



### Dear Families and Friends

It is almost impossible to believe that we are already in March of 2020. "2020" to me still sounds like some distant year in the future. A time with flying cars and robots! But it is here now and some of it is great and some of it isn't. The start of 2020 saw devastating bushfires here in Australia and it saw the world respond in a way that has been incredibly moving and inspiring.

2020 for Batten families sees amazing work being done by the team at BDSRA, new research being invested in, a new website and planning for a much anticipated Family Conference at the end of the year.

I have been lucky enough as the new Family Support Officer to be in contact with several families and would welcome anyone affected by Batten Disease to get in touch. I can be contacted via email at [support@bdsraaustralia.org](mailto:support@bdsraaustralia.org) or by phone/text on 0472 632 649. My hours are a little sporadic as I balance the role with my hospital work and family, so if I don't answer straight away, please leave me a message and I will respond as soon as I can.

This new role has allowed me a chance to reflect on my own experience with Batten disease and to think about the kinds of things that I would have found helpful when Tilda was alive, and the things that I would find helpful now that she has gone. I would love your input and suggestions on what would be useful for you and how we can better support all those affected by Batten disease.

Laura Smith  
Family Support Officer, BDSRA Australia

### 2020 Family Conference

Planning is well underway for our Family Conference later this year! Thank you so much to those who responded to our survey. We will be working with an experienced event planner to assist us with conference planning and logistics and welcome your input and suggestions. If you missed out on the survey or would like to have any input into conference planning, please send an email to [support@bdsraaustralia.org](mailto:support@bdsraaustralia.org). Watch this space and we will keep you posted!

### Childhood Dementia Partnership

BDSRA Australia is currently working closely with Dementia Support Australia (DSA), providing training and education that will enable specialist DSA consultants to better understand the impact of Batten disease and to provide timely, professional and pragmatic support to families affected by this form of childhood dementia and its related behaviours.

This exciting new partnership and training program will be showcased at the International Dementia Conference in Sydney in May, where BDSRA Australia Medical & Scientific Affairs Manager, Dr Ineka Whiteman, has been invited to present on the topic: *Childhood dementia: the confronting reality of rare paediatric neurological disease*.

To learn more about DSA or to speak to a DSA consultant visit [www.dementia.com.au](http://www.dementia.com.au) or call the 24 hour helpline on 1800 699 799.



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### COVID 19

With measures to protect the community against COVID 19 being ramped up and the situation changing rapidly, you are not alone if you are feeling particularly anxious about your vulnerable children. For the most accurate and up to date information from the government please visit: <https://tinyurl.com/rescbnb>

And please don't hesitate to contact our Family Support Officer either by [email](mailto:support@bdsraaustralia.org) or phone 0472 632 649 if you feel you need some support. We are thinking of all our families and sending our love.

BDSRA Australia

## Events

### *Annual General Meeting – BDSRA Australia*

It was wonderful to see so many families and friends at our AGM and lunch in Sydney in November. If you would like a copy of any of the reports presented at the AGM, please contact [info@bdsraustralia.org](mailto:info@bdsraustralia.org)



## Medical and scientific affairs update

### *BBDF-101 for treatment of CLN3 - Clinical Trial update*

In December last year, European biopharmaceutical company Theranexus and US-based Beyond Batten Disease Foundation (BBDF) announced its exclusive \$US 20 million agreement that will bring a new CLN3 treatment candidate to clinical trial in the USA this year.

In summary, the candidate treatment known as 'BBDF-101' is a proprietary combination therapy (intravenous and oral) intended to slow the progression of CLN3 disease. It works by activating a molecule known as transcription factor EB (TFEB), a 'master controller' of lysosome activity which, in theory, could help clear toxic cellular waste material and slow disease progression.

Enrolment in the paediatric phase of the trial is expected to commence in late-2020. This will be a two-year trial, based only in the USA, with enrolment of 30 paediatric CLN3 patients aged 4-16 years.

BDSRA Australia's Medical and Scientific Affairs Manager, Dr Ineka Whiteman, met with Theranexus Chief Medical Officer, Dr Werner Rein, and BBDF President and CEO, Mary Beth Kiser, to discuss the clinical trial and to seek out potential opportunity for the participation of Australian CLN3 patients. There is opportunity for enrolment of international patients, however to be eligible for selection, patients must be able to remain in the USA for the full duration of the trial and be available to attend weekly treatment at designated centres around the country, in addition to six-monthly clinical assessments. See the full post on our Facebook page [here](#).

For further information or to receive BBDF-101 trial updates and announcements, please contact Dr Ineka Whiteman at [research@bdsraustralia.org](mailto:research@bdsraustralia.org).

### *Research into the role of speech pathologists in early detection of CLN2 or Late Infantile Batten disease – CLN2 families needed!*

One of the earliest clinical symptoms of CLN2 or late-infantile Batten disease is delay in language development. Speech pathologists are often one of the first healthcare providers referred to in the early (pre-diagnosis) stages of CLN2 disease and therefore may potentially play a critical role in its early detection.

Current and bereaved CLN2 families are invited and encouraged to participate in a short, confidential questionnaire. To participate in this important study, the online questionnaire can be found [here](#) and the full story via our Facebook page [here](#).

