BDFA

BATTEN DISEASE FAMILY ASSOCIATION

BATCure

AUTUMN 2016

making the difference

Contents

Diary Dates

NCL 2016
Boston, USA
5-8 October 2016

Kidz to Adultz up North
EventCity, Manchester
17 November 2016

BDFA Family Conference
Crowne Plaza, Stratford-upon-Avon
18-20 November 2016

"Examining the Utility of Music Interventions for Children with Learning Disabilities" RSM Conference
Royal Society of Medicine, London
28 November 2016

Kidz to Adults in the Middle
Ricoh Arena, Coventry
16 March 2017

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,

I am writing this piece at a research conference and I am very aware that until recently, the programme of these conferences contained either nothing or very little of relevance to the NCL community. It is exciting that now we are seeing a real and hugely welcome change firstly for our CLN2 families, but also on the treatment horizon for other forms of the disease. In this edition of our newsletter you will read about some of these developments and the work of the BDFA in driving the research agenda both in the UK and worldwide towards treatments for all.

Whilst research is ongoing, the need for family support continues to be at the heart of what we do and I firmly believe that we are a small charity that makes a real difference to the families and professionals we support. I hope you will see some of those stories and their impact in the articles in this edition.

Inside we also share some hellos and goodbyes. We say hello to our new Batten disease Clinical Nurse Specialist, Laura Lee from Great Ormond Street Children's Hospital. We welcome Laura as a new and important member of the team and I am sure many of you will be in touch with her in the near future. Sadly we say goodbye (in the UK) to a dear friend and leading NCL researcher, Professor Jon Cooper who moved, in the summer, with his family to take up a new post at UCLA. Whilst this is a loss to the UK NCL community, Professor Cooper will continue his NCL work in the US, and remains an integral part of our global community. We all wish Jon and his family well for their new start.

As always, we hope you enjoy reading this latest newsletter and we would love for you to share your copy with friends, families and wider community. The more people who are aware of this disease, the louder our voice for better services, treatments and a world without Batten.

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

Letter from our Chair

Another summer has passed by and wherever you may be my thoughts remain with all those families who are trying to cope with the enormity of dealing with a child with Batten or indeed those that have lost a family member this summer. Whether you have just had a diagnosis or whether you have been living with the disease for ten years, we all share the same thoughts and understanding and the BDFA continues to be available to you to support you on your journey. I know from my own experience, and especially from this summer, how hard it is and I really encourage you to tap into all the support and services that the BDFA can provide you with.

I'm biased because I love cricket and I've always felt an enormous sense of achievement whenever I've been involved in a BDFA fundraiser, so the highlight of the summer for me was the BDFA lunch at Lord's Cricket Ground which was so kindly and generously arranged by Keith Banks. On a beautiful July day, 150 people enjoyed a great lunch and were entertained by Bob 'The Cat' Bevan and our patron and English cricket supremo, Andrew Strauss. In addition, the lunch had a serious part to it and everybody who attended will never forget the 'Hey Charlie' video or Ellen Bletsoe's moving speech on her own experiences of living with a daughter with CLN3 and what this journey has felt like to her. The lunch raised over £40,000 and I would like to say a huge thanks to Keith, Andrew, Bob and Ellen and all those involved in the making of 'Hey Charlie' for all they did to make the lunch such an enjoyable and poignant occasion. Whilst all of these people made this event, it wouldn't have been possible without Gaynor at the BDFA who lived this lunch for several months, so another huge thank you to her for all she did and continues to do in support of our fundraising efforts. Awareness continues to spread!

Finally, I would also like to say a thank you and a goodbye (for now) to Jon Cooper for his tireless work and support of the BDFA. Apart from being hugely knowledgeable, he is also wonderfully personable and I have always found being in his company highly pleasurable. We will miss you as I'm sure will all the other season ticket holders at Southampton! Good luck Jon and hope to see you again soon.

Michael O'Connor, Chair of the Board of Trustees
michaeloconnor@bdfa-uk.org.uk

New BDFA Trustees

We are delighted to announce that five people are planning to stand for election as BDFA Trustees at our AGM in November. The election will take place at our 2016 Family Conference when you will have the opportunity to vote them onto the BDFA Board. Find out a little more about some of them below:

JAMES JEYNES

James is father of Lewis Jeynes, who was diagnosed with Batten disease in 2014. For many years Lewis remained undiagnosed and James hopes to make people more aware of Batten disease through his work with the BDFA. He is Chief Executive of MemNet and its Founder Member. For over 24 years he has been working within the membership and associations sectors as a senior manager. He worked in public sector development roles overseas in South Africa, France, Switzerland, UAE, Barbados, Jamaica and Trinidad. James has a passion for creating and developing businesses and is currently an Associate Director with Advantage Public Services, Director of Executive Office Ltd, Director of the Football Argument Ltd and a proud Fellow of the Royal Society of Arts. In his spare time James is a Trustee of The Lewis Jeynes Fund and an Advisor to the NHS England Personal Health Budgets Team.

