out of hospital to peacefully leave this world wrapped in the arms of Nana and Mummy with Granddad, Daddy and Uncle David holding my hands. I'm loved so strongly it's like they are always with me.

Daniel Khan

22nd June 1997 ~ 18th October 2015

My son Daniel was born a healthy and good looking baby boy in 1997. When he was three the doctors' appointments started and my life changed for ever when we were told he was affected by Batten disease. Daniel lost the ability to walk, talk and eat, suffered chest infections and was on life support. He was always so strong and such a smiley boy in spite of so many hospital admissions. In 2015 at the age of 18, Daniel passed away in his sleep and my whole life changed. I didn't want to bury my son, and I didn't know how to end my pain. Sometimes hard work is not enough and however much you want to, you can't make things right. The happiness has gone from my life and sadly our story does not have a happy ending. I miss my son Daniel all the time.



Alisha Lennon

Born 5th May 2006, became an angel
7th August 2016, aged 10

When somebody loved me, everything was beautiful
Every hour spent together, lives within my heart
And when she was sad, I was there to dry her tears
And when was happy, so was I when she loved me.

Through the summer and the fall, we had each other that was all
Just she and I together, like it was meant to be
And when she was lonely, I was there to comfort her
And I knew that she loved me.

So the years went by, I stayed the same
But she began to drift away, I was left alone
Still I waited for the day, when she'd say I will always love you.

Lonely and forgotten, never thought she'd look my way
And she smiled at me and held me, just like she use to do
Like she loved me, when she loved me.

When somebody loved me, everything was beautiful
Every hour spent together, lives within my heart
When she loved me.

RARE DISEASE DAY

Every year, 28th February marks the International Rare Disease Day organised by Eurordis, when we focus on raising awareness both in the UK and internationally for all rare diseases including Batten disease. This year's theme was Research and how through research we have hope. With the support of Abeona Therapeutics, we saw the BDFA logo and that of many of our international partners in this fight, displayed in Times Square, New York on the NASDAQ Tower.

We ran a campaign throughout the day, with families sharing photos of children on social media to raise awareness of the faces of this disease and what it means for families every day and not just on 28th February. The posts saw traffic on our social media platforms reach nearly 20,000 people.


© Findacure, Photographer Barbara Asboth

Heather Band, BDFA Scientific Officer attended two events marking Rare Disease Day in London and Birmingham. "From Bench to Bedside: new treatments for children with rare disease" was hosted by the British Paediatric Surveillance Unit (BPSU) in collaboration with Birmingham Children's Hospital on 27th February and the following day she attended the Findacure conference in London.

In Birmingham, Dr Larissa Kerecuk (Rare Disease Lead, Birmingham Children's Hospital NHS Foundation Trust) discussed their work at the hospital including details of the world's first Rare Disease Centre for Children due to open at Birmingham Children's Hospital in December 2017. Professor Sara Mole (UCL) presented "The neuronal ceroid lipofuscinoses or Batten disease" to an audience of clinicians, researchers, policy makers, and most importantly, patients and their families. Many charities were present and there were plenty of opportunities to network.

Findacure is a UK charity, which aims to help and build the rare disease community, to drive research and develop treatments in rare diseases (**www.findacure.org.uk**). Their conference programme was diverse and interesting with a total of fifteen speakers sharing their perspectives on drug repurposing. These included talks from patients, clinicians, patient organisations, charities, researchers and representatives from the pharmaceutical industry all focused on exploring the best way to deliver repurposed drugs to patients with a rare disease.

Both days highlighted the challenges in rare disease research. A recurring theme was the importance of ensuring patient relevant outcomes and patient reported outcomes are central to future clinical trial development. All those attending were very positive – there is now the potential to take research forward to make a real difference for patients with many rare diseases.

'Improving Diagnosis, Care and Treatment for Childhood Dementia' Parliamentary Roundtable

On 14th December 2016 the BDFA alongside clinicians and other patient groups, was invited to attend a Parliamentary Roundtable at Westminster to discuss the care and treatment of children diagnosed with rare diseases which cause childhood dementia.

A number of rare and ultra-rare diseases cause childhood dementia including Neuronal Ceroid Lipofuscinoses (NCL, commonly known as Batten disease), Mucopolysacchardioses (MPS), Leukodystrophies and other metabolic disorders.

The event was hosted by Ben Howlett MP, who is the Chair of the All-Party Parliamentary Group on Rare, Genetic and Undiagnosed Conditions and was attended by Andrea West (BDFA), Mike Bewick (Former Deputy Medical Director of NHS England), Jim Shannon MP, Alex Chalk MP, Charlotte Roberts (MPS Society), Tony Heffernan (Bee for Battens), Dr Paul Gissen (GOSH and UCL), Dr Ruth Williams (Evelina), Baroness Finlay, Professor David Nutt (Edmond J Safra Chair in Neuropsychopharmacology at Imperial College London).

