WINTER NEWSLETTER

DECEMBER 2021 | ISSUE ONE

BDFA
BATTEN DISEASE FAMILY ASSOCIATION
Together we WILL make a difference
Registered Charity In England and Wales 1084908 - Scotland SC047408

INTRODUCTORY LETTER

Welcome to our first official BDFA newsletter for many years. We are delighted to be in a position to relaunch our newsletters, which will be produced four times a year in December, March, July and October. The newsletter is very much a work in progress and we want to hear your ideas for stories and regular features so please do get in touch.

It has been a very busy time at the BDFA and we are looking forward to being able to bolster our support to families in 2022 with our new Peer Befriending service as well as expanding our core family support team with a new member of staff who will work alongside Sian, our Head of Family Support and Advocacy. Many of our families have been able to access support from our family wellbeing service, delivered by The Maypole Project, which has proven to be much needed by our community. We will also be piloting a bereavement project, and the plan is that families will shape this service going forward.

We are also excited to be able to begin our own BDFA-led research project examining the diagnostic odyssey of Batten disease. Families have told us that, as their patient organisation, the BDFA has a vital role keeping abreast of the complex research and treatment landscape and we are pleased to be in a position to employ a part-time Scientific Officer early in 2022.

We have also been busy revamping our website and we will have news on this very soon and will be welcoming families to feed in their ideas. Our aim is for the new website to go live for awareness day next June.

Our strategy to diversify our income is proving to be successful and our Head of Fundraising Liz has brought funding in from many new trusts and foundations since she joined us in January. We will also be announcing the appointment of our new Community Fundraiser who will be working closely with families in local areas around the country.

In other news, Boris (the bear, not the PM!) has started his UK tour and will be spending his Christmas with a family in Scotland, who have two young children with CLN1, very recently diagnosed. You can read about the Borrows-Currie family on pages 6-7. We are really looking forward to hearing about Boris' adventures as he makes his way around the UK.

It was truly amazing to see so many exceptional people on one 'Zoom' as we announced the winners of our inaugural Pride of Batten awards. Chair of Trustees Zlatko Sisic and Secretary Pauline Docherty were bowled over by the dedication, kindness and excellence among those working and supporting families from around the UK.

Thinking of all our families this Christmas, those missing their loved ones and those still processing a very new diagnosis of Batten disease and all of the families in our community.

This is the online version of the newsletter but if you would like a print version please contact **admin@bdfa-uk.org.uk** and we will be pleased to organise that for you.

Amanda, Chief Executive

BDFA CONFERENCE 2022

Saturday 11th June - Sunday 12th June 2022

We are excited to be planning our family conference after a break of five years, and hoping that as many of our Batten families as possible will be able to join us there. Due to current times, the conference will be a hybrid event, so families can join us in person at the Crowne Plaza Hotel, Stratford-upon-Avon, or virtually for a weekend of events.

Early in the new year, the BDFA team will be announcing the plans for the weekend and letting you know how to book your places and all the details regarding hotel rooms, times, and planned events. All the family is welcome at the event, and we would especially like to welcome our community of bereaved families to join us.

We would love to see you.

PRIDE OF BATTEN **WINNERS**

2021 saw the launch of the BDFA Pride of Batten awards, celebrating best practice across the UK among people supporting and working with our children, young people, and adults with Batten disease.

All our winners and those specially commended were nominated by families, for 'going the extra mile', their kindness, and their support and insight. It was very difficult to pick out winners and commendations and every single person nominated is incredible and hugely appreciated.

Thank you to all who attended the awards ceremony via Zoom, all who took the time to nominate, and to our judges for their assistance. It was so special to see all the faces of those making a difference to our community alongside families and children and young people with Batten disease. Well done all! We hope next year to be able to do it all in person, fingers crossed. **And the winners are...**

EDUCATION

Winner: Kath Black, 1:1 Teaching Assistant, Poynton, Stockport
(nominated by Lucy Carroll, mum to Amelia, who has CLN2)
"We will be forever grateful to Kath for how much love she has shown our little girl. We thank her for being such an important part of Amelia's journey."

Special Commendation:
Lynette Iemboli, Teaching Assistant, Gloucester
(nominated by Ali Glover, mum to Joshua, who has CLN6)
"From the first day of Joshua's diagnosis Lynette has been absolutely amazing and a complete life-line to our whole family, not just Joshua. She really is the most incredible person and is thoroughly deserving of recognition which she very seldom gets."

Other nominees in this category:
- Chloe McCole, Sarah O'Donnell, Paul and all the other staff at Cosy Cottage Nursery, Edinburgh
- Claire Middleton, Head Teacher Wickersley Northfield Primary School, Rotherham
- Emma Swift, Visual Impairment Teaching Assistant, Rotherham
- Chris Rollings, Headteacher, Hadrian School, Newcastle
- Melissa Taylor, Teaching Assistant, Sheffield
- Rachel Broderick, Teaching Assistant, Birmingham
- Rachel Elliot and Emma Thompson, Teaching Assistants, Peckover Primary School, Wisbech
- The Visual Impairment Team, Rotherham Metropolitan Borough Council

HEALTH

Winner: The Great North Children's Hospital medical and nursing team, Newcastle
(nominated by Gail Rich, mum to Nicole and Jessica who both have CLN2)
"We cannot thank the team at Newcastle enough and we feel incredibly fortunate that our voices were valued and that they wanted to help us. They thoroughly deserve to be recognised because they made this happen and they supported us in achieving what has been our goal since the girls began treatment back in 2017."

