



# Batten Disease Newsletter

*Bringing light to Batten's*

Issue 20 Spring 2010

*from the UK's only dedicated Batten Disease support and research charity*

**Batten Disease Family Association**

**Registered Charity No. 1084908**

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*James Fitzpatrick (deputy director of the Roald Dahl Foundation),  
Ben & Julie Pickering and Sam Simpson*

Our Chair, Julie Pickering writes...

## Dear Members

Happy New Year. I hope you all found some warm enjoyment over the festive season despite the cold unpredictable weather.

We know the November to January period can be hard for families as winter settles in. For five families, including our own, it was time for their child affected by Batten disease to pass away. I am sure each individual family, with the loss of their child, feels as we do, and our thoughts are with them. As a family, we are deeply saddened with Ben passing away and 2010 will bring a new way of life in our home but we will never forget him and what he gave to us.

When we look back as a family at 2009, up to Ben passing away in November, we had an incredible year, with the BDFA being a big part of this. We see that yet again the BDFA has moved further towards its vision. In 2009 many great support initiatives were achieved, we held our first excellent training day for families and professionals linked to Infantile/Late Infantile Batten disease, recruited Sam Simpson, our Family Support Officer, funded by the Roald Dahl foundation and Shauna Bates, to be our Social Care Advisor. The year culminated in another wonderful family conference, a feature on which you can find on pages 4 and 5.

In conjunction with all of this we have been working with Dr Ruth Williams, our Medical Advisor, to submit an application for an NHS National specialist Batten Disease clinical service to be established at the Evelina Children's Hospital. This application was finally submitted in December and we will know the outcome by middle of 2010. If successful, it will transform the NHS Batten clinical service for all our families and professionals with effect from the autumn of 2011. Fingers crossed... these achievements are just some of what has happened in 2009.

In awarding research grants, we yet again increased both our funding and the number of projects we have supported. There are two articles

about work we have funded in the research news on pages 10 & 11. For 2010, we will continue to expand this even further. We hope over the next few years the BDFA funds available for research will grow significantly to enable us to facilitate much more research into Batten Disease. We are always looking for new and novel ways to support our scientists. Many of you have already made this possible through the amazing fundraising you have done... and plan to do in the future. As well as the valuable funds you raise, it gives others the feeling they are not alone and a little bit of hope. Attending the London Marathon and then the walk in 2009, was an amazing feeling for us as a family, both for the great days out they were and for giving us the feeling that we are able to do something positive within our life of Batten's.

On the fundraising side, Georgette Ward, our Fundraising Officer, has worked hard in her first year to ensure the fundraising side of the BDFA is solid and continues to grow with many new successful initiatives like the Batten Bulletin (please find your copy enclosed with this issue). Our available funds continue to grow with the expansion of these and your individual events, enabling us to do the work required, the support we provide and the research we facilitate. Thanks to all our members, their families and friends who help with these activities all year round.

We are now established as one of the top 15% of charities in the UK and are still continuing to grow. We look forward in 2010 to recruiting a Charity Manager to co-ordinate the BDFA's ongoing activities. Please take time to read the Trustee's 2008/09 report enclosed with this newsletter to really appreciate all the hard work that is being done. As well as our paid support and individual fundraisers and families, I would also like to thank all our volunteers, professional advisors and the Trustees for their continued input into the BDFA. Together we are making it happen... thank you.

If you want to get involved and help in anyway, please do contact us. As a special request, please really do think about promoting the 2nd Worldwide Batten disease Awareness Day on 5th June 2010. Georgette, our Fundraising Officer is always ready to embrace any new ideas... or just wear one of our orange BDFA Batten T-shirts for the day... (it gets you noticed!)... and I know from personal experience it will make a difference.

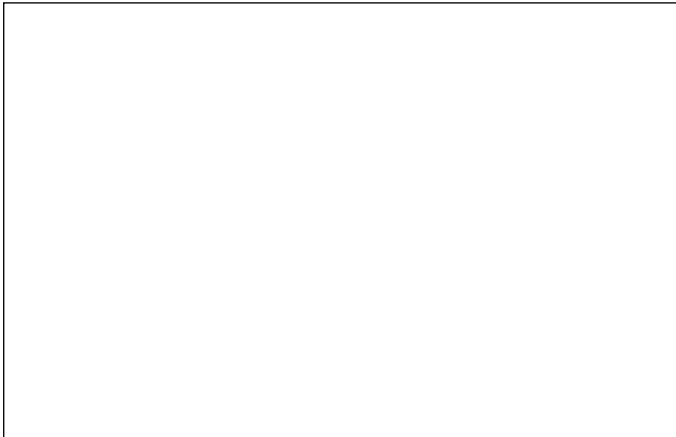
In the meantime, if we can help **you** in any way, please contact us.

