



Batten Disease Newsletter

Bringing light to Batten's

Issue 22 Spring 2011

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



Batten Disease Family Association

Registered Charity No. 1084908

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Our Chair, Pauline Docherty writes...

Dear Members,



Pauline, Jim and James

Welcome to the first newsletter for 2011! I hope you find this edition interesting and informative – we are keen to receive your comments, so please let us know what you think.

First of all, I would like to join Andrea in thanking Julie for the amazing work she has done as a Trustee and as Chair. I do not believe we would be in such a strong position if it hadn't been for the time and commitment Julie has given the BDFA.

Julie, we all wish you the very best for the future.

So, as you will have realised, my name is Pauline Docherty and I have recently become the Chair – a role I am proud to have taken on. For those of you who do not know me, my connection to Batten Disease is through both my children who had/have Juvenile Batten Disease. My daughter, Joanna, was born in 1984 and my son, James, in 1990. For the first five years of their lives, both developed just as any parent would hope and it was only after several years of misdiagnosis that we finally received the scary, horrific diagnosis of JBD for Jo – subsequent tests confirmed that James also had it. It is difficult to sum up how your life changes once you receive the confirmation of JBD – watching my daughter and my son deteriorate, being taken over by this cruellest of diseases and knowing the inevitable outcome which eventually came for Jo – it has to be the hardest thing a parent has to do.

At the same time, we have met some amazing people along the way, made many friends we would not have met and we have done our best to do as much with Jo and James as they could enjoy. They have been surrounded by love and we have lots of very special memories.

Jo died in April 2008 aged 23, James will be 21 in January 2011 and both have been lovingly looked after by Heather House. I believe we have been particularly lucky in the help and support we have had – but in the early years, the external information and support that was available for parents was very poor.

And this is where the BDFA comes in – I was one of the original members who started our Charity and I am so proud of where we have got to in a relatively short space of time.

Our aim is to ensure that no family or carer should go through this journey alone, that professionals know to give families our details

and that families know that help is always available. Being able to talk to others who understand is a great comfort and the expertise that as an organisation we can now call on is improving all the time. We have a great team in Andrea, Sam and Georgette (it seems as if Andrea has been with us forever) so, thanks to them for their hard work. Thanks to you, as well for your continued support. Please help us to continue with our work to give support to families, and invest in research and raise general awareness.

Pauline

From the Charity Manager



Andrea

It is hard to believe that it has only been a few months since I was writing my introduction to the BDFA for the last newsletter but time has flown by during what has been a very busy time for the staff, trustees and supporters.

When I took on the role of Charity Manager in September I came into a vibrant, thriving organisation with a

strong team of staff and supporters who I know will enable us to continue to face the significant challenges and difficult economic times ahead. Our hugely successful family conference and AGM gave us the opportunity to say goodbye to Julie Pickering in her role as Chair of the BDFA. Julie has been an invaluable driving force in the achievements of the BDFA as an organisation that facilitates Batten's research, supports families and raises awareness of this devastating disease. I also know that she has personally been a huge support to those families living with Batten's and whilst we all recognise that the time is right for her and her family to move on, we will miss them all.

I want to take this opportunity to welcome our new Chair of Trustees, Pauline Docherty who introduces herself on this page. Pauline, with our committed board of Trustees will continue to help the BDFA to achieve its future vision, but we are also looking for new Trustees to join the Board and if you feel you have skills and experience you would like to offer please see the advert on page 5.

Looking ahead, 2011 is set to be an exciting year for the BDFA and Batten Disease in the UK. Sam Simpson continues to provide invaluable support and information to families in her Family Support Officer role and you can read more about her work and upcoming events in 2011 on page 3. Funded by the Roald Dahl's Marvellous Children's Charity, this role has become intrinsic to the vision of the BDFA to reduce isolation and support families living with Batten Disease. With funding for this role due to finish in September 2011, we are all working hard to maintain this important resource for the future. Due to the fantastic efforts of Georgette Ward, our fundraising officer, and our supporters, we are delighted to be able to offer a programme of grant funding for research into Batten Disease. The details of these opportunities are outlined in our Research Pages and on our website, and they show a new development for the BDFA as it is the first time we are able to offer to fund a PhD student for 3 years.

Cover photo: Laura Bletsoe scaling the climbing net with support from Zena Gallyer, Deputy Manager of Fir Tree Lodge, at the children's activities during this year's Family Conference.