Special Commendation:
Steve Box, Community Nurse, ID-CAMHS, Dorset Health Care
(nominated by Lisa Hover, mum to Annabelle and Robbie, who have CLN3)
"Steve is a family man himself and is very genuine with his concerns. He is just a great, kind, honest man."

Other nominees in this category:
- The nursing team at Manchester Children's Hospital (Claire Hamilton Kay, Fiona Heap and Katrina)
- The consultant and nursing team in Edinburgh (Dr Elizabeth Pilley, Michelle Small and Celia Brand)
- Justyna Szmyd, Physiotherapist, Burton-on-Trent
- Laura Lee, Clinical Nurse Specialist, GOSH
- Professor Paul Gissen, Consultant in Paediatric metabolic diseases, GOSH
- Shani Bradwell, paediatric physiotherapist, Sheffield

SOCIAL CARE

Winner: Anna Dene, Carer, Sheffield
(nominated by Michelle Windle, mum to Olivia, who has CLN3)
"Anna is a great support to us all and has often been someone I can pour my heart out to as we go through this journey. Even though Olivia can no longer tell us, we know she loves spending time with Anna. You only have to see the smile on her face!"

Special Commendation
Sofia Khan, Social Worker, Sheffield
(nominated by Chantelle Cammack, mum to Sheyne and Amber, who have CLN2)
"Sofia has become part of our extended family, with us through the good and bad times, providing the best support possible."

Other nominees in this category:
- Sabrina Kah, Family support worker at The Rainbow Trust, Sunderland

OTHER

Winner: Barbara Moore, Camsight Family Worker, Wisbech
(nominated by Sarah Dodkin, mum to Finley and Harrison who have CLN3)
"Barbara has retired this month which is well deserved for her as she truly deserves to live life to the fullest but we and other families will miss her so very much. But I know that if we really needed her, Barbara would still pick up the phone, come round with a hug and point us back in the right direction because that's who she is. An absolute diamond of a woman."

Special Commendation: Alex Currie, Uncle, Edinburgh
(nominated by Sophie Currie, mum to Eilidh and Cameron, who have CLN1)
"We would not have been anything as organised at this stage without his help and I am really grateful for everything he has done for us and his niece and nephew."

Other nominees in this category:
- Tracy Paxton Beattie, Community Champion at Sainsburys, Throckley

BORIS THE BDFA BEAR'S **UK TOUR**

Like the rest of us, Boris had his holiday plans cancelled last year due to COVID-19 so now he's is making up for it and is off on his travels again starting this Christmas!

Boris and his suitcase are going to be on the road joining our amazing Batten families over the next year. He will stay for a week with a family and his journey can be documented in his travel journal and the website for all the other families to read and see what he has been up to. He cannot wait to see you and get cuddles from you all!

His suitcase is FULL of things to help Boris settle in and make himself at home. He is off to be with a family for Christmas and we already know he is going to be spoilt rotten!

If you would like Boris to come and stay with your family soon, please do drop Lisa an email at **admin@bdfa-uk.org.uk** and we will get him booked in for a visit! We cannot wait to see what he will get up to with you all.

NEW LONDON **OFFICE**

In early September the BDFA moved into our new home a short walk from Kings Cross station, London. Lisa and some volunteers sorted and moved us into the new office, which is bright and welcoming and is a great space for the team to meet and work from. The current COVID-19 situation means we are still predominantly working from home, but we are all getting to the office as much as we can. Families are very welcome to come and see us at our new office in the new year.

The office is also home to our special memory book of children, young people and adults whom we have lost to Batten disease. Please contact Lisa on **admin@bdfa-uk.org.uk** if you would like to include an entry for your loved one.

DIARY **DATES 2022**

BDFA office closed for Christmas
Friday 24th December (Christmas Eve) - Tuesday 4th January

Rare Disease day - Monday 28th February

Batten Disease Awareness day - Thursday 9th June

BDFA Family conference - 11th June - 12th June
We are planning that this conference will be a hybrid event, so families can join us in person at the Crowne Plaza Hotel, Stratford-upon-Avon, or virtually for a weekend of events.

BDRSA Family Conference - 8th - 10th July
35th anniversary in Cleveland, Ohio, USA. Families in the UK will be invited to join virtually and with the option of ordering t-shirts and conference 'boxes'.

CHRISTMAS **CARDS**

A huge thank you to all of the families who entered the Christmas card competition this year, the designs were amazing, and our judge, our esteemed and beloved, Barbara Cole chose the two winners, 9-year-old Evie and 16-year-old Annabelle.

Our first winner is Evie, who has CLN2 Batten disease, and Mum Rebecca Davies who worked as a team on their design said *"thank you for the suggestion of designing the card together, we had a really good time and Evie loved the praise for doing so well! Besides drawing, outlines, and a bit of shading, Evie did all of it, including the painting and stamping letters!! It was loads of fun and it really left me feeling a sense of achievement with Evie, as we had not done a little project together in a long time. It was very special and it means a lot to have created a card together with big smiles!"*

Our second winner is Annabelle Hover, aged 16. Annabelle has CLN3 and we loved her design too. Annabelle said this about her design process, *"I have CLN3 but I won't let it stop me. I have no useful sight left so drawing is hard for me. For my Christmas card, I drew around two cookie cutters, for the snowman, then I used wiki stix for me to know where the circles are and then I stuck snowflakes and a Happy Christmas sticker."*

Cards have been selling like hotcakes, and have been a real hit so our thanks to all who ordered some this year. We had:

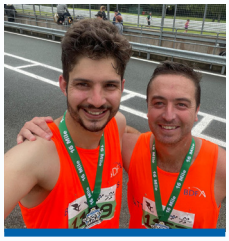
19 design entries
2 winners
127 packs sold
1270 cards sent!