TIM MARTIN

Tim Martin is Managing Director, North America Property for Guy Carpenter & Co Ltd. He had never heard of Batten disease until hearing BDFA Trustee, Ellen Bletsoe speak at an industry event in 2014. He was so inspired by this and the work of the BDFA that he felt sure the Insurance and Reinsurance community could help raise money. He therefore held highly successful fundraising lunch events in 2015 and 2016, which have raised significant funds for the BDFA and have played an important role in raising the profile of this fatal disease. He hopes these will continue to be an annual event for many years for the attendees and says "I feel very honoured to have done something to help such a great charity and to have met Laura, Ellen and Dr Mole. These events are really helping to raise the profile of this disease and we need more people to know about it".

ROGER COLE

Roger Cole worked in the British Telecom development department on remote computer management and control of equipment and retired in 2006. He has many years' experience writing technical specifications and seeing through contracts from tender to deployment. Roger is putting his skills to use in supporting the smooth running of the BDFA office. Roger has had involvement with BDFA through his wife Barbara Cole who works as an education advisor volunteer with the BDFA. Through this he has met a number of families around the UK. His leisure activities include narrowboating and travelling.

NEIL DUNFORD

Hello, my name's Neil Dunford. I have joined as a Trustee member of the BDFA this summer. My daughter, Yanna, has the CLN2 variant of Batten disease. Yanna has been enrolled on the BioMarin Clinical Trial since September 2014, which she started in Rome. We returned to England at the end of January 2016. Before her diagnosis, I worked in retail banking for several years. Since returning to London I have been developing a Befriending Network for parents with disabled children with Scope. I am interested in promoting this model within the BDFA, in order for parents of disabled children to have a coherent structure of support from fellow parents within the Batten disease community.

Welcome to our new Clinical Nurse Specialist

Hello! My name is Laura Lee and I am pleased to tell you that I will be starting the role of Batten Disease Clinical Nurse Specialist at the end of September.

I'm currently working as a Novel Therapy Research Nurse at the Somers Clinical Research Facility in Great Ormond Street Hospital. I have been working on lots of trials looking at potential new treatments for a variety of neurometabolic conditions. This includes working as the lead nurse for the BioMarin Enzyme Replacement Therapy Trial for patients with CLN2. This role has given me the opportunity to meet some of the families and professionals involved in Batten disease and the drive to be a part of this team.

Prior to this, I completed my degree in Children's Nursing at the University of Surrey. I started my career on a general paediatric ward which led me to discover my passion in caring for children with complex needs and their families. Before starting my current role, I worked as a paediatric nurse on a Neurology and Neurosurgery ward at Great Ormond Street Hospital.

I'm grateful for the opportunities that I have had in my career and hope to develop this role in collaboration with children and families affected by Batten disease. I am really excited about starting this new post and look forward to meeting lots of you at the BDFA Family Conference in November!

email: laura.lee@gosh.nhs.uk
020 7405 9200 ext. 0213

Education

In 2015, Education Health and Care Plans (EHC Plans) were brought in to replace Statements or Learning Difficulty Assessments for children and young people up to the age of 25. The new Plans span all areas of health, social care and education to form one document.

Children and young people with a Statement or a Learning Difficulty Assessment will be transferred to an EHC Plan by 1 April 2018. If your child or young person has recently been diagnosed they will receive an EHC Plan. If they do not have one, parents can write to the Local Authority to request an assessment. All children and young people with Batten disease should have an EHC Plan and your request should not be refused.

We know of many children and young people who have already been through the EHC Plan process. Unfortunately there have been a few Plans that have not had the required information and didn't contain enough information about Batten disease in them. Barbara Cole and Harriet Lunnemann can support you through the EHC Plan process and are happy to attend meetings, write reports or look at draft Plans to ensure that your children receive what they need. Please email **support@bdfa-uk.org** for more information.

The BDFA has also produced a leaflet on EHC Plans which will be available at the Family Conference in November and on the BDFA website. It breaks down the EHC Plan into the relevant sections and gives an explanation about what should be written into each different section.

Background

The aim of this study was to look at the challenges that families face when looking after a child or children affected by CLN2 disease. The study was made up of two phases. Phase 1 involved focus groups in the UK. Phase 2 involved home surveys in the UK and Germany.

Phase 1 – Focus Groups

In May 2015, a Focus Group Research Day was held in Sheffield, UK with a mixture of current and bereaved caregivers. Eleven adults from six different families took part in the focus group sessions, and one child sibling took part in an interview. The aim of the focus groups was to learn about the challenges faced by families to inform the in-depth family surveys planned for Phase 2.

Phase 2 – Home Surveys

Families whose child was not taking part in a clinical trial, were invited to the study by the BDFA in the UK and by the NCL Gruppe e.V, and University of Hamburg-Eppendorf in Germany. Between September 2015 and February 2016 families in both countries took part in the survey. Nineteen families in total took part, with one to three participants from any one family. This included parents, partners of a parent, grandparents and unaffected siblings (adult and child siblings).

Study researchers visited the families at their home or other quiet place. Interviews were conducted with one family member at a time and included open ended questions and multiple choice questions, as well as validated questionnaires.