During the event, Andrea gave a presentation detailing the lifelong journey of UK families living with Batten disease from diagnosis through bereavement. She highlighted the challenges with early diagnosis and the obstacles that families face to get timely, quality and appropriate care for their children throughout their lives. Dr Paul Gissen also outlined the research needs of these groups of diseases and in particular the ongoing enzyme replacement trial at Great Ormond Street Hospital for children with CLN2 (Late Infantile Batten disease).

The event was designed to achieve a number of aims:

- To strive to ensure relevant policy takes into account the gravity and urgency of childhood dementia
- To ensure rare disease-related actions and national rare disease plans include measures aimed at improving the diagnosis, management and care of these severely debilitating and fatal conditions
- To ensure new therapies addressing these diseases need are urgently and adequately supported and EU research funds are maintained
- To develop reliable clinical care pathways to support patients and their families which are easy to implement by clinicians
- To develop expert reference centres across Europe and allow patients from across Europe to access them.

Tom Wishart, PhD, Division of Neurobiology at the Roslin Institute, University of Edinburgh

The Roslin is probably most famous for Dolly the Sheep but can you tell us more about the Institute?
Most people know us for Dolly, but the Roslin is a very interesting place to work, with many research interests, including Neuroscience. I was recruited to diversify the work going on in that field in early 2012. The Roslin is a part of the College of Medicine and Veterinary Medicine within the University of Edinburgh.

When did your interest in the NCLs start?
The Roslin has international expertise in genetic engineering and this has led to us working on new models for lysosomal storage diseases, including Batten disease. We are developing a sheep model for PPT1, CLN1 Batten disease, and are currently waiting to see if this will be a good model for studying the disease.

What do you see as different in your approach to NCL research?
My PhD student, Maica, is working on CLN1 and CLN3 funded by the Darwin Trust. She is looking at overlaps at the molecular level between these diseases, the idea being that we look for common factors that are affected. In neurodegenerative disease, one feature is the effect on synapses, which are very important for communication between neurons. So we look at these in NCL models to see the processes taking place. Targeting the changes correctly can benefit the whole neuron. Hopefully this approach will find something to benefit all forms of Batten disease.

As a relative newcomer to the NCL research community how are you finding this?
Luckily I have some funds to spend on this research. Government investment in dementia research has mainly focused on those of advancing age, but we are promoting the cause of childhood dementia.

We have been supported by researchers in the field, such as Prof Jon Cooper, who provided tissue samples from models he was already working on. We used these to do something novel. Attendance at meetings is a good way to make contacts – until you put a name to a face you cannot really know if you can form a collaboration. Recently Prof Sara Mole has helped us with our projects.

You attended our family conference in Stratford-upon-Avon last October.
Yes, I really enjoyed meeting families at the conference. It is important for students to see those affected, to ground them again as to why we are doing this research. To see what families deal with on a daily basis reminds them that their work is not just scientific drive and that there is an end goal.

What do you like doing outside of work?
We have a house that is a real "fixer upper" and needs a lot of work so DIY most weekends! Time with the family – taking my kids to mini rugby and swimming.

What are your plans for the future?
We are waiting to see how CLN1 sheep model the disease but this could take a while. We hope that this will be a good preclinical tool. The Roslin is ideally placed as we are on the Vet School campus so can move seamlessly from species to species. This has its challenges, and the willingness of people in the group to work hard and acquire the different expertise needed is what has led to our current success and will allow us to take this forward in the future.

Photo Laura Graham (PhD student), Tom Wishart PhD, Maica Llavero (PhD Student)

DEM-CHILD

CLN2 Registry Summit

In January 2017 the BDFA, along with its US partner the BDSRA, was invited to provide the patient perspective at a CLN2 Registry Summit run by BioMarin Pharmaceutical in Barcelona. The summit was also attended by leading clinicians from across the world and was an opportunity to discuss the importance of disease registries, what data they should contain and how they should be maintained. The BDFA sent out a short online survey to CLN2 families and then presented the results from the survey to the summit. The clinicians were very interested in the family perspective on registries, what information was important to them and the value to families of contributing their children's data.

Why do we need registries?

Having accurate and up to date information is vital for the successful clinical trials and to better understand what is happening, in the disease. This can lead to more effective care and treatments and help with improved methods of diagnosis.

How can you help?

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

Evelina London Children's Hospital

Guy's and St Thomas' NHS
NHS Foundation Trust

Dear Parents/Guardians,

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

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

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

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

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

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

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

Yours sincerely,

Dr Ruth E Williams,
Consultant Paediatric Neurologist,
GMC 3057036

Congratulations to Andrew and Sarah Dawkins, their families and supporters for their amazing fundraising efforts, which have enabled the BDFA to fund two research projects in CLN5 disease on their behalf. The projects are both progressing well and the latest news from UCL and Cardiff University is featured below.