FUNDRAISING UPDATE **FROM LIZ**

Hello everyone, what a busy time we have had in fundraising! Community fundraising has really taken off again since COVID-19 restrictions eased in the summer and we have had some wonderful events happening throughout the community. Thank you to everyone who has run an event for us over the summer/autumn and to those who continue to work tirelessly in their community to raise funds for the ongoing work of the BDFA.

'RUNNERS **ROUND UP'**

It has been absolutely wonderful to see organised running events happening again and they have been quite emotional to witness, as myself, Lisa, Amanda and Sian have all been along to an event this autumn to cheer on the runners. We had runners represent and raise funds for us in the **Great North Run**, the **London Marathon**, **Royal Parks Half Marathon** and the **Manchester Half Marathon**.



We would like to thank all our runners; Team Rich of The Nicole and Jessica Rich Foundation, Duncan Brownnutt, Evan Brownnutt, Toby Bidwell, Justin Kerwin, Mat Norval, Heather Cruickshank, Tom Elliot, Sara Brookes, Dan Smith, Ryan Radford, Louise Bullock, Simon Young, Andrew Charlesworth, Jenny Mabbutt, Helen Jackson and Chris Drinkwater. You all did the BDFA proud and we are so grateful to you all.

OTHER FUNDRAISING **NEWS...**

The Cardboard Box Company in Blackburn took part in a large dragon boat race event in September, held by their parent company, W & R Barnett in Northern Ireland. Lots of companies took part to raise funds for their chosen charities. The Cardboard Box Company chose to support the BDFA because of a personal connection (to a newly diagnosed child Jeffrey, who has been recently diagnosed with CLN2 Batten disease) and they raised over £12,000 in sponsorship. Then as their team finished the race in an incredible second place, W & R Barnett Ltd provided generous matched funding, which resulted in a donation to the BDFA of almost £59,000! This was utterly astounding and we are so grateful to The Cardboard Box Company and to W & R Barnett Ltd for their amazing generosity, kindness and support.

Early in September, the BDFA team were delighted to volunteer at one of **Beefy's Charity Foundation's** special dinners, in celebration of the 40th Anniversary of the historic 1981 Ashes win! Lord Ian Botham is the founding Trustee of Beefy's Charity Foundation and he and his fellow Trustees have supported the BDFA for a number of years through their prestigious fundraising events. It was a fantastic evening at The Oval in Surrey and we enjoyed meeting Lord Ian Botham and his family, whilst helping with the chocolate raffle and historic auction.

DECEMBER **NEWS...**

Our wonderful supporter **David McGovern** from **Beyond Vinyl** record store in Newcastle, has released a special Christmas charity single. This follows the huge success of 'Beautiful Girl' for Batten Disease Awareness day in June, which got all the way to number 5 in the physical charts. David's Christmas single is a cover version of the song "Stay the Same," a solo hit in the late 90s for Joey McIntyre, previously of the boy band, New Kids on the Block. David's version is a double A side release, also featuring "Santa Claus is Comin' to Town," with guest vocals from local legend, 'Santa Ste'.

Two weeks ago it was announced by the official charts that David's single is a contender for the coveted Christmas number 1 and David now finds himself going head to head with the likes of Adele, Elton John, Ed Sheeran and Gary Barlow! Please support David by buying the single, downloading and streaming it so that we can get him all the way to number 1 and raise huge amounts of awareness.

I am continuing with the vital work of writing to Trusts and Foundations for grant support for the BDFA and I really need your help with this. I am looking for quotes/stories and also photographs that families might be willing to share. This would really help funders to see the reality of Batten disease and the need for the BDFA's work. Please do get in touch with me directly if you can help with that – **07745 210212**, **lizbrownnutt@bdfa-uk.org.uk**.

We would love to hear from anyone who is interested in undertaking an event on our behalf. We can certainly guide and support in all of that, so please do get in touch and encourage others to contact me directly.

There will also be opportunities for runners and cyclists to represent us in various events in 2022. We have already secured places in the prestigious **RideLondon** event taking place in May 2022 and the **Royal Parks Half Marathon** in October 2022. If you are a keen cyclist or runner and would be interested in taking part in these events to raise funds for the BDFA then please do get in touch.

Finally, a huge 'thank you' to everyone who nominated the BDFA for a £1,000 award in the **Ecclesiastical Movement for Good '12 Days of Giving'** initiative. We found out last week that we have been successful and we will receive the award in January. Thank you for sharing the news far and wide and for securing all those crucial nominations.

Thank you for all your support in fundraising!

REGULAR GIVING – A regular donation to the BDFA, however small is extremely valuable, as it helps us to plan our future work. Please contact **admin@bdfa-uk.org.uk** to set up a standing order or visit our website to set up a monthly payment via PayPal.

Focus On:

MUSIC & BATTEN **DISEASE**

The Amber Trust
Music for Blind Children

The Amber Trust's With Music In Mind music service aims to transform music provision for visually impaired children and young people with neurodegenerative disease by providing parents, carers, teachers, and therapists with freely available resources and one-to-one musical support.

Support can either be in the form of weekly music lessons if the child is able to benefit from them or through monthly visits by a specially trained music practitioner who works with the whole family with a view to supporting musical engagement as well as wider well-being.

The service is freely available to children and young people under the age of 18, resident in the UK, who have Batten disease or another neurodegenerative disease in which bilateral visual impairment is present and causes in its own right a significant impediment to the child's ability to learn or function independently.