I hope you enjoy reading about the work that has been completed over the last few months and thank you for your continued support. It is important to everyone at the BDFA that our publications reflect what you as members would like to see, so if you have any contributions such as stories, poems, fundraising ideas, photographs or articles that you would like to submit please let us know.

All of these achievements can only happen because everyone is working very hard to reach our goals, the more we do the more we can achieve to bring light to Batten Disease.

Andrea

Andrea West, Charity Manager

Update from the Family Support Officer

It has been a busy few months since our last newsletter and there has been a lot of activity in the support office. Back in October we had our 12th Annual Family Conference and AGM where we welcomed over 70 adults and 20 children and young people – our biggest conference so far! It was wonderful to see so many of you there and to welcome some new faces.



Read more about the conference on pages 4 and 5.

Shortly after our conference we held our 'Living Life with Batten's' workshop for families and professionals caring for a young person with Juvenile Batten Disease. Over 20 family members and professionals joined us at Heather House where we held an informal discussion session addressing some of the topics raised by those who attended. It was a very different style of workshop to those that we have previously held and feedback so far has been extremely positive. We now look towards our next workshop to be held on 5th May 2011 for families and professionals caring for a child with Infantile or Late Infantile Batten Disease. Details of the day and booking information are on the back page of this newsletter.

2011 promises to be an exciting time for the BDFA and the way we support families, with developments in our information in the form of updating our Information Sheets, the anticipated launch of our Late Infantile Information Resource book, (followed by versions for Juvenile and Infantile Batten Disease) and developments in our e-resources; making it easier for you to access the information and support that you need.

Don't forget that we have a wide range of library materials that can be sent out on loan free of charge to anyone who would like them. For more information on the publications that are available please look at the library subsection of the support area of our website.

Remember that we are here to support you. If you have a query, comment or suggestion, or you would just like to speak to someone, then please contact me on 01233 639526 or support@bdfa-org.uk

Sam



Sam Simpson, BDFA Family Support Officer
(funded by Roald Dahl's Marvellous Children's Charity)

News from the Fundraising Officer



Thank you for all that you are doing to help our work to grow. It is through the donations that you give, the events that you organise and the opportunities that you help us to seize, that our charity continues to develop – even during the hard economic times that we are all facing. As the BDFA's Fundraising Officer I love to support fundraising ideas that you might like to take on – or just to hear any ideas that you may have for our charity. So please do be in touch, fundraising@bdfa-org.uk or 01603 760111. Together we can bring light to Batten's.

Georgette

Georgette Ward, Fundraising Officer

Baking for Batten's - a Great Success!

Last October we held our first Baking for Batten's month where supporters across the UK took part in their own baking events; coffee mornings, cake sales and general baking activities were enjoyed while raising vital money to support our work and creating further awareness about Batten's. Our next Baking for Batten's event is set for October 2011 so why not take part and organise your own baking event?

Sponsored Walk for Batten's 2011

On Saturday 21st May 2011 we will be holding our sponsored walk for Batten's. This year we would like you to help us raise £20,000 to support our work. Your sponsorship helps to fund our helpline, our Batten Disease training days, information leaflets and so much more. This year's walk will take place in Southwell, Nottinghamshire. You can choose to walk the shorter morning walk of 5 miles which is fully accessible, or the full 9 mile walk. We have a good rate secured at the nearby Holiday Inn and will be organising a group meal with quiz on the Saturday evening. To request your walk pack please e-mail: fundraising@bdfa-org.uk or call 01603 760111. Let's step out for Batten's!

Ebay for Batten's

This May it's eBay for Batten's month at the BDFA. This is the chance to put your unwanted items for sale on eBay and help us raise money at the same time by nominating the BDFA to receive a percentage of your final selling price. To find out how to sell your item please visit our fundraising pages of our website: www.bdfa-org.uk and follow the simple steps. Soon you and your friends can be eBaying for Batten's!

**PLEASE NOTE: our address has changed to
PO Box 504, Fleet, Hampshire, GU51 9GE**

'Bringing People Together'



Pauline Docherty and Julie Pickering

BDFa Annual Family Conference and AGM

On Saturday 16th October 2010 we welcomed over 70 families and professionals and 20 children to Heather House in Tadley for our annual family conference and AGM. The day began with our AGM where attendees had updates on the work that the BDFa have been doing over the last year.