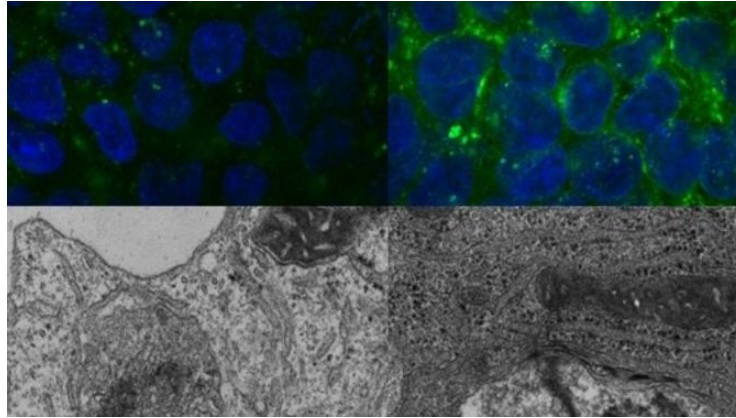
If you have any questions or require further information, please contact Lisa Tilbrook, Senior Clinical Speech Pathologist at [lisa@thrivehealthgroup.com.au](mailto:lisa@thrivehealthgroup.com.au) or Dr Ineka Whiteman, BDSRA Australia Medical and Scientific Affairs Manager at [research@bdsraustralia.org](mailto:research@bdsraustralia.org)

## News

- Federal Government Announces National Strategic Action Plan for Rare Diseases. The Morrison Government has announced it will provide up to \$3.3 million for activities to implement the first National Strategic Action Plan for Rare Diseases. The Action Plan aims to increase awareness and promote earlier, more accurate, diagnosis and treatment for around two million Australians suffering from rare diseases. Further information can be found [here](#) and [here](#).
- New research has deepened the understanding of the underlying causes of Batten disease, including organs and cell types affected as well as crucial molecular mechanisms, which can help the design of novel therapies, a review study reports. Read our Facebook post [here](#), or via the Batten Disease News site [here](#).
- Polaryx Therapeutics has announced that it has received Investigational New Drug (IND) approval

from the FDA for the drug PLX-200, allowing them to carry out a clinical trial for CLN2 with PLX-200. See our Facebook post [here](#) or the full article [here](#).

- BDSRA Australia welcomes the recent announcement by Federal Health Minister, The Hon Greg Hunt MP, that through the Medical Research Future Fund (MRFF), the Morrison Government is investing \$55 million to research rare diseases and rare cancers. See our Facebook post [here](#), or read the media release [here](#) and go to [www.grants.gov.au](http://www.grants.gov.au) or more information on the grant round, including application dates and criteria.
- Sueanne Chear, a PhD candidate in Assoc Prof Tony Cook's team at the Wicking Dementia Centre at University of Tasmania, presented their latest research at the Australian Society for Stem Cell Research conference in Brisbane in at the end of last year. Co-funded by the Australian and US Chapters of the BDSRA, this research makes some important advances in the characterisation and understanding of stem cell models of CLN3 disease. Through various microscopy and labelling techniques, Cook's team demonstrate that CLN3 stem cell models recapitulate several hallmark pathologies of CLN3 in humans. In future, these cell models may be a useful tool for testing potential new drugs for treatment of CLN3. Read our Facebook post [here](#).



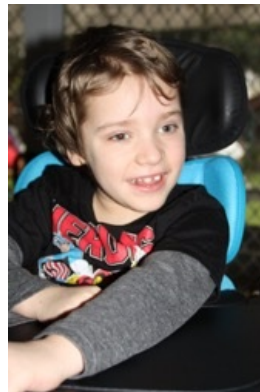
- An update from Regenxbio on potential inhuman gene therapy trials for CLN2: RGX-181 for the Treatment of Late-infantile Neuronal Ceroid Lipofuscinosis Type 2 (CLN2) "Batten" Disease REGENXBIO intends to submit an Investigational New Drug (IND) application (or foreign equivalent) for the first in-human clinical trial evaluating RGX-181 in the second half of 2020, following additional preclinical development and analyses to support clinical development. Preclinical data presented at the European Society of Gene & Cell Therapy (ESGCT) 27<sup>th</sup> Annual Congress highlighted outcomes from a murine mouse model for CLN2 Batten disease

#### Our new angel

*Nicholas Elliot*

Nick gained his wings on Sunday afternoon, 15<sup>th</sup> December, 2019. Fly high, be free we love you.

All of us at the BDSRA send all our love to the Elliot family.



## Fundraising

### *Art Union Raffle*

*Art Union Raffle is now closed, with winners being drawn on 12 March 2020 – Good luck to everyone who bought tickets!*

### *Our Everyday Heroes*

Have you considered raising money for BDSRA Australia? It is easy with "Everyday Hero", which can turn any event (large or small) into a fundraiser, with direct tax deductible donations to BDSRA Australia. It's so easy to create your own fundraiser from the Everyday Hero website and nominate BDSRA as your charity of choice.

### ***Our Everyday Heroes - "nobody can do everything, but everyone can do something"***

BDSRA Australia relies on donations and fundraising in order to support families and research.

Have you considered raising money for BDSRA Australia? It is easy with "Everyday Hero", which can turn any event, large or small, into a fundraiser, with direct, tax deductible, donations to BDSRA Australia. To learn more or create your own Fundraising Page with our charity as your choice, follow this link:

<https://give.everydayhero.com/au/get-started>



### **A special welcome**

The BDSRA would love to welcome the utterly delectable Faith-Kenzie Yvonne Harriet Foster born on the 30th December 2019. We love the Faith spam, Naomi - keep it coming!

### **Donations**

Thank you to everybody who donated to BDSRA Australia recently. Without your support, we would not exist. You can support BDSRA Australia by giving at <https://www.givenow.com.au/battendiseasesupport>

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