All applications should be made by a parent or guardian but can be supported in the process by a health care or other support worker. Families accepted onto the scheme will receive an initial assessment and the award will be reviewed annually.
For more information visit: **withmusicinmind.ambertrust.org** contact Hannah at **wmim@ambertrust.org**

RESEARCH **UPDATE**

Rebecca Atkinson, from Brighton University, is investigating the impact of music for children and young people diagnosed with Batten disease. The research specifically looks into how music therapy and music education programs can support and maintain children's communication in a school environment.

Anecdotally, parents and staff have emphasised how a child's language is better when they are singing. Pursuing this further, the study began in 2016, by following children in their music therapy sessions for 3 years, tracking changes in their communication. Findings showed that children's communication appeared to be maintained when they took part in weekly music therapy or music education sessions (assessment scores plateaued in the communication area over the course of the 3 years).

Follow up research in 2020, created the *Music Speaks* program which was trialled in the UK with children affected by CLN3. Interviews with staff members talked about how the program was empowering for the children and staff who took part in the trial. Findings demonstrated that from using *Music Speaks*, children learned how to slow their speech down using beats and draw upon musical phrases to help support their communication. The program also gave staff the skills to support children's communication through music on a general basis in the classroom.

Ongoing analysis will continue into 2022 with results expected by the end of the year. Research findings will be shared with the BDFA, our US counterparts the BDSRA and the global Batten disease community, to help support affected children, their families and educators with musical activities, ideas, and resources.

RESEARCH **UPDATE**
from our Chair of Trustees

New BDFA-led research project looking at the diagnostic odyssey

Dear Families,

Although this has been another strange year in our lives, something quite remarkable and positive has happened.

Under the leadership of Amanda, her team and the Board of Trustees and thanks to your incredible support, the BDFA has gone from strength to strength organisationally, enabling us to do many things for the benefits of patients diagnosed with Batten disease. One of the projects that we started planning this year and will execute in 2022 is a survey type study looking at the reasons for late diagnosis in NCL disorders. We intend to capture the feedback from families and healthcare professionals about the diagnostic odyssey by applying scientifically valid methodology. The results of this study will inform targeted educational strategies and will help the BDFA engage key stakeholders in shaping diagnostic pathways for children diagnosed with hereditary neurodegenerative diseases.

Thanks to a generous support from our partners Amicus and BioMarin, we will be able to start the project in early 2022. The study will involve the families of children diagnosed with CLN2 and CLN3 since these are the two most frequently diagnosed types of Batten disease in the UK. However, we expect that study conclusions will help initiate improvements across the spectrum of Batten disease disorders. We will be reaching out to you regarding the participation in this project and would be grateful if you take part in the survey.

We look forward to our partnership in creating a better future for our kids.

For further information please email Zlatko on **zlatkosisic@bdfa-uk.org.uk**

Zlatko Sisic
BDFA Chair of the Board of Trustees

FAMILY **STORY**

Sophie and Stuart Borrows-Currie family live in Edinburgh. They have two children, Eilidh, 4 and Cameron, 2. Both children have CLN1 Batten disease.

Our baby girl Eilidh was born in September 2017. It was an uneventful pregnancy, a difficult labour and she was a joyous, confident, loving baby. She walked and talked early and loved being with other people, adults and children alike. An early fondness for songs and stories soon grew into a full-blown love affair with books and her affectionate nature won over the hardest of hearts.

Her little brother Cameron arrived in October 2019. Another easy pregnancy, a much easier birth but a difficult first few weeks for the whole family this time, with Eilidh meeting 'Bay Camun' for the first time when he was in an incubator in the neonatal unit.

He soon recovered and we were well on the way to becoming a solid foursome. He shared his sister's early love of books (sometimes not the best news for a sellotape-wielding Daddy!) and although he was not quite as keen to get started on walking and talking he got there in the end.

In March 2020 when the lockdown hit our little ones were 2 and a half and a small baby. Eilidh took it particularly hard, as she was sent home from nursery one Tuesday lunchtime and never went back. She stopped seeing grandparents, going swimming, seeing friends, visiting cafes and all the other little pleasures she had grown used to in a busy couple of years. Our bright bubbly little girl became a little less sure of herself, more easily upset and always checking that Mama and Daddy weren't going to disappear too.

Looking back now, it's clear that at some point over this summer her CLN1 started to make itself known too. When we were allowed to see friends again, we began to see that our clever cookie, who had found learning so easy, had started to fall behind her friends. When nursery started up again in August 2020 she found it very hard to settle in, a complete contrast to her usual self.

By Christmas last year it was clear that there was something different about her. We initially suspected ASD, or ADHD, or some other similar difficulty. Even when she began to have absences around the start of the year, we were assured that they could be a behavioural feature of a potential ASD diagnosis and I wouldn't have even considered something like Batten disease at that point.

However, things deteriorated quickly for our little girl. By April she was barely able to sit still, unable to concentrate, her balance was very poor and she was clearly having significantly more absences than could be explained by ASD.

We took her to Edinburgh Sick Kids hospital twice because we were so concerned, and when an EEG gave us a diagnosis of epilepsy at the end of May we were relieved. She began medication the same day and we instantly saw an improvement- she wasn't back to normal yet but we could see that the potential was there.

We reassured friends and family this is okay, epilepsy is manageable, it might take a while but we will get on top of it. She will be able to lead a normal-ish life, she will be happy, we can cope, it could be worse.

In July the routine testing associated with an epilepsy diagnosis in Edinburgh was carried out.

Urine sample - normal.
Lumbar puncture - normal.
Routine bloods - normal.
MRI - wait and see.
Genetics - wait and see.

So we waited in regular contact with the epilepsy team managing Eilidh's care and not really expecting anything further. Life carried on, we saw family, went on holiday, had Stuart's birthday.