BIOMARIN

Findings

1. Diagnosis and Symptoms

- Early symptoms of CLN2 disease (e.g., seizures, developmental delay) often lead to initial misdiagnosis of epilepsy.
- Caregivers reported frustration and anger related to their fight for a correct diagnosis, which could take as long as two years.
- Symptoms of the children at the time of the survey included: seizures, difficulty sleeping, and problems with/lack of movement, vision, and/or communication.
- Caregivers mentioned that symptoms such as loss of sight were difficult to confirm as they were unable to communicate with their child.
- The affected children were prescribed a large number of medications to help manage their symptoms.
- Some caregivers mentioned that it was hard for them to tell the difference between symptoms of CLN2 disease and the side effects of the medications they gave their children.

2. Education

- All affected children are or have been in education, except for one child from Germany.
- Most affected children attended special needs schools where they received one-to-one support throughout the day.
- Many caregivers experienced difficulties in getting access to the special needs schools or necessary support in mainstream schools.
- Attending school was seen more as providing social experience and stimulation, rather than for educational reasons.
- Caregivers particularly reflected on sensory rooms as a positive experience for their child.

3. Healthcare Professionals (HCPs)

- Caregivers were generally very positive about HCPs providing care and support.
- Several caregivers reported that the quality of care and support from HCPs improved after their child was diagnosed.
- Most caregivers had different experiences with different HCPs, reflecting individual differences in HCP personality, experience, and communication style.

4. Level of Care and Support

- Generally, caregivers were satisfied with the current level of care and support their affected child received.
- A few caregivers felt that some services needed the option of being fast-tracked when a need for something arose.
- Several caregivers reported how the quality of support and care they received from HCPs differed between individuals, and this was linked to their familiarity with CLN2 disease.
- Negative experiences with HCPs related to seemingly low levels of interest taken in the child and the family, not being listened to with care or understanding, and frustration with communication style.
- In general caregivers were less satisfied with social services, compared to medical and educational services.
- Care and support needs mentioned included: care packages designed around the nature of CLN2 disease and access to 24/7 nurse support, as well as support for professionals involved in the care of their child to better predict and plan for the next steps.
- Examples of services that the caregivers thought were missing included: being able to get correct diagnosis sooner, support to cover care needs during the night, someone to coordinate all the care and support services the families need, and advice and care services to guide the caregivers through the process immediately before and after their child passing away.
- Caregivers, particularly in Germany, mentioned that accessing appropriate support required a lot persistence and knowledge about the relevant services.

5. Caregiver Well-being

- Adult health-related quality of life was lower in UK and Germany compared to the general population.
- The majority of caregivers reported at least slight problems with pain or discomfort, and anxiety or depression. About a third of caregivers reported at least slight problems with usual activities.
- Compared to the general population caregivers overall also reported lower life satisfaction and lower happiness with their partner. Caregivers reported or recalled more hours on caregiving each week (on average 73.45 more hours per week) and had fewer hours sleeping per night during the last month compared with parents with a child of the same age from the general population. (1.32 fewer hours sleeping per night).
- Overall, caregivers' level of comfort with their current financial state was no different compared to parents of a child of the same age.
- Caregivers of children in the severe stage of CLN2 reported a greater number of hours caring, and less sleep than caregivers of children in earlier stages and bereaved caregivers. Bereaved caregivers still had slightly less sleep compared to the general population (1.19 fewer hours sleeping per night).
- While overall caregiver happiness lowered in relation to the stage of their child's disease, with bereaved caregivers reporting the lowest level of happiness, life satisfaction was similar across stages.
- Caregiver quality of life measured by two questionnaires was lowest for those caring for a child or children who were in the more severe stage of the disease.

Conclusion

Across the course of caring for a child or children with CLN2 disease, families cope with many difficult emotional, physical, professional, financial and organisational challenges, including: coping with the shock of diagnosis, the often exhausting impact of caring on sleep, physical health and family relationships, the financial impact of having to stop working to care for their child as well as pay for care equipment and home adaptations and to deal with funding and care support. Despite the many difficulties they deal with, caregivers reported they simply need to accept the situation as it is and deal with it for the sake of their children.

Impact

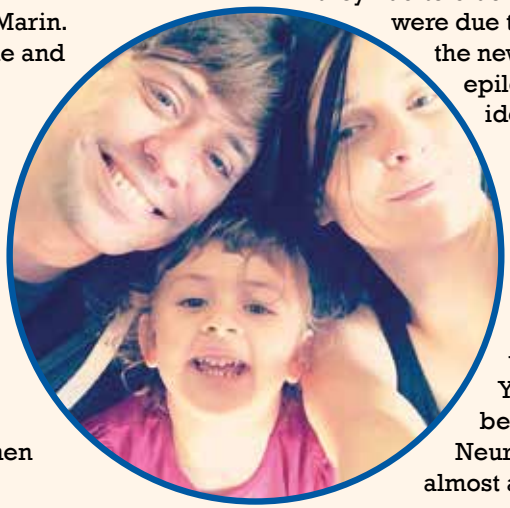
Results will be presented by Andrea West at the NCL Congress in Boston, USA in October. We also plan to make results available to families at the BDFA conference in November and to the scientific community in a journal publication. We wish to thank ICON plc, BDFA, NCL Gruppe e.V. Klinik und Poliklinik für Kinder- und Jugendmedizin, Universitätsklinikum Hamburg-Eppendorf and the families who gave up their valuable time to take part in this study.