BDFA
BATTEN DISEASE FAMILY ASSOCIATION
Together we WILL make a difference
Registered Charity No. 1084908

Development of a drug screen for CLN5 Batten disease

The group at UCL (Dr Dan Little, *pictured*, Professor Paul Gissen, Professor Sara Mole, and Dr Robin Ketteler) has used patients' skin cells, which have the mutation or "mistake" in the CLN5 gene to create a type of cell (iPS cells) that can then be turned into nerve cells (neurons) which are the cells that die in CLN5 disease.

Dan has successfully turned the first of the iPS cell lines into neurons. Work is well underway to look for significant differences between these CLN5 cells and healthy cells to exploit in a drug screen. Drug libraries can then be tested to find compounds with potential to make the CLN5 cells healthier. The new cell lines also represent a valuable resource for further study into CLN5 disease.

UCL

Looking at what goes wrong in CLN5 Batten disease

Dr Emyr Lloyd-Evans and Katie Shipley (PhD student), *pictured,* at Cardiff University are working to identify key differences in cells derived from CLN5 patients compared with healthy unaffected cells. The aim is to find what goes wrong first and then to look for potential treatments.

"Our project focuses on identifying key differences of CLN5 patient cells compared to healthy cells to try to understand which change happens first in the cell. If we know what these are, we can try and treat this to prevent other changes happening which lead to the cell not working properly." Katie Shipley

Emyr's group has special expertise in studying the role of calcium within the cell. Calcium is not just an important mineral for strong bones; it has a vital role in sending messages between different parts of the cell. Too much or too little calcium can cause a great many problems and have a very damaging effect on the health of the cell.

The results so far indicate that there are differing levels of calcium in several cell compartments and Katie is now looking at this process in more detail. An existing drug has also been identified that improves these harmful effects in the CLN5 cell model. Katie will continue to investigate these very promising areas in the next two years of her PhD.

CARDIFF UNIVERSITY
PRIFYSGOL CAERDYDD

The National Institute for Health and Care Excellence (NICE) is the body in the UK which specifically evaluates the funding of rare disease drugs through their Technology Appraisal and Highly Specialised Technologies programmes. Recently, along with NHS England, they launched a consultation on proposals for changes to these evaluation and funding arrangements. In summary the proposals were as follows:

- To introduce a 'fast track' NICE technology appraisal process for the most promising new technologies, which fall below an incremental cost-effectiveness ratio of £10,000 per QALY (quality adjusted life year), to get these treatments to patients more quickly.
- To operate a 'budget impact threshold' of £20 million, set by NHS England, to signal the need for a dialogue with companies to agree special arrangements to better manage the introduction of new technologies recommended by NICE. This would apply to a small number of technologies that, once determined as cost effective by NICE, would have a significant impact on the NHS budget.
- To vary the timescale for the funding requirement when the budget impact threshold is reached or exceeded, and there is therefore a compelling case that the introduction of the new technology would risk disruption to the funding of other services.
- To automatically fund treatments for very rare conditions (highly specialised technologies) up to £100,000 per QALY (5 times greater than the lower end of NICE's standard threshold range), and provide the opportunity for treatments above this range to be considered through NHS England's process for prioritising other highly specialised technologies.

Whilst we recognise that we have a health system which is under pressure and has limited resources and we also recognise that the pharmaceutical companies need to be mindful of the cost and accessibility of the treatments they develop, these new proposals would mean that none of our potential treatments would be routinely funded and would be considered under a process that lacks transparency and accountability. These changes would impact on the availability and access to future treatments for children and young people with ultra rare diseases, so alongside our close colleagues in the Lysosomal Storage Disorder Collaborative, we took part in the consultation.

We gave our informed opinions on how these treatments should be funded in the future.

We await the outcome of the consultation and will update our membership through our website, social media pages and direct email of the outcomes.

We hope you enjoy the first edition of the BATCure Newsletter which is enclosed as an insert. BATCure is a 3-year research project with the goal of advancing the development of new treatment options for patients living with CLN3, CLN6 or CLN7 Batten disease. The BDFA are leading part of the project and will be launching an online Family Survey in Spring/Summer 2017. It is really important that as many families as possible participate and we hope that you will be able to take part. Further details can be found on the back of the BATCure Newsletter.

For any BATCure enquiries, please contact Laura Codd BATCure Administrator at the BDFA:
lauracodd@bdfa-uk.org.uk

www.batcure.eu **BATCure** **BAT_Cure**

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

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BDFA Family Conference

24th-26th November 2017

Crowne Plaza, Stratford Upon Avon

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