In July we had the MRI results, Eilidh's scan had shown some minor cerebral atrophy. Because Eilidh hadn't had an MRI before it was impossible to say if this was brand-new and progressing fast, had always been there and meant nothing or somewhere in-between.

It was the first indication that something that wasn't fixable might be happening to our little girl. We tried our best to ignore it and carry on.

On August 27th 2021, two days before the due date and two weeks before her 4th birthday, we had the results back from the epilepsy gene panel that all new epilepsy diagnoses are sent to in Edinburgh.

It was a catastrophic diagnosis for us. Our smart, quick, joyful, loving little girl was going to waste away in front of our eyes and we would be powerless to do anything but watch. That first weekend (when we didn't know it was an unusual variant and we weren't sure if she would still be around, walking and talking and eating Christmas dinner with us) we talked over lots of things I never thought I would need to do for my daughter. What would her funeral be like, should I stop buying reduced clothes at the end of summer and saving them for next year, will she be able to recognise my voice when she can't see me, how much will she suffer, does Cameron have it too, if Cameron doesn't have it how do we ever make it okay that he will lose his sister, will there ever be a point in our lives where we are completely okay again? Probably not.

continued overleaf...

Time passed and things settled. Eilidh responded really well to a new medication and was much more her old self. Some things were gone never to return, but we accept that and we focus on what is still there. She is much happier, more engaged with the world around her and without a peer present to compare her to it is easy to convince yourself it will be okay.

On October 29th, less than 2 weeks after he turned 2, we were given the worst news we could have had about our little boy. Our vanishingly rare gene, which we had innocently passed on without knowing the risk, had struck twice. There was the same chance of neither of them even being a carrier and a far higher chance of neither being affected. How had the inexplicable happened? How was it possible that my darling boy, so cheerful and just starting to become his own little man, would soon start to suffer the same losses as we had already seen his sister suffer?

On the upside, there will be no 'oh it might be behavioural absences' and he may well retain more skills for longer. He will never have to experience that period of time that Eilidh did where we just didn't know and couldn't treat it.

There are, of course, lots of negatives. Cameron is completely fine at the moment. Eilidh's slow-motion train crash had already derailed and was rolling gently down the embankment by the time we knew anything about it.

Cameron is by contrast chugging merrily along, blissfully oblivious to the disaster lying in wait that everyone else can see.

And so we tick on. This is not a sad story. Our children are surrounded by loving family and friends. The medical support and the charitable initiatives that are available to us have been outstanding in our short experience so far, and I have no doubt that will continue as we go along. We know about CLN1 now, and we are lucky that we do still have time and the children do still have the health to do the fun things in life and will continue to experience happiness for as long as we can provide it for them.

So it is not a sad story. But it is no longer a happy story either…

by Sophie Currie

FAMILY SUPPORT **UPDATE**

Hi all,

I just wanted to say a huge thank you for all of your kindness and for making me feel so welcome in the BDFA. I am still in the process of reaching out to all of you. If I haven't spoken to you yet, I will do, and if you have something that you need to discuss, please do not hesitate to call **07876 712553** or email **sianfisher@bdfa-uk.org.uk**. I will be happy to assist you in any way possible.

Over the last few months, we have been helping many new families who have reached out to the BDFA and current families that have needed support. We have attended meetings including EHCPs, annual reviews, social services, etc. We have trained schools, sent out grants, referred families to Maypole, written letters to support DLA applications and guided families trying to find their way through the first few weeks of diagnosis.

Amanda and I have been to the Times Educational Supplement (TES) SEN show and spoken with other charities, including the Royal National Institute for the Blind (RNIB), who offer RNIB Bookshare. They provide free access to over 320,000 books from early years to higher education, audible stories. Here is the **link https://www.rnibbookshare.org/cms/**

New staff update: we have recruited a new member of the family support team, with the help of Chantelle (mum to Amber and Sheyne) and Laura Lee (thank you). Elizabeth, our new staff member, will work alongside me, working 21 hours a week. Both Elizabeth and myself will work across the UK. Elizabeth has wonderful experience supporting families who have children with disabilities, and her kindness shone through in the interview. We very much look forward to welcoming her to the team in the New Year.

Families have now started to request home and school visits which we are more than happy to do so: please get in touch with me if you would like one. We are pleased to travel to the north or south, and we have recently offered a home visit to Scotland.

I look forward to speaking to you all very soon, and please do reach out if you need us.

Be kind to yourselves
Sian

FAMILY **STORY**

In October, one of our lovely BDFA families came to us with some extremely sad news. Their seven-year-old daughter with CLN2 was in intensive care, and dad stayed with her day and night. Mum is pregnant, and they have two other children. Due to this, dad was unable to work, so he was unable to pay their bills, and this added pressure was only adding to the families stress load. Sian Fisher and Laura Lee started to look into charities that could help; each said 'no', and it was becoming heart-breaking for the family to be told that there was no help.

It wasn't until Liz, Head of Fundraising, suggested a charity she had worked with before. This fantastic charity paid for their rent, other bills, food and Christmas presents. When dad was told what help they could give, you could hear the relief, and it was the first time we had heard him laugh. Dad said he was highly grateful for the support from the BDFA and 52 Lives; he said, 'you have saved my family'. Things have remained a rollercoaster journey for this family. Since then, their daughter has been out of the hospital and has gone back in to intensive care. The family does know they have the support of the BDFA and 52 Lives to support them every step of the way.

NCL CONFERENCE 2021
SUMMARY

NCL 2021, or the 17th The International Congress on Neuronal Ceroid Lipofuscinosis (Batten disease), took place in St Louis, MO in early October 2021. This was one year later than originally planned because of Covid-19, and for the same reason NCL 2021 took place as a 'hybrid conference'.