*Julie*

## Forthcoming Events 2010

25th April	London Marathon (3 runners)
29th April	Living with Batten's – a new opportunity for Juvenile Batten Disease families, carers and professionals. Please see back page for details.
22nd May	BDFA Sponsored Walk
5th June	Batten Disease Awareness Day
15th June	Lab Day at Jon Cooper's Paediatric Storage Disorders Lab, in London
11th July	London 10K run
16th/17th October	BDFA Family Conference and AGM

[Cover photo: The children went to an Adventure Farm while their parents attended the AGM, and here are Laura Bletsoe and Anna Pickering having a great time on the swing!].



## Family Support Officer Update

Hello, I've been in post as Family Support Officer since the beginning of September. It has been an exciting time for me and my first three months have flown by. I've been busy learning the ropes and getting stuck into some exciting projects. The highlight for me so far has been having the opportunity to meet families and professionals at our annual Family Conference in October. One of my first tasks has been to support the organisation of "Living Life with Batten's", a new opportunity for families and professionals caring for a child with Juvenile Batten Disease. This event is set to take place on 29th April 2010 and the proposed agenda can be found on the back page of this newsletter. There will be a further "Living Life with Batten's" event in November 2010 which will focus on caring for young adults with Juvenile Batten Disease.

In the last two months I have also been working on our Family Networking Scheme, setting up support pages on the website, planning for the B DFA Family Conference 2010, managing membership, networking with other organisations and, most importantly, taking helpline calls and emails.

It has been a privilege to speak to so many families and professionals already, and I do hope that you will continue to get in touch with any support needs, questions or suggestions that you may have.

I look forward to hearing from you...

Best wishes,

*Sam*

support@bdfa-uk.org.uk  
01233 639526

Sam Simpson, B DFA Family Support Officer  
(funded by the Roald Dahl Foundation)



### Our website is growing!

Log onto [www.bdfa-uk.org.uk](http://www.bdfa-uk.org.uk) to see our Fundraising and **NEW** Support Pages.

## Fundraising Officer Update

I'm pleased to say that it has already been a busy start to the year with lots of supporters taking part in races, enquiring about our new tandem sky dives or organising their own local fundraising event. Our work is funded through your generosity and to enable us to continue, and to grow, we do need your support.

Please do look at our fundraising pages on our website, and the Batten Bulletin for new ways to fundraise. In the meantime we need supporters to help cheer our London Marathon Runners on Sunday 25th April, and for runners to take part in the British London 10K race on Sunday 11th July – could you help?

### Walking for Batten's

Our Fifth Annual Sponsored Walk for Batten's will take place on Saturday 22nd May 2010 at Bourton-on-the-Water, The Cotswolds. This is a great chance to raise vital funds. So why not bring along your friends, work colleagues, families – there is even a chance to make a weekend of it as we have secured an excellent rate at a nearby hotel.

The walk itself will be two loops – a shorter distance in the morning for all ages and abilities (approx. 5 miles) and then in the afternoon there is an additional loop (approx 6 miles). You can choose to do just the shorter morning walk or the whole route of 11 miles.

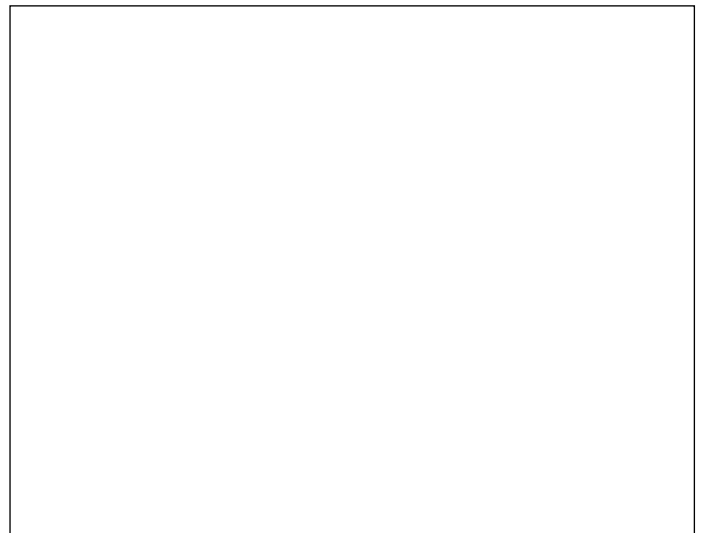
To sign up to this year's walk, or to take part in our other fundraising events, please visit: [www.bdfa-uk.org.uk](http://www.bdfa-uk.org.uk) or e-mail: [fundraising@bdfa-uk.org.uk](mailto:fundraising@bdfa-uk.org.uk) or call 01603 760111.

Together we can bring light to Batten's.

*Georgette*

[fundraising@bdfa-uk.org.uk](mailto:fundraising@bdfa-uk.org.uk)  
01603 760111

*Orange is the colour!! The 2009 B DFA Annual Sponsored Walk*



## BDFA Family Conference and AGM – Saturday 10th October 2009 – Heather House, Tadley, Hampshire

It was a pleasure to welcome so many families and professionals to our Annual Family Conference and AGM at SeeAbility Heather House in Hampshire. We had sunshine and a great turn out, with families and professionals from around the country coming together to learn more about Batten disease.