Thomas Butt
Associate Director HEOR, EUMEA region
BioMarin Europe

At the end of January 2016, we landed at London Heathrow. We had just spent 17 months living in Italy where our daughter, Yanna, was enrolled on an experimental Enzyme Replacement Therapy, sponsored by a pharmaceutical company named BioMarin.

Since returning to England, Yanna has continued to receive fortnightly infusions, directly into the cerebrospinal fluid surrounding the brain. She is usually sedated for the whole infusion, lasting four hours. This treatment will hopefully continue for the rest of Yanna's life.

There are currently four trial sites around the world delivering the treatment on behalf of BioMarin. Hamburg opened first, then London, Rome and finally Ohio. When Yanna was diagnosed with CLN2 Batten disease in June 2014, the London site had been temporarily suspended and wasn't accepting new children.



So, at the beginning of September 2014 I got a sabbatical from work, we packed what we could into three bags and flew to Italy, expecting to stay for one year. In the end we stayed for 17 months. Even now, there are many relocated families in both Rome and Hamburg with no idea when they can return to their home countries.

"But no matter what happens, no matter how long Yanna lives, no matter what the future holds, with Batten disease it's always ever going to be one day at a time."

It's impossible to describe how surreal it is, coming to terms with the diagnosis of Batten disease, in a foreign country where you don't speak the language. It was an isolating experience.

Similarly, there's little point attempting to describe how draining it is; seeing your child repeatedly undergoing invasive medical procedures, often once a week, not knowing if it was doing any good, or if we were simply depriving her of the little childhood she had left.

For Batten parents, words lose meaning. Trying to describe how you feel is like shouting into a void. It's not that people can't imagine how painful it is for us, that's easy; the problem is that we can no longer imagine what it's like to live without the pain. On the day your child is diagnosed with Batten disease you become alienated.

I've thought back to the day of diagnosis a lot. We had been called urgently into Leeds General Infirmary. A week earlier, they had told us Yanna had dyspraxia and the seizures were due to epilepsy. I had felt almost elated at the news; we could deal with dyspraxia and epilepsy. There were books on it. It wasn't ideal, but it was manageable.

I remember the Neurologist telling us they had received the final test results. Apparently there was an enzyme missing in Yanna's brain. The seizures would increase. Yanna would lose her motor functions. She'd go blind. It wouldn't happen straight away. By five Yanna would be in a wheelchair. She'd be bedridden by ten. At twenty? The Neurologist seemed to shrug her shoulders almost apologetically.

She gave us a printout with some information about the disease and the contact details of the BDFA. No follow up appointment, no crisis counselling. Just a website and telephone number for an obscure charity we'd never heard of. She told us to call them about a clinical trial.

And now, two years later, the results are in. Apparently they far exceed BioMarin's expectations. The doctors are even talking about Yanna's life in terms of decades. That's pretty big. But no matter what happens, no matter how long Yanna lives, no matter what the future holds, with Batten disease it's always ever going to be one day at a time.

Neil Dunford

Together for Short Lives

Hearing the news that your child is likely to die young is devastating. It's an incredibly distressing and confusing time. For tens of thousands of families in the UK this is the reality.

Together for Short Lives is a UK charity that speaks out for all children and young people who are expected to have short lives. Along with everyone providing care and support to these children and families, they help them have as fulfilling lives as possible and the very best care at the end of life.

www.togetherforshortlives.org.uk hosts a wealth of information for families about the help and support available to them. Each family is unique and everyone's journey will be different. *The Family Journey* section provides information and advice to help every step of the way and answers to common questions received through the Helpline are shared under *Your Questions Answered*. The website also hosts free resources to download or order to help families get the most of their local services and enjoy their time together as a family.

> "Getting in touch with Together for Short Lives and using the resources on the website made me realise I was not alone."

Any family caring for a child with a health condition that may shorten their life can join Together for Short Lives' family community. Members receive a quarterly newsletter full of news and updates on policy, services and resources. They are also invited to join reference groups to help influence change where change is needed.

James has Infantile Batten disease and his mother, Amanda is a member of Together for Short Lives' family community. She says:

"Finding out your child has a life limiting condition is devastating and can be incredibly isolating. You feel like you're the only family going through it and I often asked myself 'why us?' Many friends and family don't know how to react so simply avoid you. Getting in touch with Together for Short Lives and using the resources on the website made me realise I was not alone. Reading the family stories gave me comfort and I realised there were many families experiencing the same issues we were. Becoming part of Together for Short Lives means we can support each other and we become a bigger voice. On our own we are lost in a system that at times fails our children, but together we are bigger and louder and our voice is heard so that change can happen."

If you have any questions or would like to request a publication, please call the Together for Families Helpline on **0808 8088 100** (10am - 4pm Monday to Friday) or email **info@togetherforshortlives.org.uk**

together for short lives

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.**

Amber Louise Jutla

29th January 2011 ~ 23rd February 2016

Our precious Amber, just five years old. In those five short years, she taught us more than we could have learned in a lifetime.

A girl unable to speak, taught me to listen
A girl who could not reach out and hold my hand, deeply touched my heart
A girl who lost her sight, opened my eyes to true love
A girl who could no longer walk, led me down the path to understanding the value of life
A girl who could not voluntarily move her body, moved my soul
A girl who could no longer laugh, showed happiness in her eyes and bought joy to so many
A girl who showed true dignity, bravery and grace that touched so many hearts
A little girl I am proud to call my daughter.

