

Dear Families and Friends,

I am honoured to be the new president of BDSRA Australia. The other members of the 2019 management committee are Michael Fietz (vice president); Mandy Meredith (treasurer); Julia Mickler (secretary); Peta Murchison; Jay Riddle and Ineka van Zyl (general); and Vanessa Anderson (public officer), together with a number of extremely committed volunteers.

The management committee has recently set a number of objectives for BDSRA Australia in 2019 and beyond, including (1) to improve the support we provide to families; (2) to put Australia on the map for clinical trials and clinical research; and (2) to educate Australian health practitioners about Batten disease. The need to improve communication with all of our stakeholders was also identified as a priority.

As part of an effort to meet those objectives, Julie Ojeda has been appointed as executive assistant to BDSRA Australia. Julie will deal with all inquiries and ensure that messages for officers and volunteers are directed to the relevant person within 24 hours. Julie can be contacted at info@bdsraaustralia.org and 0475428691. We are also thrilled to announce that BDSRA Australia will be launching a new, user friendly, website – watch this space.

Finally, I would like to thank Hamish Murchison for his hard work and excellent management of BDSRA Australia over the last two years. Under Hamish's leadership, BDSRA Australia committed AUD \$150,000 to fund Batten disease research, which was an excellent effort.

Kate Beattie, President



New president, Kate Beattie, with her son, Tom (CLN2).

BDSRA Australia funds \$150,000 of research

Since July 2018, BDSRA Australia has given or committed to give over AUD \$150,000 to three Batten disease research projects. Dr Nadia Mitchell at the University of Otago, NZ, is leading one of those

projects. Dr Mitchell reports some exciting interim results with CLN5 gene therapy in affected sheep. Read more at: <https://tinyurl.com/yyahzj5r>

Rare Disease Day - 28 February 2019

Did you know there are over 6,000 rare diseases that affect over 300 million people worldwide? The Rare Disease community around the world will rally together to raise awareness on Thursday 28th February. 'Bridging health and social care' is the theme this year, calling for better coordination of all aspects of care for people living with a rare disease. Show you care by joining the social media campaign – just paint your face and share a photo using the hashtag #ShowYourRare. Find event details and more information at: <https://tinyurl.com/y5xua5vg>



Cerliponase alfa (Brineura) - Life Saving Drugs Program

In October 2018, Kate Beattie and Peta Murchison spoke to the Expert Panel of the Life Saving Drugs Program about why cerliponase alfa (Brineura), the only treatment for any form of Batten disease (CLN2), ought to be funded on the Life Saving Drugs Program. Following the advice and assistance of the Expert Panel, the Commonwealth Chief Medical Officer was to make a recommendation to the Minister by early January 2019. More information can be found on that process here:

<http://www.health.gov.au/internet/main/publishing.nsf/Content/lsdp-applications>

We eagerly await further news on this important decision.

Matilda receives her wish

Gorgeous Matilda recently received her wish through The Make A Wish Foundation - a new puppy called Symbol. Matilda's mother, Liz, says, "Now that Symbol is here, I couldn't think of



anything more perfect, a wish that can give every day for Matilda until she is gone and also joy for the whole family." Please read Liz's full story here: <https://tinyurl.com/y2lyn7hq>

You can contact The Make A Wish Foundation at: www.makeawish.org.au

News

BDSRA United States - family conference, 18-21 July, Colorado

The US chapter of the BDSRA will be holding their family conference in Denver, Colorado. Join the facebook group below for further information and updates if you are interested in attending:

www.facebook.com/BDSRAFamilyConference

CLN3 - recruitment for gene therapy trial

Amicus Therapeutics is recruiting children aged 3-10 years with CLN3 Batten disease for a Phase 1/2 Gene therapy trial in Ohio, Columbus at Nationwide Children's Hospital, USA. For more information, please see: <http://ir.amicusrx.com/news-releases/news-release-details/amicus-therapeutics-announces-phase-12-study-gene-therapy-cln3>

CLN2 – benefits of enzyme replacement therapy continue after 3 years

Biomarin recently announced that the patients treated with Brineura (cerliponase alfa) every fortnight in the ongoing open-label extension study across UK, USA, Germany and Italy continued to show a reduced rate of decline compared to a natural history cohort over a three year period.

For more information see: <https://seekingalpha.com/pr/17406141-biomarin-announces-ongoing-study-demonstrates-durable-treatment-benefit-brineura-cerliponase>

CLN2 - potential gene therapy

REGENXBIO has received rare paediatric disease designation for RGX-181. RGX-181 is a potential gene therapy treatment for children with CLN2 disease. For more information see: <https://regenxbio.gcs-web.com/news-releases/news-release-details/regenxbio-receives-rare-pediatric-disease-designation-rgx-181>

Useful links and resources

The following websites have some great resources for the Batten disease community.

Clinicaltrials.gov - information about all the clinical trials that are being conducted around the world.

Battendiseasenews.com - the latest research news on Batten disease.

Courageousparentsnetwork.org - an organisation created by parents for parents, providing information, tools, support and skills needed to help care for a seriously ill child.

Globalgenes.org - provides news, resources and updates relating to Batten disease.

Mia Murchison

In August last year, Mia Murchison became an angel. In Mia's short life, she achieved more for Batten disease awareness than anyone could have expected, including as the champion of the 'Bounce4Batten' campaign. The BDSRA would like to express our deepest sympathy for Mia's family and friends. Fly high Mia, fly high sweet girl.



Our new angels

Imogen Eve Stone 17 November 2006 - 8 October 2017

"We have been so very blessed to be Imogen's family, to share in this life with her. Such a pure soul. Imogen taught us the truest meaning of love and life. She is our Angel and Imogen will live on forever, treasured in our hearts." Lynda-Jane (Imogen's mum)

Katie Archer 22 June, 2008 - 25 March, 2018

"Full of enthusiasm, love, laughter, cheekiness, and determination. Katie taught so many about what is really important in life. She loved animals and Dora and was so deeply loved by her family and friends. Katie is the brightest shining star in the sky and in the hearts of all who miss her, especially Lisa, Brett, Amy and Caleb."

Michael Francis Murnane (Mick) 13 October, 2000 - 24 May, 2018

"Dearly loved son of Leo & Julie, loved brother of Paddy (dec), much loved grandson of Moira & Paddy (dec) and Heather & Allan (dec) Clarke, loved nephew of his aunts & uncles and a dear cousin. Aged 17 years. May he Rest In Peace."

Mia Johannah Murchison 27 July 2009 - 17 August 2018

"So incredibly sad and still so beautiful, we are overwhelmed and thankful for the out pouring of love and tears for Mia. And feel so privileged to have been her family. With all our love, gratitude & tears Hamish, Peta, Toby, Jasper and our forever shining star Mia."

Donations

Thank you to everybody who donated to BDSRA Australia in 2018 and 2019, with a special acknowledgment to our five largest donors: the Champion Foundation; the Mia Johannah Trust; Masonic Care Fundraiser; the new silks of the Victorian Bar; and the Nancy Rhoades Bequest. Without all of your support, we would not exist. **You can support BDSRA Australia by giving at** <https://www.givenow.com.au/battendiseasesupport>