The children and young people enjoyed a trip to a nearby family adventure farm whilst the adults stayed at Heather House and took part in our workshops and forums. This year we focussed on aspects of social care and the help that Social Services provide. Thanks to all who contributed, especially our speakers, Shauna Bates & Anne Caldwell (Social Worker, The Children's Trust).

### Here is what some of our family members and supporters thought of the day...

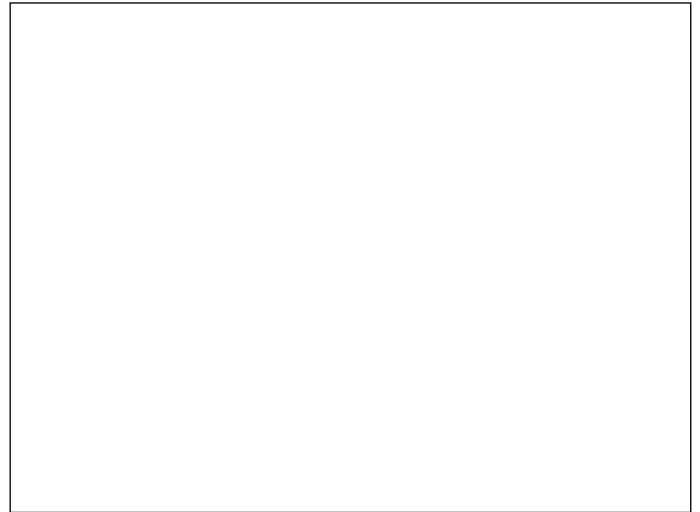
*"So, it was with some trepidation and interest that we attended the Family Conference. I had inferred that, like the website, it was going to be organised, informative, frank and approachable. And so it was. We were warmly welcomed by friendly faces. Those there understood our plight (of course). They were parents, professionals, researchers, doctors and staff. There were formal lectures on topics of interest to all, such as dealing with Social Services. There were smaller, flexible, group discussions on topics of a narrower interest, such as 'newcomers' like ourselves. And, very usefully, there were the informal discussions, contacts and networking during the breaks. We left shaken but fulfilled and with lots of useful information. The venue was excellent, the food was excellent and the atmosphere was excellent."*

**Michael Neliki**

*"The conference was enlightening and eye opening. It was nice to see other families and to have an opportunity to talk to Dr Ruth Williams."*

**Roger Dawkins**

**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](mailto:support@bdfa-uk.org.uk) to update your details.



*Shauna and her sisters ready for the journey home in their new BDFA T-shirts*

"On the 9th October this year my sisters Caroline and Catherine and I set off for our first BDFA conference. Although I had been to a BDSRA conference in 2007 this was my chance to see what we could do for Batten Disease on this side of the world and to meet families that I could hopefully form friendships with. Being from Northern Ireland it was quite a trek and although I would have loved for my daughter Rhiannon, who has Late Infantile Batten Disease and my husband, Alan, to join me it just wasn't possible at the time. I asked my sisters to keep me company and provide moral support as I presented two workshops for the first time in my role as the new BDFA Social Care Advisor.

I was so nervous about the workshops I was going to be giving that day. As a Social Worker I have always strived to do my best but the work I do for the BDFA means so much more to me. I am going to be trying to help people who are going through the same things as I am. I really know how they feel and what it's like to face the daily battles with various professionals.

The best thing that the weekend brought was the time that I got to spend with my sisters. Sometimes it's hard to explain all the different ways that Batten Disease affects me, my husband and Rhiannon. My sisters got a better understanding that weekend, we realise that there is no doubt that having a child in our family with Batten Disease has been hard, but the love she has brought has benefited us as a family in the most wonderful way."

**Shauna Bates**  
Social Care Advisor for the BDFA

**Shauna's husband Alan has shared with us a special poem about their daughter Rhiannon on page 8.**

Here are some photos of the children enjoying their time at the local adventure farm.



*Come on girls can you give me some help?*

*Ben Marlow, with Anna Pickering and Joy Parkinson*

"I really enjoyed my day out with my son Ben, whilst my wife, Heather (a B DFA Trustee) attended the conference. It was an opportunity for us to help look after siblings and enjoy some fresh air and fun at the local adventure farm. We all look forward to the B DFA conference each year as it is a great family time."

**Andrew Marlow**

*Zac Griffith & Ben Marlow on the move F1 style!*

## Meanwhile back at the conference...

Families, scientists and care professionals were also able to mingle and talk informally over tea between the talks and workshops.



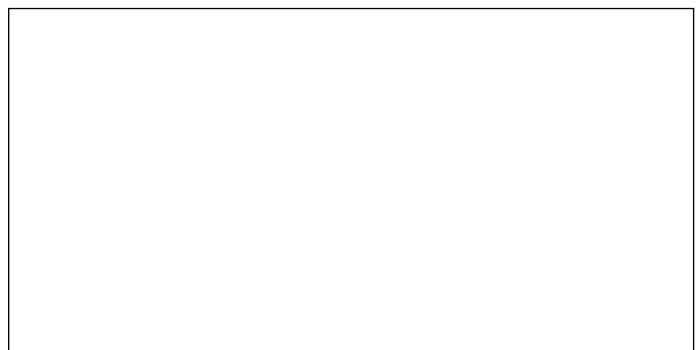
*Special thanks to Steve Bayliss for doing such a marvellous job supplying everyone with tea and coffee*

Scientists and professionals were able to give one-to-one sessions with parents.