Amber Louise Jutla, the girl who stole our hearts to heaven. Her infectious smile will forever be imprinted in our hearts. Sleep tight and sparkle bright.

Zac Robinson

13th April 2010 ~ 29th March 2016

To our gorgeous boy Zac,

Writing this you are now hopefully sleeping peacefully and are on your way to joining other stars in the sky. When you were born I never imagined that I would be writing this for you but you have given us so many memories and ones that I will certainly treasure forever. You always had your own agenda or 'Zac timing' and you certainly did things your own way right from the very beginning but everyone who met you fell in love with you and I think you were the most hugged and kissed boy that I have ever known. Although you couldn't speak, your eyes told me what you thought and how you were feeling and I will miss so much those gorgeous and beautiful sparkly green eyes which lit up when you knew we were cuddling and talking to you.

Daddy and I will miss you so much. Despite your smiles and happy demeanour, we hated to see you in pain or how tired you got after having a seizure. The last thing we wanted to do was say goodbye but at least we know you are now free of pain and suffering. Both of us will forever talk about you, and together with Molly, we will make sure that your new baby sister Sophie grows up knowing she has the most amazing big brother.

Sleep tight my gorgeous boy and whenever we see a rainbow or the stars twinkling in the sky we will remember all the happy memories you were able to share with us as one big happy family.

All our love forever,
Mummy and Daddy

Caitlind MacPhail

17th November 1991 ~ 31st March 2016

Poem for Cait

"Are we there yet, are we there yet,
Are we there yet?" she says,
A wicked sense of humour,
On a cheeky smiley face,

A little bit of insight,
Is what I want to give,
On a little girl of 24,
Who really tried to live.

Sometimes grumpy, or in pain,
You really couldn't tell –
She looked at you with smiling eyes,
She hid it really well.

A look that would say "yes I'm fine,
I'm watching Jeremy Kyle"
She laughed when they were shouting –
You see that was her style.

Her big blue eyes, or were they green?
They sometimes changed you know.
There was always something funny,
That made her face to glow.

Get on your nerves,
Get on your nerves,
Get on your nerves,
You see,

A little song,
From Joe Pascale,
With an added touch
From Cait and me.

Sitting in her wheelchair,
She loved to take the mick,
"Who's your favourite uncle?"
She liked to play a trick.

Her Disney films she use to watch
Although she could not see.
She'd listen so intently,
She saw more than you and me.

She loved to shop for everything,
Though I think this was her mum,
Her favourite was Build-A-Bear
with 10, 20, 30, and then some!

The cinema was a favourite treat,
With all her playback friends –
The shows, the bowling, and
the swimming,
The fun it never ends.

The music that she listened to,
It really did complete her.
The songs were very varied,
Like the purple people eater.

A princess who loved pink and purple
They were her favourite hue.
The sky that day, it turned that shade,
To say they had received you.

Her fairies were a favourite thing
Of which she had a few.
And now she has a garden
for mum and dad to view.

"Are we there yet, are we there yet,
Are we there yet?" she says.
I know you're there now, my special girl,
In your special place.

My goddaughter, my princess,
My very special niece,
My love for you will never wane,
May you rest in peace.

Tia Kerry Crouch

26th January 2003 ~ 11th February 2016

Long dark hair, big brown eyes with rose bud lips,
a button nose. You sure are the brightest star
to enter the sky.

An angel on earth to an angel up above.
Watching us all, we miss you with every piece of love.

Pinkies were your favourite treat, sausages you loved to
eat and 'more mushrooms' we would hear you say,
mushrooms every day.

Big brave girl, every step of the way. On your journey
in life, you gave the biggest fight.

My big sister, my best friend to the very end, and for
eternity you will remain in our hearts. We will meet again.

With no more tubes and suction machines, you have gained
your angel wings. Fly high and be free to play again.

This song was written by Tia's sister Jessie, aged 9.
She wrote the song while at Chestnut Tree House Hospice
for Tia's funeral.

Tia Kerry Crouch was born on 26th January 2003 and gained her angel wings on 11th February 2016. Always in our hearts, Princess Tia. We will miss and love you forever XX

Amelia Rose Roberts

18th March 2005 ~ 17th March 2016

My Angel up in Heaven

My angel up in heaven, I wanted you to know,
I feel you watching over me, everywhere I go.
I wish you were with me, but that can never be,
Memories of you in my heart, that only I can see.

My angel up in heaven, I hope you understand
That I would give anything if I could hold your hand.
I'd hold you oh so tightly, and never let you go,
And all the love inside of me, to you I would show.

My angel up in heaven, for now we are apart.
You'll always live inside of me, deep within my heart.

Long-time Batten disease researcher and BDFA Scientific Advisor
summer to take up a new post at the Harbor UCLA Medical Center
predominantly on Batten disease, and his new post will allow him t
academic activities.