There were nearly 100 people at NCL 2021 in person and almost 200 attending virtually. Such NCL conferences bring together Batten disease scientists, clinicians, and family organizations to listen to and discuss the latest advances in research. In organizing the conference, we wanted to present science of the highest scientific standard, but also to do our best to make this understandable for families. In addition to 'explainer' events before and during NCL 2021, this document is meant to provide a summary of the most important 'take home' messages of the work presented and what this means for families. These are split up into the different sessions of the conference, with a short paragraph on each.

For more details of the presentations or to download the conference booklet containing all the abstracts, please visit: (**ncl2021.org**). We have also prepared a brief lay summary of each presentation that can be downloaded separately. Any comments and questions can be addressed via email to (**cooperjd@wustl.edu**).

Genetics and Cell Biology (Underlying causes of disease & what Batten proteins normally do)

These studies can seem very far removed from affected families, and can appear very complicated and difficult to understand. However, they are the foundation on which our understanding of disease is built and without them none of the experimental therapies or potential treatments would be possible. While we all know Batten disease is caused by mistakes in the DNA that is the blueprint for making proteins inside our cells, what these Batten proteins do has long remained mysterious. In this session we heard new information about how the CLN3, CLN5, CLN6 and CLN8 proteins interact with other proteins inside the cell. This influences how things move between compartments inside the cell, or how things get into or out of cells. If such very basic processes go wrong then this will cause cells to become sick, and may explain why a problem in the cell's waste disposal system or lysosome can have such a devastating impact on a child. How things get to the right place at the right time is especially important in brain cells, and at the synapse (where nerve cells or neurons talk to each other). New 'iPSC' cell models have been made from human skin cells and 'reprogrammed' genetically so that they be turned into many different sorts of cells (for example brain cells) for studying disease. Such cells can be used to test or 'screen' many hundreds or thousands of drugs at once to identify promising compounds for testing in animal studies and several examples of doing this were presented

Disease mechanisms (What goes wrong in disease, and where and when does this happen).

This group of studies are important for finding out how Batten disease affects the brain and other parts of the body. They are also the next step in finding clues of how to potentially treat these effects of disease. In this session we heard about the importance of the CLN1 enzyme PPT1 at nerve terminals and how it's absence may affect how nerve cells communicate with each other. It appears that the microglial cells that make up the brain's immune system are especially vulnerable in both CLN1 and CLN3 diseases and their failure to function properly may be an important step in how the brain is damaged. Simple brewer's yeast with CLN3 disease can be used to identify promising new drugs for testing in mice. Data about the eye in CLN3 disease was presented, including how the cells of the retina at the back of the eye degenerate. While the brain is severely affected in all forms of Batten disease it appears that nerve cells in the rest of the body also fail to function properly in CLN1, CLN2 and CLN3 disease mice. This includes the peripheral nerves that relay pain and touch information, and we also learnt how nerve endings in muscle are affected by disease. It is also clear that nerve cells in the bowel of CLN1, CLN2 and CLN3 disease mice are attacked by disease, and this not only appears to cause severe bowel problems, but may be treatable by gene therapy.

New Technologies (How can we speed up research using new methods to help our work)

Scientific research can seem to take forever to make sense of something very complicated. However, technology is always improving and some powerful new methods may help speed up our work. One group of methods for doing this depend on studying everything that is happening inside a cell or organ at once. Such 'big data' or "-omic" methods look at patterns in how many things have changed as a result of disease to identify the key processes that have gone wrong. Examples include genomic studies (looking at which genes are switched on), proteomic studies (how much of individual proteins are present), and we learnt more about how these can be used to study disease pathways and processes. These methods can also find biochemical markers ('biomarkers') used to follow disease progression and to judge if therapies are working. It is also possible to modify or change a protein to alter its chemical properties or how it behaves, and this is being used to make new and potentially more powerful forms of lysosomal enzymes that can be taken up inside cells better. We also learnt of other ways to possibly deliver such enzymes to the brain in small particles and these are being tested in Batten mice.

Preclinical translational work (Testing experimental therapies in animal models).

This type of study involves identifying potential treatments and testing these in an animal model of Batten disease. The goal is to find methods that can treat animals (therapies that are effective) and appear to be safe to use, so that they can then next be tested in people. Such 'pre-clinical' work depends on having a good understanding of how disease affects these animals, and this allows scientists to judge both safety and efficacy. Most of this work is done in Batten disease mice, and we heard about several new approaches, especially in CLN3 disease. This included how antisense oligonucleotides (small fragments of especially designed genetic material) can be used to skip past the mistakes or mutations in the CLN3 gene to improve disease, and how certain drug combinations (trehalose and miglustat) can be used to stimulate or jump start the cells waste disposal system. Both have showed promise in mice, with the second strategy being the one that will be used in the 'BBDF 101' CLN3 clinical trial that was announced recently. Directing gene therapy for CLN3 disease to different types of brain cells can influence their disease, and this tells us more about how important nerve cells and their supporting astrocyte cells are for brain function. Gene therapy has also been tested in CLN7 mice and these scientists plan to move to a clinical trial shortly.