It was at the AGM that Julie Pickering, Chair of the BDFa, stood down after 3 years supporting the BDFa in the role. The BDFa took this opportunity to thank Julie for all of her hard work for the charity and the tremendous support that she has provided to the families over the years. We then welcomed Pauline Docherty, former support team trustee, as our new chair.



Sara, Jon and Ruth

During the course of the day we heard a clinical update from Dr Ruth Williams, Consultant Paediatric Neurologist and scientific updates from Dr Jon Cooper and Dr Sara Mole, Research Scientists. Sarah Kenrick, Registered Manager of Heather House and BDFa Trustee, also shared her presentation on person centred care and Batten's with a small 'b'.

There were several opportunities during the day for attendees to join separate discussion groups for each of the 3 main types of Batten Disease and also Mums, Dads, Adult Sibling and Wider Family and Professionals discussion groups. These sessions allowed each group to discuss the issues that were relevant to them and to look towards how the BDFa can support them with some of these in the future.

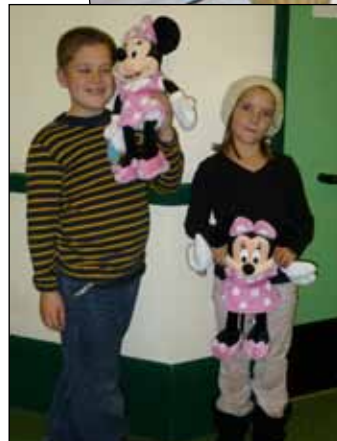


Fun at the Park

Sam, James and Pauline



Rocco Oran and Abdula Abbasi



Max and Angel McLeod



Laura Bletsoe

Whilst all the adults were busy at the conference the children went off on an adventure to a local wildlife park where fun was had by all! At the end of the conference the BDFA were delighted to be able to give the children a special gift of a cuddly Disney character, kindly donated by the Disney Store in Norwich.

After the conference ended it was back to the Apollo Hotel for a well deserved drink and a relaxing evening meal. We would like to say thank you to everyone who helped to make this year's conference possible. It was wonderful to see so many of you this year and we look forward to seeing you again in 2011!

Here's what one family made of the day...



"We travelled down, in two cars, from York on the Friday to Basingstoke. There was Heather (5) my youngest granddaughter, who was diagnosed with Late Infantile Batten's Disease, last January. She was in the first car with my son Brian and his wife Becky, and my eldest granddaughter Corinne (12). I travelled with Holly (7) my middle granddaughter and my niece, Jo, who was hesitant to come along as she felt a bit of an interloper but offered to drive us and turned out to be a great support.

As a family this was the first annual BDFA conference we had attended. In fact it was the first most of us had had anything to do with the BDFA and to say some of us were unsure whether we wanted to attend would be an understatement. I mean being in a room full of strangers, feeling vulnerable, not sure if we could keep our emotions in check, all of this and much more was going through our minds.

Because Batten's is so rare, we felt no-one really understood what we were going through, even among our own family group. I've been devastated by this disease but how could that possibly compare to how my son and his wife are feeling? I'm Heather's grandmother but that day, at Heather House, within our own groups, you had the feeling of empathy and a certain amount of camaraderie. These people not only knew how you felt, you knew how they were feeling because we were all on the same journey, some had started before others but we had all started off at the same point with the despair and bewilderment of how had this happened to your precious child? Why had this happened to your family?

We learned a lot about how Batten's progresses, the barriers you have to get over to have your child's interests served, and the professionals that are available to help you achieve this. We also learned that most people want to help but often don't know how they can so you have to ask them. The most important thing we learned was from the families and carers who, by way of showing us how they coped, gave us the tools to help us cope and live with Batten's."

Jan Atkinson

Trustees Needed

The BDFA are looking to recruit Trustees to provide governance and to guide the Charity to meet its strategic aims for the future. Specifically we are looking for individuals with fundraising experience but are keen to talk to people who are committed to raising awareness, facilitating research and supporting families living with Batten Disease. Should you require further information, wish to discuss or formally show interest in becoming a Trustee, please contact Andrea West, Charity Manager 01252 416110 info@bdfa-org.uk

Dates for your Diary

April 2011	London Marathon
5th May	Living Life with Batten's Infantile/Late Infantile Event
May 2011	eBaying for Batten's month
21st May 2011	Sponsored walk for Batten's
3rd & 4th June 2011	Batten Disease Awareness Day
14th June 2011	Lab Day
10th July 2011	London 10K Run
8th October 2011	AGM & Family Networking
October 2011	Baking for Batten's month
27th - 31st March 2012	NCL 2012

SAVE THE DATE...