Jon's journey started as a graduate in Anatomy & Cell Biology from
Neuroanatomy at Bristol (1990). He writes:

*"I had a phone call in the summer of 1996 from someone I knew
work on a disease you've never heard of?" which turned out to
shortly after I arrived when I met my first Batten family, Liz and
them I learnt just what Batten disease is and how it impacts a fa*

A move back to the UK and an opportunity at Kings' College
Laboratory at the Institute of Psychiatry in April 2000. Jon cor
conference in 2000.

Jon writes about his favourite moments with the BDFA:

*" A personal highlight was being presented with the wonderful pi
summer. The sponsored walks have always been a highligh
message that can't be ignored, and a real chance to wa
PSDL open days has also been very important in giv
to day basis, and communicate the science behin
been especially important in helping the many
understand the human side of the disease, and
real people. The annual family conferences h*

*Being part of the NCL2012 organising comm
conference was also a great moment, and wa
together. It has been fantastic to see the BDF
highlighted by attending the opening of the
to California is the next step."*

**On behalf of all its members, the BDFA wo
years. We know that we will continue to work
to Jon and his family for their new venture, the**

Professor Jonathan Cooper left King's College London this
in Los Angeles. As he did in London, Jon will continue to work
o concentrate on his research rather than teaching or other

Sheffield University in 1986 followed by a PhD in

in San Francisco who basically said "Do you want to come and
be Batten disease. I accepted and the real turning point was
Dave Aureolio from San Jose and their daughter Natalie. From
amily and I decided that this is what I needed to work on."

London saw the beginnings of the Paediatric Storage Disorders
ntinued to meet UK families and attended his first BDFA

cture of families from the BDFA just before I left London this
ht, with the snaking ribbon of orange shirts sending out a powerful
lk, talk and listen to parents' thoughts. Establishing our annual
ing a chance to show parents what we actually do in a lab on a day
d it in a way that everyone can (hopefully) understand. These have
wonderful people who have worked in my lab over the years to
that it's not just a name in a book or scientific paper, but involves
ave always been special, and reinforced this message.

ittee and working to set up such a big science and family
s another great example of scientists and the BDFA working
FA grow over the years to the position it is in today. That was
BDFA's first office last year. It's been an amazing journey... returning

uld like to thank Jon for his fantastic support over many
k with him closely in the future on NCL projects. Best wishes
e PSDL lab in LA.

LA BioMed
Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center

Gene Therapy to Treat Visual Failure in CLN2 disease

The BDFA is delighted to welcome Dr Mikel Aristorena, who has recently started work in Professor Sara Mole's lab at the LMCB, UCL. Mikel will divide his time between UCL and the Institute of Ophthalmology (IOP) on a 3-year project funded by the BDFA and the Wellcome Trust.

"I'm 33 years old and I'm from Spain. Although this is my first contact with Batten disease, the focus of my research has always been in rare diseases. There are currently two clinical trials to correct the deficiency of CLN2 in the brain but there is no study focusing on the loss of vision. The aim of my project is to address this gap by developing a gene therapy treatment for the loss of vision in CLN2 disease. Currently I'm testing new potential vectors – ways to deliver a functioning (good) copy of the affected gene – to the place where it is needed in the eye, in my model system."

This work builds on the very successful BDFA-funded project on "Gene therapy to treat visual failure in CLN3 disease" and Mikel joins Sophia kleine Holthaus in the group headed by Professor Robin Ali. Our thanks go to Dr Sander Smith and all the members of the IOP for their continuing support to the BDFA, and the NCL projects.

Announcing a new CLN8 research project for 2016–2019

The BDFA is delighted to announce the award of £33,000 to Dr Emyr Lloyd-Evans for a three year PhD studentship. This is a joint venture between the BDFA and Cardiff University, with matched funding from the Life Sciences Research Network Wales.

CLN8 has challenging biology, as the exact function of the protein remains unknown. It is believed to have an effect on possibly two other important structures within the cell (not just the lysosome). There is a major unmet research need and CLN8 is under-represented in current active research worldwide. Potential experts on CLN8 need to be engaged and new groups attracted to the field. This project aims to do this with collaborators, including experts from other lysosomal storage diseases and some of our colleagues from the BATCure consortium (see page 19).

Rafael Andrés Badell-Grau will begin his studies in October on the project *'Characterising the fundamental cell biology of CLN8 disease for the purpose of developing drug screening strategies and potential therapies'*.

"I have always had great interest in pharmacology and neurological diseases. Whilst studying towards my degree in Biochemistry, my interest was focused onto lysosomal diseases, which I then took forward and expanded during my Masters in Research where I worked on the role of lysosomal dysfunction in Huntington's disease in Dr Emyr Lloyd-Evans' Laboratory at Cardiff University.

Using the knowledge and skills I have learnt during the Masters I am looking forward to starting my PhD at Cardiff University with Dr Lloyd-Evans, exploring and expanding our knowledge of the mechanisms behind CLN8 disease. What are the key differences between healthy cells and CLN8 cells? Based on our findings we will then consider if these key events and differences are potential therapeutic targets and how they could be exploited to make an effective drug screen."

Rafael Andrés Badell-Grau

British Society of Gene Therapy

On Saturday 14 April Heather Band, BDFA Scientific Officer attended the British Society of Gene Therapy satellite meeting on Gene, Cell and Molecular Therapies for Inherited Metabolic Disease at the Institute of Child Health in London.