*Heather Sickelmore learning about the latest research on CLN3 from Sara Mole*

We would like to thank SeeAbility Heather House for allowing us to use their wonderful facilities and for Sarah & her team for all their hard work on the day to support us. Special thanks to Marion, pictured below, with Julie for all her inspiration. We are already beginning to plan this year's Family Conference. Members will have received a questionnaire so that we can get your perspective on ideas for what format it could take. If you do feel able to complete and return the questionnaire, it will provide us with valuable feedback, to ensure that when we meet up on 16/17th October 2010 it is an even bigger success.

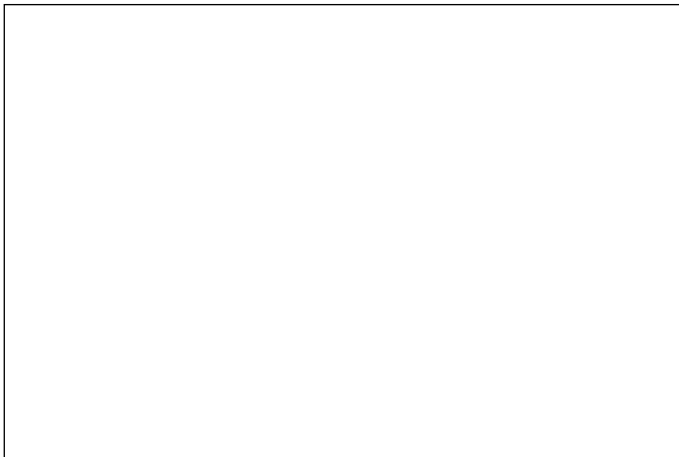


# Building the Team

*The BDFA's Vision is to bring light to Batten Disease by being the central point of excellence in the UK for supporting affected families and to facilitate research into the disease*

## Introducing the BDFA Trustees – who we are and what we do

Pictured below are **Sarah Kenrick**, Secretary, **Julie Pickering**, Chair and **Alison Jenkins** our Treasurer.



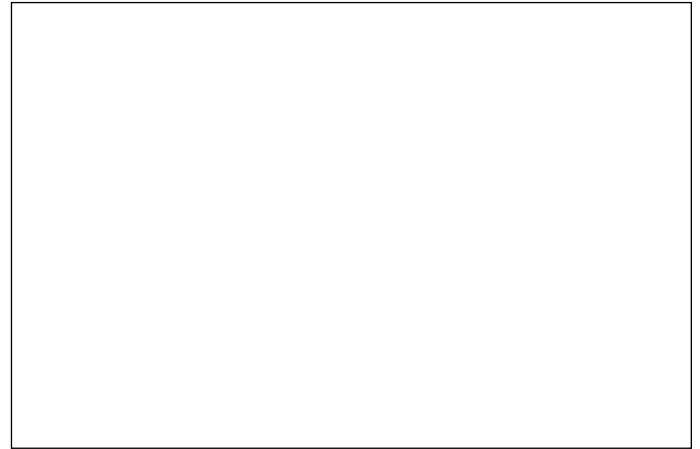
At the AGM changes were approved to change the way that Trustees are elected and so we thought it was a good opportunity to feature our team for the coming year.

It was agreed that from now on a Trustee will be elected for 3 years. Anyone can, of course step down at any time during this period, but it is hoped that Trustees will be able to stand for that time without having to seek re-election each year.

We all attend Board meetings 4 times a year and our AGM in October. We are divided into teams for the projects that we work on during the year namely: Support, Fundraising and Research.

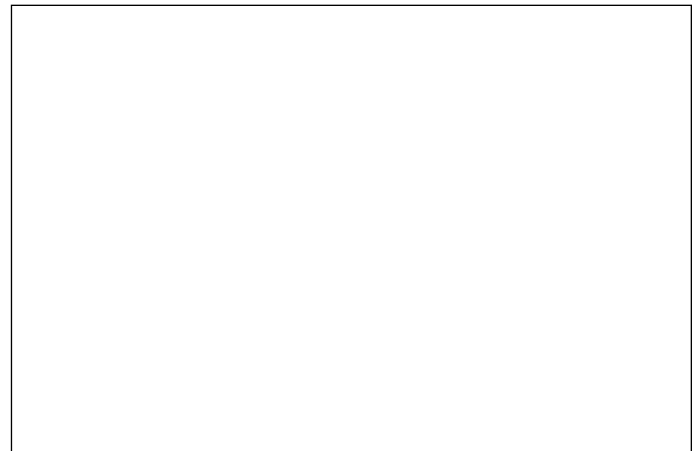
Trustees link closely with Sam and Georgette our paid support and as the charity has grown we feel it is the right time to recruit a charity manager. Their role will be to co-ordinate the running of the charity, including future strategy planning, networking with other relevant organisations, looking after internal Human Resource and Financial Management Issues. In the future the roles of a Financial Officer and Research Officer will also be defined.