Batten Disease Awareness Day 2011

On 3rd and 4th June 2011 we will be promoting Batten Disease Awareness Day. Why not order your T-shirts and our NEW ribbons and wristbands to do your bit to raise awareness? Don't forget to spread the word to family and friends too... Together we can bring light to Batten's.

For more information get in touch with Georgette Ward on 01603 760111 or fundraising@bdfa-uk.org.uk

Have we got your current email address? Don't forget that we can keep you posted on updates and events via email too! Please contact Sam on support@bdfa-uk.org.uk to update your details.

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Making contact with other families...

Our son was diagnosed with Infantile Batten Disease in January this year. James was 22 months old at the time. It was not the diagnosis we expected and came as a complete shock. Our world fell apart and we felt completely alone. All the professionals we met had never encountered Batten Disease and were unable to support in the way we needed. Our local GP gave us the contact information for the BDFA and suggested that it would be good for us to get in touch and make contact with other families. To be honest this initially terrified us! We were frightened about what we would be told. We took some time to adjust to James' diagnosis and when the time was right for us we contacted the BDFA who put us in touch with other families. We have not looked back since! Talking to other families, usually through email, has been a real comfort. We no longer feel isolated. We are now in regular contact with other families who understand our worries and can help us with the everyday difficulties we find throughout our Batten's journey. We can be just having a bad day or needing advice about medication or tried and tested equipment. There is always someone to help. We would be truly lost without the Family Networking Scheme.

Amanda and Ian Harrison

Would you like the opportunity to make contact with other families? For more information about our family networking scheme then please contact Sam Simpson on support@bdfa-org.uk or 01233 639526.

Grants to Individuals and Families from Roald Dahl's Marvellous Children's Charity



Roald Dahl's Marvellous Children's Charity (formerly known as The Roald Dahl Foundation) is very pleased to have been able to support the work of the Batten Disease Family Association by funding the post of Family Support Officer. Since being set up by Roald Dahl's widow, the Charity has made grants across the UK totalling more than £7m – funding charities, specialist NHS nurses and grants to individual children and families. The Charity's grants to individuals provide practical help to children (and their families) who meet our criteria for haematology and neurology, including Batten Disease. We help with needs caused by the medical condition that the child is living with. The family must also meet our financial criteria and must be in receipt of income support, working tax credit, housing benefit or have an earned income under £22,000 per year. Grants must be applied for on behalf of the family by social services, a specialist charity or a healthcare professional. Our maximum grant is £500 but applicants need to apply for a specific purpose for which they will need to give costings. Further information and application forms may be downloaded from our website at www.marvellouschildrenscharity.org and follow the link to grants to individuals or contact our Grants to Individuals Manager on 01494 892170 or email: individualgrants@marvellouschildrenscharity.org

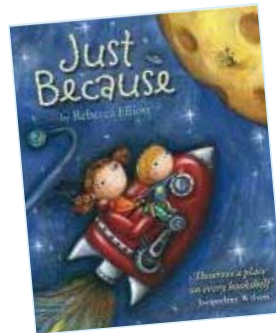
Siblings Corner...

Here at the BDFA we are keen to develop the support that we can offer to all members of the family. We know that each family member is unique in the support that they require when a member of their family has a diagnosis of Batten Disease. We are aware from discussions with siblings in particular that they are one group which do not currently feel that they have an opportunity to network with one another directly and would like to do so. If you are a sibling and would like to be involved in supporting a scheme to assist siblings to network with one another then please get in touch with our Family Support Officer, Sam Simpson on support@bdfa-uk.org.uk and register your interest.

Just Because...

"My big sister Clemmie is my best friend. She can't walk, talk, move around much... cook macaroni, pilot a plane, juggle, or do algebra. I don't know why she doesn't do these things. Just because."

