

## The Australian Chapter of Batten Disease Support and Research Association Inc. - BDSRA

May 2015

Australia/ New Zealand

Issue 32

Dear families, friends and members,

What a milestone of a year this is. 20 years ago 3 parents aimed to become an integral part of a worldwide charity and initiated the Australian Chapter. The Committee took shape in the following years and one of those founders still remains as our President and has continued to support the Charity's endeavours—thank you Harry Partridge.. I was fortunate to be offered a role on the Committee in 2000 and how we have grown... Many have supported this Charity in the 15 years of service, volunteers and donors, and I am so proud to say that **together** we have given "Batten Disease affected persons a voice". Here are some Charity achievements.

Cheers from Vanessa, Manager.

### BDSRA Australia 1995 — 2015

Here are just a few special activities, to name a few ....

- ☺ 2000 merchandise items sold into many homes/businesses
- ☺ Donation Tins distributed around Oz: 320
- ☺ Registered families affected in Oz & NZ: 130 (BDSRA records)
- ☺ Volunteers: 74 (Committee, projects & fundraising)
- ☺ Books (Perils of Motorcycling) sold: 200 postal orders filled and over 1000 copies sold
- ☺ Family Meeting/Conferences: 8 (took place in QLD & NSW)
- ☺ Research Grants Funded: over \$500,000 from fundraising
- ☺ And, 1st Australian Research Scholarship to be awarded in 2016

⇒ **Help us to celebrate this milestone. Send us an email on what the BDSRA has meant to you. A commemoration will be placed at the website.**

⇒ **Attend the 2015 Annual General Meeting of the Australian Chapter on Saturday 26th September at Kirribilli, Sydney. Be a part of the Battens mission. Lunch included. Please put this in your diary to come. Dinner & accommodation is available for those staying over.**



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AWARENESS in OZ via the Moomba Festival in Melbourne and Awareness Day 31st March: Both these events created 2 big visit spike periods of visits at our website and Likes/shares on our Face-book page.

Www.battens.org.au - 889 new users, with 2,441 page views  
www.facebook.com/AustralianBDSRA - 567 likes with 4,598 users reached.

# Research & Services

## Updates — Programs locally

**A greater understanding of how altered cell calcium handling contributes to impaired brain cell function in Batten disease.**

**Alexandra Grubman and Anthony White, Department of Pathology, University of Melbourne**

Recent research from the US and Australia is shedding light on how brain cell function is impaired in some forms of Batten disease. Scientific work published by the teams of **Susan Cotman** at Harvard Medical School, USA, and **Anthony White** at University of Melbourne, Australia have shown that in CLN3 and CLN6 forms of Batten disease, the important element calcium is not handled correctly by brain cells. The mis-handling of calcium leads to impairment of key cellular processes designed to remove 'junk' proteins in the cell, resulting in accumulation of these proteins and other cell components, and neuronal cell damage. Susan Cotman's group has found that when the CLN3 protein function is impaired, this leads to abnormal movement and storage of calcium in neurons, resulting in defective autophagy, a process where the cell removes unwanted material. In closely related work, **Alexandra Grubman** and Anthony White have shown that neurons with impaired CLN6 protein function also have dramatic changes to handling of both calcium and zinc, and that control of the two elements is closely related. Treatment of neurons with a zinc delivery compound was able to substantially restore normal calcium handling in CLN6 cells suggesting that there may be potential in developing therapeutic approaches based on control of zinc and/or calcium handling in neurons.

For further information, you can email Anthony White at [arwhite@unimelb.edu.au](mailto:arwhite@unimelb.edu.au)

The National Referral Laboratory (Adelaide, South Australia) has recently expanded its genetic testing for Batten disease. In addition to testing already offered for the genes associated with infantile (CLN1), late-infantile (CLN2) and juvenile (CLN3) Batten disease, we are pleased to announce we have increased our scope and are now using Next Generation Sequencing technologies to also test for CLN4 (DNAJC5), CLN5, CLN6, CLN7 (MFSD8), CLN8, CLN10 (CTSD), CLN11 (GRN), CLN12 (ATP13A2) and CLN14 (KCTD7). Since introducing this new testing, we have diagnosed our first-ever CLN6 patient. We hope this extended testing can help others with suspected Batten disease to achieve a definitive diagnosis. More information at:

Genetics and Molecular Pathology, SA Pathology, Women's and Children's Hospital, [www.wch.sa.gov.au/nrl.html](http://www.wch.sa.gov.au/nrl.html)

## Updates — Research worldwide

### CLN2 Enzyme Replacement Trial -

### January 2015 update:

Other current locations for the trial are occurring at:

- ◆ We are very happy to announce that the **first United States site** for the BioMarin CLN2 enzyme replacement study has been listed on [clinicaltrials.gov](http://clinicaltrials.gov). Dr. Emily De los Reyes at Nationwide Children's Hospital in Columbus and her co-PI, Dr. Lenore Leowald are currently assessing children to begin the study in December. To learn more about the official guidance, and criteria, go to <http://1.usa.gov/1Hr3lbl>.
- University Hamburg-Eppendorf, Germany
- Bambino Gesù Children's Hospital, Italy
- Evelina London Children's Hospital, London
- ◆ The **estimated completion date of this study** is March 2016
- ◆ In January 2015 BioMarin Pharmaceutical Inc. **announced interim results** from its Phase 1/2 pivotal study for BMN 190— interim data indicates that in all nine of the BMN 190 patients who have been followed for at least six months and up to 15 months, the treatment appears to show stabilization of the disease compared to the natural history based on a standardized measure of motor and language function. To read more of this release: <http://investors.bmrn.com/releasedetail.cfm?ReleaseID=890846>

For more Research Information, please visit BDSRA-USA at this link: <http://bdsra.org/clinical-news/>

# Programs and Support

## Information Sharing

### National disabilityinsurance Scheme

The National Disability Insurance Scheme (NDIS) commenced in July 2013. For the first three years, it is being introduced at trial sites at selected locations because it's a big change, and they want to get it right and make it sustainable. Roll out of the full scheme will commence progressively from July 2016.

<http://www.ndis.gov.au/people-disability>

At this link you can find fact sheets and contact details who will help you with questions such as:

- ◇ What is the NDIS?
- ◇ Where the trial sites are
- ◇ What help can I get?

What type of services  
will be funded?

**BDSRA recently updated its “National” and “State-wide” helpful information and referrals for families. If you haven't received yours via email, please let me know.**



### Batting For Battens Inc., QLD

Persons affected by Batten Disease and their families are eligible to apply for a grant. You need to be a permanent Australian resident to be eligible to apply.

The need can be any purpose which will offer relief to the persons affected and their families. The scope is wide enough to allow the Foundation to support families with any need. Examples include renovations to the family home, relevant equipment, cost of attending a conference, etc., for information visit:

<http://www.bfbf.org.au/Grants.html>

The families of these very special people would like you to know of their passing and to

Remember them fondly:

**Marsden Hill of NZ**

**Shaniya Smith of WA,**

**Torben Dougan of NZ,**

**Sam Brinin of QLD.**

### EpilepsyAustralia

Epilepsy Australia is the national coalition of Australian Epilepsy Associations raising our voices as one to advance the cause of all Australians living with epilepsy.

Actively delivering counseling, support and information to all who access our services, Epilepsy Australia is committed to raising awareness and understanding of the very real issues faced by those living with epilepsy. A goal of Epilepsy Australia is for every household in Australia to be seizure aware.

Download a seizure guide today at:

<http://www.epilepsyaustralia.net/userData/docs/Ist%20Aid%20Poster%2009.pdf>

**Epilepsy Australia**  
**National Help Line**  
**1300 852 853**



Ronald  
McDonald  
Family  
Retreat®

Our Family Retreats are there to help relieve a little of this stress. They offer a welcoming and peaceful environment, often near the beach or close to local nature settings. Families with a seriously ill child, or those grieving the loss of a child, can come together in a calm and relaxing setting, far from the hospital environment.

Our Family Retreats are open to any family that has had a child diagnosed with a serious illness.

The entire family is welcome to stay for up to a week, although each retreat does have a maximum number of occupants.. The Family Retreats are located in: Bunbury (WA), Jurien Bay (WA), Forster (NSW), Bateman's Bay (NSW), Palm Cove (QLD) and Ocean Grove (VIC). For bookings, visit <https://www.rmhc.org.au/our-programs/family-retreats>

# Fundraising and Awareness

## NATIONAL BATTEN DISEASE AWARENESS DAY: 31ST MARCH And RECENT FUNDRAISERS

Many activities and media stories supported this annual Day. From being featured on the big screen at Moomba Festival, Melbourne and bouncing any which way in Bounce4Battens to supporters putting their best foot forward in National runs (including 8 year olds!), to highlights by partnering organisations and media. Batten Disease has been seen and spoken about in large numbers around Australia. Mission achieved. The fundraising that occurred is appreciated and will contribute to the future of a treatment or cure to this currently unstoppable disease...



**in NZ :** The Archer family held a gold coin day for Katie at Bayfield School and Early Intervention Centre, inspired by Rare Disease Day celebrations on 28/2, raising \$650



**in QLD :**

- \* Melinda and friends held a gold coin work collection and raised \$116
- \* The Bent Banana collected in Battens Tins



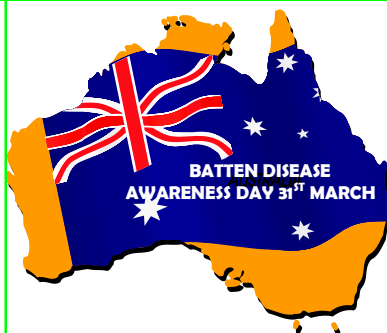
**in NSW :**

- \* Olivia and her school mates raised \$520 by the sale of merchandise



**in S.A.:**

- \* Well done to Wendy Smith & Lorraine with their Movie Night, raising \$350
- \* Ruth Carter & the Golden Grove Lutheran Primary School sold merchandise and fundraised \$326



- \* Bounce4Battens promotion by the Murchison family raised \$5946 and created a whole lot of awareness via the sharing of hundreds of photos.