## Support Team



The Support Team Trustees are **Barbra Higgins**, **Marion Bayliss** (pictured above with Sam our Family Support Officer) and **Pauline Docherty**. They share knowledge, experience and ideas in order that the BDFA can provide the best possible support to families and professionals alike.

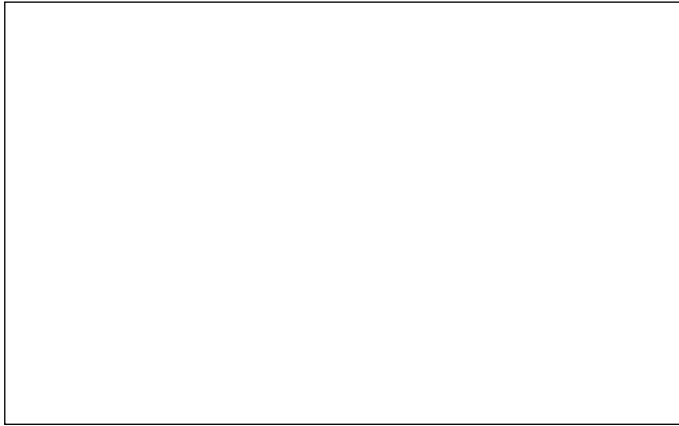
## Fundraising Team



The Fundraising Trustees are **Philip Glazebrook** (pictured above with Georgette our Fundraising Officer), **Kathryn Thompson**, **Pam Hughes** and a special welcome to **Melanie Hall** as a new Trustee to this group.

Mel is already a very active fundraiser for the BDFA and will be a great addition to the team. The Fundraising trustees bring to the BDFA their expertise and interests, which enables the very active fundraising programme to take place and develop.

## Research Team



*Heather Sickelmore, Heather Band & Julie Pickering*

The Research Team Trustees, pictured above, is co-ordinated by Heather B. They put their expertise to developing and administering our research grants programme, keeping up with the latest research, attending scientific meetings and any other science related projects.

We would like to say a big thank you to those Trustees who stood down at the last AGM.

**Debbie Jordan** was a very active and valued Trustee for 6 years, during which time she also served as secretary and is stepping down for family reasons but will continue to support us in the future.

**Liz Gatrell** was part of the Fundraising Team for 2 years. Using her professional fundraising background she played a key role in establishing the team and will continue to help with fundraising initiatives.

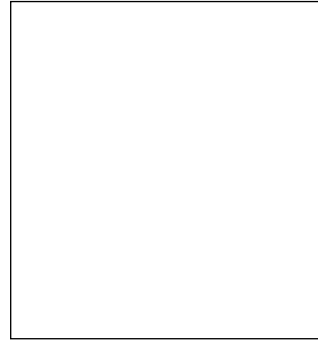
**NEW TRUSTEES ARE ALWAYS WELCOME** and if you or a family member or friend have time, skills and commitment to support our charity's work as a volunteer Trustee and would like to find out more please contact Julie Pickering via [info@bdfa-uk.org.uk](mailto:info@bdfa-uk.org.uk) or on 01603 760111.

### EVER THOUGHT OF BECOMING A VOLUNTEER?

The BDFA also has many volunteers who give their time and expertise to help in specific areas and if you would like to know more please contact Georgette on 01603 760111.

This year **Nicola Plumb** has designed a fantastic poster for the annual sponsored walk. Special thanks also goes to **Maggie Chisholm** for all her work with the website, **Anne Solbe** for all her help researching grant making organisations and **David de la Riviere** for his IT support.

## Professional Forum



*Dr Ruth Williams*

At the BDFA we have also been very fortunate to have the support of many professionals within the field of Batten Disease.

Dr Ruth Williams our Medical Advisor has been working very hard putting together the bid of National Commissioning Group (NCG) funding for a multidisciplinary Batten Clinical service. The proposed service will provide for all affected children and young adults, and their local professionals. It has been designed to give access to Batten experts, from diagnosis throughout the disease course to end of life, and will be delivered via co-ordinated diagnostic services, a Clinical Nurse Specialist providing advisory and outreach support as well as specialist multidisciplinary clinics at the Evelina Hospital.

In conjunction with this application the BDFA would like to know about the burden of health care for families looking after children with Batten Disease. Enclosed is a sheet asking you for information about your visits to hospital and visits to your home by professionals over a three month period. Please take a little time to see if you could help with this, the results of this will be used to inform the development of the Batten Service under the leadership of Dr Ruth Williams, and to monitor any impact such a service may have.