My daughter Clementine was born with profound mental and physical disabilities, whilst I am painfully aware of the hardships and heartbreaking consequences of parenting such a child more than anything she has brought into our family immense happiness and joy - she is perfect! Unfortunately there are very few children's books featuring severely disabled children and I wanted to share some of Clemmie's wonderfulness with the world so I wrote 'Just Because'. It's a fun, funny, heart warming picture book aimed at all children about Clemmie and her beautiful relationship with



her younger brother Toby. It was published in August and so far the response has been immense – I think disabled children and their siblings enjoy reading a book which features characters they can relate to whilst other children can still relate to the main themes of sibling love and adventure and just take Clemmie's 'differentness' at face value and accept her as she is. Children are far better than us adults at that!"

Rebecca Elliot
(Children's writer and illustrator)

'Just Because' is widely available to buy. The BDFA also has a library copy which is available for loan. For more information contact support@bdfa-uk.org.uk or 01233 639526.



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



Hannah's Wish Comes True!



Back in March this year, I decided to contact Make-A-Wish to see if they could grant my daughter, Hannah, a wish.

She was diagnosed with Juvenile Batten Disease 4 years ago, and probably like most families who receive that awful news, I was set on making Hannah's time with me as special as possible.

We've been to lots of

places and done lots of things, but I wanted her to have her own personal wish that I didn't influence.

The lovely volunteers from Make-A-Wish arrived in April to talk to Hannah about her wish. She's quite a shy girl at first, so it took them a while to establish that she is Harry Potter mad! Once they knew this, ideas and suggestions were thrown around and they went away with a few ideas.

A couple of weeks later, Hannah received a letter stating they were going to send us (Hannah, me, my partner Nick and his son Owen) to Florida for a week in November all expenses paid to visit The new Wizarding World of Harry Potter in Universal Studios !! Hannah was absolutely delighted!

The day finally came and we were taken to Manchester the night before the flight to enjoy a meal and relaxing evening. The flight was problem-free and we arrived in Florida to a sunny 84 degrees. We were met by a representative from Give Kids The World Village (where we were staying) and given a free rental car! Hannah loved the car with its automatic sliding rear doors!

Give Kids The World is the most amazing place I have ever been! Nearly the whole place is run by volunteers, with everything being donated by local and international businesses. Everything on site is free – food, drinks, ice cream (as much as you want – even at 7.30am!). We were given free passes to Walt Disney World, Universal Studios and SeaWorld – including a special badge for Hannah to wear which meant we didn't have to queue for any rides! Truly fantastic!

The week passed quickly but the two days Hannah spent in The Wizardly World of Harry Potter are ones we are never going to forget! Hannah drinking Butterbeer and Owen being selected in Olivanders for a special wand are just two special memories. Another is Lilo & Stitch in Walt Disney's Animal Kingdom, who upon realising Hannah is blind, spent time getting her to feel their faces – truly emotional.

Debbie White

Recognition for Nurse

Katarzyna Wentka, a senior nurse at SeeAbility's Heather House has won a special award in recognition for her outstanding contribution to developing a new system for the care of young people with Juvenile Batten Disease (JBD).

SeeAbility's Heather House has a worldwide reputation as a leading palliative care home for young people with JBD. Due to the degenerative and ultimately terminal nature of the condition, the process of nursing individuals changes according to their needs. Despite the challenges, the team at Heather House always ensures that the quality of life of the young people they care for is maximised through a wide range of therapeutic treatments and activities. End-of-life care has always been an emotive and difficult subject and consequently Katarzyna's review of best practice in this area has led to a groundbreaking system of nursing.



Katarzyna's approach, involved a review of current evidence and best practice of palliative care and introducing new staff training. She developed innovative new tools such as pain scales and end-of-life care plans. Katarzyna acknowledges the invaluable role families can play in the decision-making process of end-of-life care: Katarzyna used her creative skills to design a room at Heather House where families can spend reflective time to come to terms with their impending loss and

she works closely with family members to determine an appropriate schedule for emergency care and advance care planning.

Sarah Kenrick, Registered Manager of SeeAbility's Heather House comments, "We are delighted that Katarzyna received this special award and it is thoroughly deserved. Katarzyna has shown tremendous resourcefulness and dedication in addressing the difficult issues surrounding the final stages of a young person's life. She has developed a unique approach to end-of-life care which is one of the areas we all find difficult and Katarzyna's ability to mentor others as well as working closely with families has been a notable achievement."

For more information about SeeAbility's work with Juvenile Batten's Disease visit www.seeability.org.uk/ourservices

Do you have something you would like to share?