In addition to mice we also heard about other animal species that are being used. For example, small zebrafish are valuable models for studying disease, and both CLN2 and CLN8 zebrafish have been made and how disease affects them has been studied, so that potential therapies can be tested in them. Work like this is being used to screen many drugs at once in these fish models to identify those which may be best to test in mice. There were also several presentations about 'large animal' models of disease, which include sheep, pigs and non-human primates (a type of monkey). These all have much larger and more complicated brains than mice, and appear to get Batten disease that is more like that seen in children. All of these models need to be studied in great detail to see how they are affected by disease. We heard about such work in genetically engineered CLN2 and CLN3 minipigs, and naturally occurring CLN7 macaque monkeys. This work includes how disease attacks their brains and affects their behavior and how disease progression can be followed by imaging methods and biochemically. This gives vital information for judging if experimental therapies have been effective and can be scaled up to treat a larger brain. This was the approach used to develop Brineura for CLN2 disease, and we learnt that similar work is being done to develop enzyme replacement for CLN1 disease. Giving the missing PPT1 enzyme to the brains of CLN1 mice or sheep is producing encouraging results, but more work will be needed to improve this strategy. We also heard more about directly treating the eye in CLN2 dogs and CLN5 sheep, as blindness may not be treated properly if we only treat the brain. The alternative is to treat both the brain and eye at the same time, and we learnt of how gene therapy is being used to do this in CLN5 sheep in New Zealand.

New Clinical Perspectives (Understanding more about Batten disease in people).

Just as learning about disease in animal models is important, so is finding out more about how the different forms of Batten disease affect people. You might think this information would all be already known, but for finding out if therapies have been effective or safe we need much more detailed or 'quantitative' measurements from people. Such vital information on 'natural history' has led to rating scales that can be used in a clinical trial. Studying Batten disease in people can also reveal new aspects of the disease that can allow medical doctors to improve the care or support they provide. The new work we heard about included a new rating scale that can be used for children with CLN1 disease, evidence for autistic like behaviors in CLN2 disease, an ophthalmic scale to assess vision in CLN3 disease, and much has been learnt by following CLN3 disease for nearly 20 years in the same people. We also heard of new guidelines for diagnosing, assessing disease and treating children with CLN2 disease. Fitting a pacemaker can help stabilize the heart problems that occur in CLN3 disease, and developing educational interdependence and understanding behavior can provide better quality of life for individuals with CLN3 disease.

Clinical translational work

The goal of all the 'basic' science work to understand the causes and mechanisms of disease, and to test experimental therapies in animals is to find treatments for Batten disease children and young adults. There is no guarantee that any approach that has been relatively successful in an animal model will have the same effects in humans, and clinical trials are effectively still experiments that need to be done to test both efficacy and safety in people. Such clinical trials depend upon having solid foundations of careful preclinical work and detailed knowledge of disease in people. Even if a clinical trial proves successful and a treatment is approved by the FDA, as happened for Brineura in CLN2 disease, we cannot predict how treated children will respond in the long term. Most of the presentations we heard in this last session were about how Brineura-treated children with CLN2 disease are responding to receiving this missing CLN2 enzyme, including how their immune system responds, how is best to manage their care and the practicalities of repeatedly delivering Brineura every two weeks. New ways to follow disease by MRI imaging or monitoring biochemical changes in CLN3 disease were also presented.

Overall summary

Scientific conferences are always a time for scientists and clinicians to meet together and exchange information and ideas, with many new ideas and collaborations coming from these encounters. NCL 2021 was rather different as we couldn't all be together physically in the same place, but as a hybrid conference we wanted to give the best possible experience for everyone who couldn't travel to St Louis. A conference app helped make this possible with only very minor technical problems, and also allowed anyone to ask questions of the presenters. Yes, we missed the face to face interactions, but even under these unusual circumstances we were able to bring the Batten research and family communities together. A great deal more detailed information is emerging, and more treatment strategies are showing promise in animal models, with some of these advances now beginning testing in clinical trials. Although this work can never go fast enough, there is an increased sense of momentum and progress being made.

There are many young talented Batten researchers, many of them women, who had the chance to present at NCL 2021. We will all look forward to hearing more about their work at the next international congress, NCL 2023, which will take place in Hamburg, Germany.

Following the conference, the BDFA was pleased to hold a webinar with Jon Cooper and Sara Mole which is available for families. Email: **admin@bdfa-uk.org.uk**

Jon Cooper and Joshua Dearborn,
Washington University in St Louis

BRINEURA IN THE EYES

Eight children with CLN2 Batten disease are part of a unique compassionate use programme at Great Ormond Street Children's Hospital (GOSH). The children are all accessing cerliponase alfa (Brineura©), with infusions given directly into the brain. However, this treatment does not help with the deterioration in sight. In this programme, clinicians at GOSH use the tiny amount of drug left over from the brain infusion and inject it directly in the back of the eye, an intravitreal treatment. At this stage, Mr Rob Henderson and his team are treating one eye every two months and after a year they will compare the effect of the drug on the loss of cells from the back of the eye in treated versus untreated eye. This treatment could preserve children's eyesight for longer.

This is an unusual programme as parents raised the funds, fundraising over £250,000. It is hoped this treatment will be available to other CLN2 children at other accepted infusion centres after the initial period at GOSH and eventually available on the NHS.

IN MEMORY

Caleb William Brownnutt

16th May 2010 – 15th November 2019

Caleb so often amazed us all with his resilience in the face of his condition that it's worth remembering that, his size notwithstanding, he was still just a little boy, albeit the funniest, toughest and smartest little boy you could ever hope to meet. He was a gift to us and carried the family through the tragedy of losing Ellie Mae.

Caleb had a way of making everyone love him and so his fan club over the years grew to include a large number of people, mainly girls, who he could trick into doing his bidding. We are of course hugely grateful to those people who were friends to Caleb but they were also hugely rewarded by him with a laugh, a smile, and, for the most highly regarded, the special greeting that Buzz Lightyear taught him. High 5, Fist Bump, Boom.