**Dr Ruth Williams**

Consultant Paediatric Neurologist at the  
Evelina Children's Hospital, GSTT, London  
[Ruth.williams@gstt.nhs.uk](mailto:Ruth.williams@gstt.nhs.uk)  
Evelina Children's Hospital, London

### Why should you become a member of the BDFA?

Your membership means that the BDFA can continue to grow as an organisation and reach all of those who need support from us. The wider our membership, the stronger and louder our voice can be. We welcome members whether they are family members, friends, friends of friends, professionals, scientists or someone who just has an interest.

**Your membership will cost just £10 per year.**

If you want to support us to continue our work then please fill out a membership form, available from Sam Simpson, email: [support@bdfa-uk.org.uk](mailto:support@bdfa-uk.org.uk), 01233 639526 and encourage your friends and family to do so too!

## Why God Made Me

Nearly everyone goes to school,  
Most people learn to read and write,  
They learn maths and history, English and art,  
But for some people there are other lessons to be learnt,  
Other things that need taught,  
And that, you see, is why God made me.

He looked down from Heaven on high,  
and all the wrong in the world made him cry,  
seeing a young couple lost in the world he turned  
to an angel and said,

"Let's give them a chance, send them their own special teacher".

So I was born to my mummy and daddy,  
their very own heaven sent teacher,  
Though only a few years have passed since that beautiful day,  
I think the Lord is pleased with the lessons I've taught.

Now that young couple stand out in the world, lost no more,  
For they have learnt lessons of a most special sort,  
It's been hard work for me and an uphill struggle for them,  
But now they know the true meaning of love, strength,  
patience and compassion.

They have an appreciation of all that is good and  
beautiful in the world.

Not bad for a few short years work, less time than it takes to  
learn Hamlet, or the history of two World Wars.

My job is almost done,  
and when they have learnt all that I have to teach,  
and said their tearful goodbyes,  
God will call me back to his side to wait for someone else,  
Someone who needs a special teacher, because you see  
That is why God made me.

by Alan Bates 23/12/05

We would like to thank Alan for sharing this poem that he has written about his daughter Rhiannon (9) who has Late Infantile Batten Disease.

## Remembrance

November and December has been a particularly sad time for the BDFA with the loss of Ben Pickering, Matthew Hall and Amy Jordan all within a few weeks of one another. During this difficult time our thoughts are with all of the families and friends of Ben, Matthew and Amy.



### IN LOVING MEMORY

Our thoughts and prayers are with the  
families and friends of:  
Benjamin James Pickering, aged 12 (Late Infantile)  
Matthew Hall, aged 7 (Late Infantile)  
Amy Jordan, aged 12 (Juvenile)

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](mailto:support@bdfa-uk.org.uk) or 01233 639526 for further information.

## New Family Networking Scheme

At the BDFA we understand the importance of giving families the opportunity to make contact with others who are living with Batten Disease so that “no family will go through the devastating journey of Batten Disease alone”.

The Family Networking Scheme now works slightly differently. Upon joining the BDFA or by making contact with the Family Support Officer, a family or individual can choose to be added to the Family Networking Directory. The directory will be circulated to all those who wish to be included. Upon receipt of the directory, families are then free to make contact with others on the list in their own time.

The information that we currently hold will need to be updated and therefore all those who are already part of the networking scheme will be contacted in the New Year.

As part of the new Support section of the BDFA website we are also looking to develop a means of family networking taking place directly from our website. Watch this space for further developments...

If you are interested in joining our Family Networking Scheme then please contact Sam Simpson, Family Support Officer (funded by the Roald Dahl Foundation) on: [support@bdfa-uk.org.uk](mailto:support@bdfa-uk.org.uk) or 01233 639526.

## Grants to Individuals and Families from the Roald Dahl Foundation



The Roald Dahl Foundation is very pleased to have been able to support the work of the Batten Disease Family Association by funding Sam Simpson in the new post of Family Support Officer. Since being set up by Roald Dahl's widow, the Foundation has made grants across the UK totalling more than £6m – funding charities, specialist NHS nurses and grants to individual children and families.

The Foundation's grants to individuals provide practical help to children and young adults up to 25 years (and their families) who meet their criteria for haematology and neurology, which includes Batten disease. The family must also meet their financial criteria and must be in receipt of either 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 a member of the social services, a specialist charity or a healthcare professional. The Foundations maximum grant is £500 but applicants need to apply for a specific purpose for which they will need to give a detailed costing. Further information and application forms may be downloaded from their website: [www.roalddahlfoundation.org](http://www.roalddahlfoundation.org) and follow the link to grants to individuals or contact Jo Smith, Grants to Individuals Manager on 01494 890465 or email: [josmith@roalddahlfoundation.org](mailto:josmith@roalddahlfoundation.org)

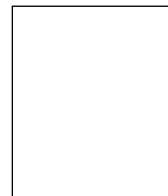
## Family Stories

The development of a new support section on our website is underway. As an organisation, the BDFA would like to ensure that the voices of families that we support are heard. We have developed a “Family Stories” section on our website which will give families an opportunity to share their story.

If you feel you would like your family story to be added to our website then please contact Sam Simpson, Family Support Officer on [support@bdfa-uk.org.uk](mailto:support@bdfa-uk.org.uk) or 01233 639526.