This is your newsletter and we would like it to reflect your voices as families and ensure that we share information that is useful to you. If you have a story that you would like to write for the newsletter or there is something that you would like us to feature then please get in touch with Sam Simpson on support@bdfa-uk.org.uk or 01233 639526.

Resources for Bereaved Children

The understanding that a child has about death is dependent upon their stage of development and any other losses that they may have experienced in their lives. What we do know is that they need to be supported to share their feelings and to be given every opportunity to remember the person that they have lost.

The Child Bereavement Charity is an organisation that supports families and educates professionals when a child dies and when a child is bereaved. They have a wide range of resources, a support and information service, online forums and a Buckinghamshire-based family bereavement support service. We would like to share two of the books that are sold by the Child Bereavement Charity which are designed to help children explore their feelings and to remember the person that has died.



'Someone I Know has Died' by Trish Phillips (£9.99)

This is an innovative activity book with many unusual and interactive features, including tags to pull and dials to turn. This book was written for bereaved children to help them understand what it means when someone dies and to explore their thoughts and feelings with an adult. Although designed with pre-school and early years age groups in mind, some older children may also enjoy this book. There is lots of space to write and draw and even a 'sedomometer' allowing a child to show an adult how they are feeling. There is special paper chest at the end of the book that allows a child to write a message to the person that they have lost and store it safely inside.

'Remembering' by Dianne Leutner (£5.00)

'Remembering' is a keepsake/memory book for children when someone special in their life has died. It is part book, part scrapbook and was created to help keep a child's memories alive after the loss of someone special and to give them a place to return to whenever they wish. There are pages for a child to draw pictures and write all about their special person as well as extra space at the end which can be used for any writing, drawing or photographs.



These books can be ordered from The Child Bereavement Charity's website: www.childbereavement.org.uk. The BDFA also has a library copy of each of these books which are available for loan free of charge. For further information contact Sam Simpson on support@bdfa-uk.org.uk or 01233 639526.

Remembrance



In Loving Memory

Our thoughts and prayers are with the families and friends of:

Stuart Ian Bayliss, aged 29 (Juvenile)

Jake Michael Nelki-Smart aged 4 (Late Infantile)

Daniel Smith, aged 32 (Juvenile)

Corbyn Thomas Mason Brown, aged 8 (Late Infantile)

Saoirse Heffernan, aged 5 (Late Infantile)



The BDFA is here to support families at all stages of the Batten's journey. We can offer a listening ear, put you in touch with another bereaved family and other support organisations.

We believe that our families deserve the best possible emotional support and an opportunity to remember their child in a special way. The BDFA has a Remembrance Book for families to share memories as a tribute to their child. If you would like a page to go into the book then please get in touch with Sam Simpson, Family Support Officer on support@bdfa-uk.org.uk or 01233 639526 for further information.

Grants for Scientific Research

We are now into our 3rd year of awarding grants for scientific research and have now launched our new grant call for grants for 2010-2011. Due to the fantastic efforts of all of those who have supported the BDFA over the last year, in November we were able to announce a new grant funding programme in addition to our current programme for research in the field of Batten Disease in the UK. For 2010-2011 we are again pleased to continue to support researchers by awarding research grants to UK scientists under our existing scheme.

The award levels are:

Tier 1 up to £500

Applications in this category will be used for scientists to attend conferences, seminars, workshops and to aid them in the publication of scientific literature to promote and increased awareness of Batten Disease in the scientific community and beyond.

Tier 2 greater than £500 up to £5000

This funding is designed to provide much needed part-funding for new or existing research projects allowing scientists to gain valuable initial data to help them apply for larger grants from other sources, or to support for research studentships.

These have proved very successful and articles from Jon Cooper reporting from the Australian Batten Disease Association conference, and from Brenda Williams about her latest research are featured in this issues research pages.

Substantial funds raised by Ascot Underwriting combined with our own funds have enabled us to offer:

- Higher awards greater than £5,000 for 2010-2011
- In this new category for 2010/11 the BDFA are seeking to fund two types of Higher Award grants for research in Batten Disease. These will be 2 awards, up to a maximum of £10,000 per award for Juvenile Batten Disease Research Projects
- And most excitingly a fully funded PhD studentship award of £100,000 over 3 years.

In the coming year over £130,000 will be awarded to Batten Disease research in the UK and these awards represent an enormous achievement for everyone who has been involved with the BDFA.