**in W.A.:**

- \* Rogue Runners have gone above and beyond through their 2014/2015 support to Battens, reaching their goal by raising over \$21000. This has been a team effort with 29 members running in events around the country, a Memorial Run in Toowoomba and raffle. The Baton Relay was a huge effort, visit the 9 baton facts here:

<https://www.facebook.com/roguerunners.aust/videos/879878892069119/?fref=nf>



- \* Darryl & the Masonic Club, Belmont sold 900 tickets raising \$1,800.00 which will be matched by their Grand Lodge Charity "Masonicare". I.E. together raising \$3,600.00 (drawn by Naomi & Lilly)



- \* Craigie Baptist Church Craft Group continued their craft and presented proceeds of \$150

*job well done...* 

Joining in the HBF Fun Run and together raising \$8547, these supportive persons, including our youngest runner, Ryan (shown here just about to race) individually raised:

- |                     |                                     |
|---------------------|-------------------------------------|
| ◇ Merrick Law \$701 | ◇ Ross (Worley Wunners) \$264       |
| ◇ Helen Dowd \$326  | ◇ Toby Marsh (Worley Wunners) \$745 |
|                     | ◇ Sara & Ryan Halley \$7256         |





# Fundraising and Awareness continued...

The 'bounce4batten' awareness campaign was a great success. Media coverage was featured on National television, radio and newspaper with Channel 7 Weekend Sunrise, 2DayFM, The Sunday Telegraph and The Manly Daily all running stories. There were also numerous company websites, blogs, Facebook and Instagram profiles with strong media presence within social media providing an extensive reach and great impact for awareness.

A short video explaining the concept was created and shared among networks online. [https://m.youtube.com/watch?v=45t5P\\_Y8NtE](https://m.youtube.com/watch?v=45t5P_Y8NtE)



Within our network of friends and friends of friends postcards and fliers were distributed through cafes, letterbox drops, school and childcare centres and local sports associations. People were keen to show their support posting bouncing photos, liking and sharing articles and providing comments. The photos delighted and inspired and people engaged positively with the campaign from all over Australia and across the globe in the UK, Europe, America and Asia. The Gracetales website with over 26,000 engaged followers on Instagram featured a story and encouraged their followers to 'bounce4batten' as well as Facebook and website readers to their site. Redmagazine in the UK also shared an article "Why your kids need to bounce" with over 30k Instagram followers and an engaged audience on the magazines website.

Friends, family, friends of friends, strangers and celebrities bounced4batten! People and companies with strong media presence included magazine editors, James Tobin (weekend sunrise), Joanna Grigg, Nudie Juices, The Bachelor crew, MKRules contestants, Sissu, printebebe, notsomummsy and stylem. There were over 800 images people shared on Instagram alone with the hashtags #bounce4batten #bounce4battens #bounceforbatten, there was more photos, videos, sharing and likes on twitter & facebook too.

## Monthly On-Line Donations

Anthea Weeks  
Dean Apostolou  
Vidya Finlay

Shirley Tsui  
Jessica Page

Penny McNiff  
Nicola Edwards

## General donations

Algis Kullukas  
Meredith Fonti  
Joanne McLennan  
Donna-Marie Sheldon  
Cathy Norris  
Martin McNiff

Danielle Watson (in  
honor of Kianna's  
Quest)  
Ellen Jarrett  
Liza Anderson-Scott  
Alison Fowles

Robert Smith  
Lyndal Spence  
Mem Lane (in honor of  
Mia & family)  
Dr. Trevor Olsen  
Lucinda Millard

Denise Clarke  
Jan Van Helsen  
Lutz & Associates  
L M Abberfield  
Nicola Maddocks

## Donations made 'in memory'



### In Memory of Sheree & Sonya Kuhr:

Katie Eschler

### In Memory of Jai Longfield-Seychell:

The Bent Banana

### In Memory of Sam Brinin:

Collection from friends & family

### In Memory of Jordan Anderson:

Sandi Herbert  
Nancy Rhoades  
Joan Ciclovan (upon her passing)

### In Memory of Chloe Hampson:

Jessica Page (my mums little angel)  
Debra & Glen Hampson

### In Memory of Isla Donegan:

Tanya Elson  
Nanny and Poppy

### In Memory of Joey Weeks:

Anthea Weeks

### In Memory of Gus Mainwaring:

Francine McMullen

### In Memory of the children of Battens:

A gift (in lieu) to celebrate the wedding of Allan and Pam Innis, by Ian Tweedie

### In Memory of Matilda Berger

In lieu of leaving gift - Michelle Clift opted Battens  
Louise Golshevsky

### In Memory of Mackenzie-Rose

Jamie & Wendy Foster (a gift for her 12<sup>th</sup> birthday)