## Buggy looking to go to a new home

### A family wish to donate this Pushchair



It is a New Bug 4 wheelbase - size 4. Suitable for up to weight 45kg and child aged approx 8-14ish. It is crash tested and therefore suitable for sitting in and for transport in cars and buses. Splits into 2 parts for easy transportation and the back tilts, side and head supports so both can be changed to different positions.

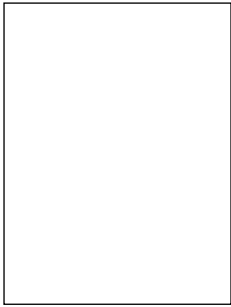
See [www.rainbowmobility.co.uk](http://www.rainbowmobility.co.uk) and click on Pushchairs for the Ormesa range for more information. Location for collection London or delivery could be arranged. Please contact Sam Simpson on 01233 639526 if you are interested.

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## Welcome to Research News



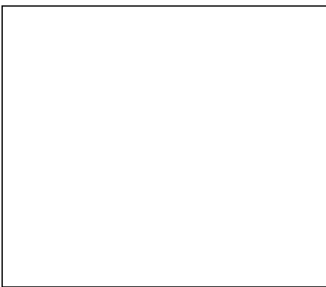
Heather Band

We are now into our second year of awarding grants for scientific research. Grants awarded under Tier 2 of our scheme (awards greater than £500 up to £5000) usually have a time period of one year and so those given out in 2007/8 have now been completed and the reports received. The program has proved very successful and is continuing to grow with over 30K awarded in the last two years. A further 15K, Tier 2 Grants

are to be awarded in 2010 and we also intend to give a higher award of 25K. So, for this issue we are featuring two reports, one from Sara Mole and another from Jon Cooper and Hannah Mitchison on what they have achieved and how having a BDFA research grant has enabled them to further their research into Batten disease. We look forward to giving more exciting news on this year's awards in our next newsletter but if you have any questions on our research grants please contact me via [research@bdfa-uk.co.uk](mailto:research@bdfa-uk.co.uk).

**Heather Band**  
Research Grant Co-ordinator

## Research Grant: Investigation of Golgi morphology



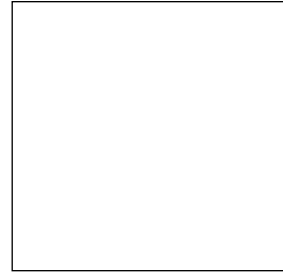
Jennifer Winter

Jennifer Winter, a PhD student at UCL, combined her interest in cell biology and use of model organisms to study disease by choosing to spend her third rotation project of 3-4 months working on Batten Disease in Dr Sara Mole's Lab which studies yeast as the simplest model

organism in which it is possible to understand JNCL.

Jennifer's aims were to (1) establish the best and a robust experimental protocol to view the Golgi apparatus in yeast using electron microscopy EM. (An electron microscope magnifies so

much that the insides of cells can be seen in great detail) and (2) carry out a study of different yeast strains and growing conditions to understand how the Golgi are affected by loss of the gene *btn1*, which is the equivalent gene to CLN3 which causes JNCL. She achieved aim (1) since we now have the details of each step of this protocol worked out.

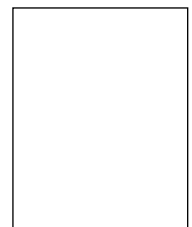


*(An example of one of her first EM pictures of a yeast cell with abnormal Golgi is shown – the Golgi are the dark string-like structures).*

However, when she set up all the experiments to achieve aim (2) and processed them for EM (which can take several weeks), in true experimental fashion, she had problems with one of the processing steps and she was not able to complete this part of the project. All is not lost as the samples are prepared and sitting in liquid nitrogen, and this work will be taken up either by another student who rotates in the Lab, or by Sara in parallel with other planned EM work. Jennifer has chosen to use a larger model organism, the nematode worm, for her PhD project in which she looks at brain function in the worm.

A previous PhD student in the Mole lab had used this same worm to investigate the function of the *CLN1/PPT1* gene that causes INCL. So, a grant from the BDFA allowed Jennifer to help further the use of EM in the Mole Lab and gain some invaluable research experience. Although Jennifer is now working on something different, she may return to Batten Disease in the future, bringing with her new approaches and knowledge.

**Sara Mole** PhD  
Reader in Molecular Cell Biology, MRC  
Laboratory for Molecular Cell Biology, UCL



Sara Mole

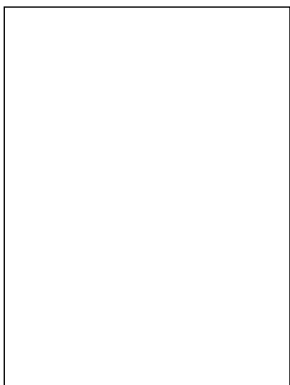
The Golgi apparatus are the parts of the cell that add the finishing touches to proteins before they are sorted and sent on their way to different locations within or outside of the cell. Dr Mole's Lab recently reported the novel finding that the yeast *Btn1* protein is located in the Golgi and that its complete loss affects these organelles very significantly. The same may be true for CLN3 and human cells.