Scientists and clinicians from the UK and worldwide were invited to attend to exchange ideas on how to translate gene therapy and novel technologies into clinical treatment for this group of rare diseases. The BDFA exhibition stand attracted many visitors keen to learn about Batten disease and how the BDFA funds research. In the morning session on novel treatments in brain disease, it was really encouraging to see Batten disease so well represented, with three talks devoted to the NCLs.

"It was really encouraging to see Batten disease so well represented."

In the opening talk Prof Tammy Kielian, Professor of Pathology and Microbiology, from the University of Nebraska Medical Centre presented new studies from her laboratory describing the use of a gene therapy approach for CLN3 disease. Her work demonstrated that a vector or vehicle containing the human CLN3 gene was able to reverse motor deficits and attenuate inflammation in a CLN3 mouse model when delivered intravenously. This gene therapy has been licensed by Abeona Therapeutics and additional preclinical studies are being performed in Dr Kielian's laboratory with the goal of a future Phase I clinical trial to evaluate its safety profile in patients with CLN3 disease.

Professor Paul Gissen presented data from the Cerliponase Alfa clinical trial in CLN2-type Batten disease. Dr Sophia kleine Holthaus followed with a presentation on the BDFA funded project "Gene Therapy to treat visual failure in Batten disease" which included the work for her recently awarded doctorate. At the end of her talk Sophia also spoke about how continued BDFA funding, with the generous support of Beefy's Foundation, enabled her to continue her work last year leading to significant advances in discovering what is going wrong in the eye in her model system of Batten disease.

NCL Resource – A gateway for Batten disease

www.ucl.ac.uk/ncl

Professor Sara Mole has been awarded a 2-year grant by BioMarin Pharmaceutical to fund a part-time researcher to maintain and expand the NCL Mutation Database and NCL Resource hosted at University College London.

Up until now Sara has single-handedly maintained the website, however this new funding will allow the resource to be updated, expanded and maintained to continue to act as a crucial gateway for current information on all forms of Batten disease. The website is an important resource for families, clinicians and researchers and the BDFA welcomes this very valuable funding opportunity.

UCL

Developing new therapies for Batten disease

Batten disease or Neuronal Ceroid Lipofuscinoses (NCL) is a life-limiting neurodegenerative disease for which there is currently no cure.

What is BATCure?

BATCure is a 3-year research project funded under a European Union call (New Therapies for Rare Diseases). The goal of the project is to advance the development of new therapeutic options for patients and their families living with CLN 3, 6 or 7 Batten disease.

Who is involved?

A consortium from seven European countries made up of ten leading scientific research groups, three companies and the Batten Disease Family Association (BDFA) who is leading part of the project and ensuring that the voice of patients and affected families is heard.

The BATCure project focuses on the most prevalent types of NCL, CLN3 (Juvenile Batten Disease or JNCL), CLN6 and CLN7 diseases.

BATCure Coordinator

Professor Sara E Mole
MRC Laboratory for Molecular Cell Biology,
UCL Institute of Child Health and
Department of Genetics,
Evolution & Environment, University College London
s.mole@ucl.ac.uk

BDFA Project Contact

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www.bdfa-uk.org.uk

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

The second BATCure meeting took place in Riga, Latvia in June 2016. Our hosts were Dr Maija Dambrova (pictured below) and her colleagues from the Latvian Institute of Organic Synthesis, who made us all so welcome.

The meeting was opened by Professor Sara Mole from UCL in her role as BATCure Co-ordinator. Members of the Consortium then provided their updates on the first 6 months of the project including initial scientific work on the establishment of sound models for CLN3, CLN6 and CLN7.

Heather Band presented the BDFA's work providing the patient perspective and voice in research. One of the first tasks was to produce a range of materials for raising awareness and providing information about BATCure. The BATCure flyer (page 18) is an example of this work and is now available in ten European languages. Translation of materials is important as it enables the project to reach as many affected families as possible. We also had a tour of the Institute where we saw their excellent work first-hand and we will be working closely with Maija on a later part of the project.

Further BATCure news...

Laura Codd joined the BDFA in April 2016 as the BATCure Project Administrator.

Her background is in the market research industry where she has worked on a wide range of qualitative and quantitative research projects both as a Researcher and, more recently, as a Project Manager. Prior to starting her career, she gained an MA in Social and Cultural Geographies and a BA in Geography from the University of Sheffield.

"I am really enjoying contributing to the amazing work that the BDFA does. It is a privilege to be part of the BATCure project and to be playing a role in ensuring that the voice of patients and affected families across Europe will be heard within the research".

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

Can gene therapy treat CLN5 Batten disease?

Following her recent trip to New Zealand, Ana Assis, Research Assistant for this project, tells us about her experiences and how the work is progressing. The Principal Investigator for the project is Professor Jon Cooper.

Can you tell us what the overall aim of the project is?
CLN5 disease is a late-infantile variant of Batten disease, and this project was developed to test how effective gene therapy might be in a sheep model of the disease.

Gene therapy is based on the injection of a virus in the sheep brain, which replaces the faulty gene with its corresponding healthy copy in the sheep DNA. The cells can then produce the CLN5 protein that would otherwise be missing, which causes the symptoms associated with this disease.