He was fanatical about trains, something he inherited from his Grandad. It's a testament to Caleb's intelligence that he could recognise and name all of the main trains from Thomas the Tank Engine, and a demonstration of his stubbornness that he insisted every one of them was blue.

Even as he was losing his speech he clung on to a few words and I think it's relevant that the last clear words I heard him say were Grandma, Grandad and Poppy. Put together those were all the most important people in his life.

He achieved so much in such a short time, meeting Buzz Lightyear, Pooh & Tiger, feeding a giraffe, and kissing a sea lion. He met Cliff Jones and Ossie Ardiles and sat in Pocchetino's chair in the dug-out at White Hart Lane. He drove the Mallard and saw all 6 surviving A4 Locomotives, 3 of them under steam (for a train-lover like Caleb, that was a big deal). He travelled the World and always seemed to find a Princess he could hug.

James Harrison

14th March 2008 – 3rd July 2021

Our beautiful brave James passed away at home on Saturday July 3rd 2021 surrounded by his family. From the moment James entered the world he wrote his own plan and we've been flying by the seat of our pants since. His life was not what we had planned but we made sure it was full of fun, laughter and adventures.

We had 13 years, 3 months and 29 days with James. That's 4859 days filled with happiness and love. Despite his disability he had a zest for life and a true determination to live it his own way.

James has left a huge hole. Life will never be the same again. He will never be forgotten and forever talked about. He was a wonderful son and brother. James was the most amazing boy we've ever met and we had the honour and privilege of being his parents.

"How long will I love you?
As long as stars above you
And longer if I can"

Blainie White

28th October 2008 – 20th September 2021

Time for me to go now,
I won't say goodbye,
Look for me in rainbows,
way up in the sky.
In the morning sunrise
when all the world is new,
Just look for me and love me,
as you know I loved you.

Fly free my special little bee
forever loved and missed so dearly

Frank Peter Billy Thompson

1st February 2008 – 9th October 2021

Frank light of our lives. How fortunate we were to have you. Watching you relish life despite the struggles you faced. You remained so joyful, giggling in the face of adversity.

You taught us so much, but mostly how to cherish life no matter what it throws at you. You taught us to find joy in the smallest and simplest of things; bashing a tambourine, the taste of food on your lips, swinging in the garden, the warmth of the sun on your skin, the wind in your hair, listening to music or floating in a pool.

You touched so many in your short life. We have been overwhelmed by the outpouring of love for you, and our family. As we grieve we will remember what you taught us. Life is a privilege to be lived to its fullest.

Bertie Petty

4th November 2014 – 12th September 2019

We miss our beautiful boy Bertie every day. He had a constant peace about him which everybody noticed and when he smiled it brought us so much joy.

He never stopped recognising our voices and broke into huge smiles when we told him it was the morning and played his favourite song, 'My Lighthouse.'

We will never stop loving him and can't wait until the day we can be together as a family again in Heaven.

Mimi, Andrew, Stuie and Henry x

IN MEMORY

Laura Elizabeth Bletsoe

28th April 1999 – 2nd October 2019

Laura loved and was loved. She enriched the lives of all who knew her. She was our songbird, our sunshine, and our inspiration. Laura was full of fun and adventure. She loved to play games and had an easy laugh. She mastered Braille Uno, loved to play word games, and make up stories. She treasured her library of audiobooks. Her favourite being *The Chronicles of Narnia*.

Epic is a word that was often used to describe Laura. Her entry into this world was epic, as she took less than 1 hour to arrive, and we barely made it to the hospital on time. Her exit was also epic, sudden, and traumatic. She died at Heather House in Tadley due to a series of complications relating to her feeding tube.

Her life was short but impactful. Laura achieved many things including awards for blind dressage riding, running (with a guide), singing and fundraising. Laura was rightly proud of her achievements. Despite the cruelty of Batten disease, she maintained her sense of place and purpose throughout her life.

Family and friends were hugely important to Laura. She adored her brother and her cousins. She had an amazing ability to remember everyone's birthdays (unlike her mother). Laura loved to go shopping for birthday and Christmas gifts. She would take a great deal of care to get exactly the right thing. This was often a challenge for the person assisting.

Laura had a very clearly defined and exacting opinion on many matters. Once an idea had formed in her head it was virtually impossible to persuade her from it. This played out in almost every aspect of her life, from choosing a DVD to what to have for breakfast. Laura helped us to master the art of patience.

In her lifetime, Laura had three residential placements outside of our home. These were New College Worcester, Linden Lodge and Heather House. We found much amusement in watching how each of these establishments adapted to accommodate Laura. Each placement would start by explaining their daily routines and how Laura would fit in. After a short time, each would tell us how they found that things run much more

smoothly when Laura gets her own way. For example, residents at Heather House routinely had a big full English breakfast just once a week as a treat. Laura liked to have sausage, bacon and egg every morning. Need I say more?

Laura's ambition was to run a riding stable, get married and have four children: two boys and two girls. She didn't achieve any of these things, but all those of us who knew and loved her, are better people because of her. That's quite a legacy to leave behind.

Laura's funeral service was held on 25th October 2019. This poem is from that service:

A Butterfly Lights Beside Us
Author Unknown

A butterfly lights beside us, like a sunbeam …
And for a brief moment, its glory
And beauty belong to our world …
But then it flies on again, and although
We wish it could have stayed,
We are so thankful to have seen it at all.

Miss you Laura xxxx

The In Memory section is fundamental to our newsletters. If you would like to include a tribute to your loved one with Batten disease please email Amanda on: amandamortensen@bdfa-uk.org.uk