## Research Grant: Investigating enhanced neurogenesis in Batten Disease

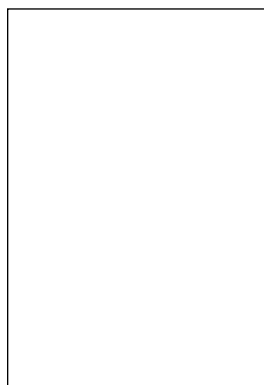
Jonathan Cooper (Institute of Psychiatry, King's College London) and Hannah Mitchison (University College London) were awarded joint BDFA funding in July 2008 to allow them to continue working together to investigate brain repair mechanisms in Batten Disease (BD). It is known that our brains can respond to some types of damage or disease by increasing the production of the stem cells or neural progenitor cells, from which all brain cells are made during development. New brain cells or neurons can be made from these neural progenitor cells, and if this internal brain repair system could be stimulated then it might provide new opportunities to replace the brain cells that are lost in BD.

Interest in this new area of research was first sparked by results from the Paediatric Storage Disorders Laboratory (PSDL) showing that sheep models of BD do show this 'neurogenesis' or continued birth of new brain cells. The aim of the BDFA-funded work was to take this work further by finding out whether this also happens in mouse models of infantile and juvenile BD (made by the Mitchison group at UCL), which we did in two different ways. The first way was by labelling newly generated cells within the brains of these mice and seeing how many new cells were being made at different stages of the disease, something that is important for understanding what triggers the production of new cells. Secondly we isolated and grew these progenitor cells in tissue culture and looked at what they were capable of becoming, which is crucial for finding out if these cells can be used to repair the brain. The work was performed primarily by two post-graduate students in the PSDL: Deesha Majithia who successfully completed her MSc project on this project last year, and Sybille Dihanich who is now in the last year of her PhD studies.

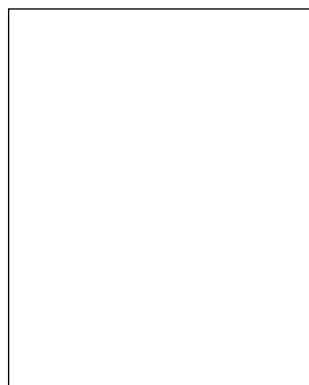
Using the cell labelling method we found that more cells are indeed being made in the brains of mice with both types of BD, but this happens much more in infantile BD. Most of these newly generated cells appear to be brain cells, and the number of new cells produced changes during disease progression with more being made at its later stages. We were able to grow these progenitor cells in tissue culture, but at the time Deesha's project was done we could only grow very few of them. Since then, we have learnt much better methods of doing this and can now grow large numbers of progenitor cells. The next challenge will be to see if we can fix their gene defect and find out if these cells can be used for therapy. Meanwhile, Sybille is investigating what triggers this neurogenesis within the brain, with one possibility being the abnormal glial responses we know occur in the BD brain.



*Jon Cooper*



*Deesha Majithia*



*Sybille Dihanich*



*Hannah Mitchison, UCL Institute  
of Child Health*

*Paediatric Storage Disorders Laboratory (PSDL)  
Institute of Psychiatry, King's College London*

### PSDL LAB DAY. 15th June 2010

Please come and visit Jon Cooper's lab to find out about the latest Batten Disease work on how different forms of Batten Disease affect the brain. □

Please contact Sam Simpson on [support@bdfa-uk.org.uk](mailto:support@bdfa-uk.org.uk) or on 01233 639526 if you would like to attend.

## Living Life with Batten's

Thursday 29th April 2010  
 Sherman Education Centre, Guys Hospital, London.  
 10.30am – 4.00pm



### A new event for families and professionals: Caring for a Child with Juvenile Batten Disease

<b>Morning:</b>	<b>Talks on key issues</b>
10:30 - 10:40	Coffee, Welcome and Introductions
10:40 - 11:00	Juvenile Batten Disease – The Basic Facts
11:00 - 11:30	Maintaining Good Physical Health and Mobility
11:30 - 12:00	Choices for Education
12:00 - 12:30	Epilepsy – Videos and Emergency Medication
12:30 - 1:45	Lunch and chat
<b>Afternoon:</b>	<b>Practical Workshops and Discussions</b>
1:45 - 2:30	<i>Anxiety, Hallucinations and Sleep Problems</i>
2:30 - 3:15	<i>Workshop: "What do we tell the children?"</i>
3:15 - 3:30	Tea break
3:30 - 4:00	<i>Juvenile Batten Disease – A Person Centred Approach</i>
4:00 -	Close of Meeting and Goodbyes

For further information and a booking form please contact Sam Simpson  
 on [support@bdfa-uk.org.uk](mailto:support@bdfa-uk.org.uk) or 01233 639526

Organised by the BDF A Batten Disease Professionals Group  
 Batten Disease Family Association  
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