We look forward to giving further information on the projects which have been awarded in our next newsletter. We are also continuing to follow up and evaluate our previous research awards and look forward to NCL 2012 – put the date in your diary!

Heather Band
Research Trustee



Rare Disease UK is a group of key stakeholders who work together to develop strategic planning for rare diseases in the UK. They also work with Genetic Alliance UK, the national charity of over 130 patient organisations supporting all those affected by genetic disorders. They recently published a report: Experiences of Rare Diseases: an insight from patients and families. It can be downloaded from the news section of the BDFA website: www.bdfa-uk.org.uk and more information is available at the Rare Disease UK website: www.raredisease.org.uk



The BDFA received a **Jeans for Genes Grant** which enabled us to commission Dr Ruth Williams at the Evelina Children's Hospital to develop a standardised scheme for the clinical assessment of children and young people with Batten Disease.

It is envisaged that this scheme would be used in clinical and educational settings to help predict future needs and monitor symptoms and disease progression. The final research report is being compiled and will be shared with members in due course.

NCL 2012 the 13th International Congress on Batten Disease AND 1st Parents Organisation International Conference

This will take place at the Royal Holloway College,
London on 27th - 31st March 2012
Updates will be posted at www.ncl2012.org

This is an important opportunity for families to access the latest scientific research and to meet those working within the Batten's field. Over the next couple of months the BDFA will be asking for feedback on what families would like from the conference but if you have any ideas of what you would like to see at the conference please contact Sam Simpson, BDFA Family Support Officer, 01233 639526, support@bdfa-uk.org.uk.

Pediatric Storage Disorders (PSDL) Lab Open Day Tuesday 14th June 2010



The PSDL's next annual Lab Open Day will take place on Tuesday 14th June starting at 10.30am (with tea and coffee served from 10.00am). As usual the day will consist of short presentations from PSDL lab members, an update on the team's work and an overview of progress towards therapies. It is a great opportunity to see the work of Dr Jon Cooper's team, to hear how the BDFA is funding research and to ask questions. All parents and others interested in the NCLs are very welcome. For further information please contact Sam Simpson, BDFA Family Support Officer, 01233 639526, support@bdfa-uk.org.uk

Astrocyte dysfunction in Juvenile Batten Disease

Lotta Parviainen, Jonathan Cooper, Brenda Williams

Pediatric Storage Disorders Laboratory and Department of Neuroscience, Centre for the Cellular Basis of Behaviour, Institute of Psychiatry, King's College London

One of the major goals of our research group is to discover which of the different sorts of cells that make up the brain are most affected by Batten Disease. We've found where and when nerve cells (neurons) are lost, but are now turning our attention to the 'glial' support cells that help these nerve cells to work properly. Our work suggests that these glia are also impacted by disease and this may open up new therapeutic avenues, using drugs to specifically treat these cells.

Astrocytes – not just 'brain glue' after all? Glial cells come in 3 varieties (astrocytes, microglia and oligodendrocytes), each having important jobs to do. Although astrocytes were originally thought to purely play a supportive role, it is now clear that these star shaped cells are crucial for the development, survival and proper functioning of neurons. This means that anything that affects astrocytes is likely to cause severe problems for neurons too.

Astrocytes as key players in disease progression. Any sort of brain injury or disease results in the astrocytes becoming activated or 'switched on'. To do this astrocytes change their shape, divide and start releasing into the local environment a range of chemical messengers that can have a powerful influence upon both neurons and other glial cells. Depending on the situation, this activation of astrocytes can have either a positive or negative influence on the disease. It will be important to find out which sort of influence astrocytes have in Batten Disease.

Role of astrocytes in Batten Disease. We have discovered that in all types of Batten Disease both astrocytes and microglia, (the other sort of glia cell that makes up the brain's immune system), are activated very early in the disease, long before brain cells start to die. However, in Juvenile Batten Disease (JNCL), this activation process appears to 'get stuck' in its early stages. Both astrocytes and microglia switch on a range of activation markers, but fail to change their shape properly. This suggests that the biology of these cells is compromised in JNCL. As some of you may have seen at the lab's open day earlier this year, we have been investigating this possibility by growing astrocytes in tissue culture, thus allowing us to explore the biology of these cells in a highly controlled environment. What we do is grow astrocytes from healthy and JNCL mice and stimulate them to see how they respond. This valuable research, and what we have discovered, would not have been possible without financial support from the BDFa.