What did you learn from your time at Lincoln University, New Zealand?
For the first part of my project, I travelled to New Zealand to spend two months collaborating with Professor David Palmer's lab. During my stay I witnessed the behavioural tests used to assess the CLN5 sheep's motor abilities, involving a maze made out of metal gates set up on a field on the University farm.

I learnt valuable techniques to help me analyse what has been going on in brains of the treated sheep that will enable me to test whether the therapy has been successful and/or if there may be any harmful long-term effects.

What is a typical day in the lab at Kings College for you?
I am now back in London, where I will complete the analysis of the results. I usually get to the office in the morning and after catching up on emails I get to the laboratory, where I either stain tissue using histology techniques or I spend some time in the microscope room taking pictures of brain sections for analysis. There is a very good work environment at our Institute (the Maurice Wohl Clinical Neuroscience Institute) and people from different groups are always happy to help each other out, so I consider myself very lucky!

Always a challenging question, but when could we expect the first results from your analysis?
This will take some time – the rest of the one-year project – but by early next year I am optimistic that I will have some interesting results to share.

Outside of the lab, what do you like to do?
I enjoy going for the occasional jog in the park and expanding my cooking skills whilst bringing friends together around a table. Being Portuguese and living in London, I generally try to soak up any ray of sun that breaches the clouds!

And your plans for the future?
When this project is finished I would like to keep pursuing the topic of neurodegenerative diseases by starting a PhD on this topic. We'll see what the future brings!

Ana B. Assis

DEM-CHILD

Introducing BDFA Funded Researcher Dr Susanne Lezius

Dr Susanne Lezius is a bio-mathematician at the Institute for Medical Biometrics and Epidemiology at the University Medical Center Hamburg-Eppendorf. Her collaboration with the DEM-CHILD database team began in 2014, analysing data on the course of Batten disease and defining clinical endpoints for clinical trials. Susanne brings a wealth of experience to her new expanded role for the NCL International Registry, made possible by the joint funding initiative of the BDFA, BDSRA, Noah's Hope and Hope for Bridget.

"We are deeply grateful for the generous financial support from the funding partners which will allow Dr Lezius to continue her work with us on the International NCL registry"

Dr Angela Schultz
MD, Children's Medical Centre, Hamburg-Eppendorf

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 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. In the summer Heather Band, BDFA Scientific Officer, arranged visits to see first hand how the work at UCL and Cardiff is progressing.

Development of a drug screen in CLN5 Batten disease

Andrew and Sarah Dawkins with Dr Daniel Little (UCL) and Heather Band (BDFA) in June at UCL

The group at UCL – Dr Dan Little, Professor Paul Gissen, Professor Sara Mole, and Dr Robin Ketteler – have 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, the cells that die in CLN5 disease. The first stage has been completed and they are now working on producing the nerve cells. The aim is then to use these cells to test different drugs to find treatments that could make them healthier.

Looking at what goes wrong in CLN5 Batten disease

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

In the first year of Katie's studies some key differences have already been found. CLN5 affected cells have changes in the way they communicate, make and use the "energy" they need to work properly and even have differences in their structure.

Andrew and Sarah Dawkins with Heather Band (BDFA) and the Neuronal Ceroid Lipofuscinoses team on their recent visit to Cardiff University

Eurordis Membership Meeting 2016, Edinburgh

The European Organisation for Rare Diseases (EURORDIS), founded in 1997, is the umbrella group and alliance of patient organisations and individuals involved in the field of rare diseases across Europe. It represents 716 patient organisations across 63 countries and covers more than 4000 rare diseases.

Each year it holds an annual membership meeting. This year it took place in May in Edinburgh in conjunction with the European Conference on Rare Diseases and Orphan Products.

After the EURORDIS members' General Assembly the remainder of the meeting included workshops to encourage learning and knowledge exchange between patient groups. The themes of the workshops included:

- European Reference Networks (ERN) and European Patient Advocacy Groups (ePAGs) by ERN Groupings
- RareConnect
- Social innovation: patient experience and expectations
- Good clinical practice in the context of ERNs

Andrea and Heather represented the BDFA at the meeting and presented the BDFA work in developing relationships with research professionals to drive the research agenda for the NCLs. Meetings like these are important ways in which the BDFA, as a rare disease organisation, is able to connect, learn and share with its peers and colleagues both in the UK and beyond.

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**). One of the organisation's key aims is to empower patient groups, and as part of this work they run events for rare disease charities.

Heather Band, BDFA Scientific Officer, was asked to speak at Findacure's training workshop 'How patient groups can work with researchers' in London in June, to share the BDFA's experience and knowledge in funding research.

The central theme of Heather's talk 'Together We Will Make A Difference' was how an innovative approach is needed for success and how collaborations are key when working together to drive the research agenda.

Heather highlighted many BDFA-funded projects to illustrate its research strategy including how the BDFA partners with universities, foundations and charities to fund excellence in research.

"There was lots of positive feedback about my talk and it was great to take part in a lively panel discussion at the end of the day."

A proactive approach in relationships with key stakeholders has been very successful. Finally, Heather spoke about how the BDFA supports families to fund research, for example in CLN5 research (p22).

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

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