What have we discovered so far? When we stimulate astrocytes from healthy unaffected mice they behave as expected; they change shape, divide and release a range of different chemicals. However, if we stimulate astrocytes from a mouse model of JNCL (*Cln3* deficient mice) they don't change their shape, and don't proliferate, mirroring what we see in the JNCL brain. These diseased astrocytes release reduced levels of key chemical messengers, as well as the important anti-oxidant glutathione. All of these defects may have negative effects on neuron survival and so might be an important part of the disease process in JNCL.

What remains to be discovered? We now need to find out if these unhealthy astrocytes really do have a direct effect on the health of neurons. We will do this by growing these two cell types together and seeing if this makes neurons get sicker and whether we can block this. Such studies will pave the way for testing whether drugs that target astrocytes would be effective in treating JNCL.



Brenda Williams



Lotta Parviainen



Lotta looking at astrocytes under the microscope

A long way to go for a weekend...

Conference report from the Australian BDSRA Family Conference, Sea World, Gold Coast, October 8th - 10th 2010
Jon Cooper, Institute of Psychiatry, King's College London

As any of you who have been to the BDFA's conferences will know well, the chance to get together with other Batten parents is invaluable. Knowing that there are others going through similar experiences is all-important and time spent seeing each other for a mix of science and support updates, plus sharing social activities is very precious. This is especially the case for the families 'down under', since Australia is a big country and families are scattered over long distances, with several more being over in New Zealand. Their family conference also only takes place every two years, so these families don't meet up very often and as we all know a lot can happen in a Batten family over two years.

Being relatively isolated, literally on the other side of the world, it's also difficult for these 'Oz/NZ' families to meet up with the scientists who are doing the research. This being a time when new clinical trials are starting and scientific papers regularly appearing with new information. As such, it was especially important that there would be someone there who is involved in these studies to give a first hand account of what is going on, provide up to date information and to answer questions from families. Thanks to the generous support of the BDFA and Australian BDSRA, I was able to be that person and attend the conference, not just as a scientist but also to represent the BDFA. Since none of the other speakers from outside of Australia were able to be there, it was especially valuable that I was there.

I was lucky enough to be at the Australian BDSRA's first two family conferences, one of which was at the same venue in 2003, but various things had stopped me from getting back since then. Vanessa Anderson, who organised the conference, was at the US BDSRA meeting in St Louis last year and persuaded me it would be a good idea to return. The prospect of flying to Australia just as the autumn starts to get cold might seem an attractive one. However, being right in the middle of my busiest teaching period, just before two big grants were due AND two PhD students finished their theses (!) it was always going to have to be a 'quick' visit, especially as there was the BDFA annual conference the next weekend!

Having a Batten family conference at a holiday resort can be a rather surreal experience, with the hotel full of tourists since this was the last week of the Australian school holidays. Sadly, the stereotyped vision of a sun kissed Gold Coast failed to materialise, as there was a tropical rainstorm for most of the three days we were there. But as UK parents can testify it takes more than a 'little' rain and gale force winds to keep a good family conference quiet! The structure of the conference may have been similar to those we have at Heather House (talks, workshops and discussion groups), but everything had a typically Aussie/Kiwi twist. The conference dinner was naturally a 'barbie' and the Dad's group discussion time was held in the bar (with free beer tokens – something the BDFA Dads were all in favour of!) and somehow the evening charity auction DID get hosted by Vanessa's helicopter pilot husband Graeme wearing one of her dresses!



Social Dinner



Boys' Business

So was it worth two days travelling there, two days travelling back and jet lag? Yes, it certainly was. The chance for me to be there for a newly diagnosed family; to meet up with Batten Facebook friends for the first time; to talk with parents whose children have passed away; to just be a listening ear. The bonds that are formed over such long distances are very strong ones and bringing the worldwide Batten community closer together and strengthening those bonds is a wonderful thing. The BDFA has played a key role in doing this by supporting a meeting between organisations from Europe and the US. With the first International Parent Organisation Conference planned to run together with the NCL2012 science congress next year, this was a perfect time to be reinforcing those links with the families in Australia and New Zealand. I would like to thank the BDFA and Australian BDSRA for making it possible for me to be part of that process.



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Thursday 5th May 2011

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For more information please contact
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