



Impact Report 2021 - key highlights



This is my son Tom. He is one of the children BDSRA Australia supports. This year BDSRA Australia awarded over \$100,000 for research in Australia and New Zealand. During our 'meet the expert' evenings, our families heard from international Batten disease experts, Professor Sara Mole (UK), specialist music therapist in Batten disease, Rebecca Atkinson (UK) and our own head of Research, Scientific and Medical Affairs, Dr Ineka Whiteman, with updates on research and clinical trials. Our family support officer, Liz, sent care packages, held 'virtual' coffee-catch ups and provided much needed emotional support in difficult times. BDSRA Australia made formal submissions to the Expert Panel of the Life Saving Drugs Program on the 24 month review of cerliponase alfa (Brineura) and we continued to work with and train Dementia Support Australia to support our families. And we continued to advocate to bring clinical trials to Australia - and to create opportunities for Australians to join trials overseas - through extensive, ongoing, communications.

Thank you to our volunteer management committee, our volunteer Medical & Scientific Advisory Board, our consultants and our supporters who make all of this happen. Please support our work this Christmas: <https://bdsraaustralia.org/index-donate.php>

We hope you have a lovely summer and a much deserved holiday.

Kate Beattie
December 2021

Congratulations to our 2021 BDSRA Australia Research grant winners!

BDSRA Australia is pleased to announce the two winners of this year's Research Grants, with over \$100,000 awarded.

Congratulations to chief investigator Associate Professor Anthony White (pictured left) from QIMR Berghofer Medical Research QLD who has been awarded \$49,600 for the project 'Integrating computational and in vitro approaches to achieve drug re-positioning for Batten disease'.

Congratulations also to chief investigator Associate Professor Anthony Cook (right) from the Wicking Dementia Research and Education Centre at the University of Tasmania, who is a second-time recipient of a BDSRA Australia research grant. This year, A/Prof Cook has been awarded \$55,000 for the project 'Dissecting the effects of CLN3 variants on the blood-brain barrier'.

BDSRA Australia is very proud to support such world-class, promising research right here in our region. Further details on the successful projects and research teams head to <https://bdsraaustralia.org/research>.



BDSRA Australia AGM

The BDSRA Australia Annual General Meeting was held on Saturday, 27th November 2021. We would like to welcome our new Board members, Bobbie Riddle and Amanda Gilpin and thank our continuing Board members: Kate Beattie (president), Dr Michael Fietz (vice president), Mandy Meredith (treasurer), Julia Mickler (secretary), Dr Ineka Whiteman (research, medical & scientific affairs), Amber Rundle (general) and Vanessa Anderson (public officer). You can find more information about all of our Board members here: <https://bdsraaustralia.org/about>. We are also grateful to Meg Garrido and Naomi Foster who continue as volunteers for BDSRA Australia.

We would like to thank outgoing Board members Laura Smith and Jay Riddle. We would also like to thank Ms Julie Ojeda for her hard work as our executive assistant to the Board for the last two years and we

wish her well in her future endeavours.

We are looking for new members of our community to join our Board in November 2022. Please contact us at info@bdsraustralia.org if you are keen to get involved.



Welcome to our new EA & communications manager

We welcome our new executive assistant and communications manager, Nikki Hopkins. She's a Sydney based mum with 3 kids. Her oldest son has Autism which has sparked a keen interest in transferring her skills across to the NFP sector. Nikki has recently worked at The Sanfilippo Children's Foundation as well as helping to launch the A-List. Please reach out and say hi to Nikki at info@bdsraustralia.org.

Dementia Support Australia - support service available to our families

BDSRA Australia and Dementia Support Australia (DSA) work together to support families affected by Batten disease. DSA has trained consultants who understand Batten disease, childhood dementia and its related symptoms, and the impact of dementia on children and youth, their families and carers. Through individualised assessments and behavioural management plans, DSA provides professional, practical strategies and recommendations to support clients and their families, and to improve their quality of life.

The service is free, with no referral required. Call the 24/7 service on 1800 699 799 to talk to a trained DSA consultant, or visit DSA Childhood Dementia Support Service.

As one parent writes: *"The support provided by Dementia Support Australia was the most practical, useful and effective service that I've accessed since [my children's] diagnoses."*



Medical & Scientific Advisory Board

The BDSRA Australia Medical & Scientific Advisory Boards is comprised of leading medical and allied health experts with experience in diagnosis, treatment and management of Batten disease. The Advisory Board was established in 2019 as a resource for other healthcare practitioners seeking advice on Batten disease management. It also supports BDSRA Australia's

Prof Monique Ryan

We would also like to take this opportunity to sincerely thank outgoing Advisory Board member Prof. Monique Ryan from the Royal Children's Hospital Melbourne for her service and support of BDSRA Australia over the past two years. Prof. Ryan has been an active contributor to the many

research, education and awareness-raising efforts. We would like to express our deep gratitude to our Advisory Board for all of the work that they do and all of the time that they volunteer to support our children. You can read more about them here: <https://bdsraaustralia.org/about>

Prof Michael Fahey

We would like to welcome Prof. Michael Fahey, who is joining our Advisory Board. Prof. Fahey is Head of Paediatric Neurology at Monash Children's, Director of Neurogenetics at Monash Health and Professor of Paediatrics at Monash University. Prof. Fahey has a keen interest in Batten disease and other neurogenetic disorders, with an extensive research track record in the areas of cerebral palsy, neonatal brain injury and Friedreich's Ataxia. We are delighted to have you on the team, Prof. Fahey!



activities we have undertaken as an organisation, and a great advocate for all our Batten families. Our very best wishes go with you Monique as you embark on your exciting new endeavours, which you can read about here: <https://www.moniqueryan.com.au>



Life Saving Drugs Program – 2 year review of Brineura

In October, the Life Saving Drugs Program (LSDP) undertook a planned 24-month review of cerliponase alfa (Brineura). BDSRA Australia was invited to provide a submission to the LSDP Expert Panel regarding patient experience on Brineura. We would like to thank the CLN2 families who provided their valuable input.

The Director of the LSDP Review wrote to thank BDSRA Australia and contributing CLN2 families for our prompt and thorough submissions. Feedback from the Panel was that our contribution was of great assistance and provided a helpful basis to address many of the patient experience questions. While the Review outcomes are still to be finalised, the Director has reassured BDSRA Australia that there is no intention to remove cerliponase alfa from the LSDP.

A fabulous outcome, and we will be sure to keep our community updated when the final outcome is announced.

Meet our new-ish Family Support Coordinator

Liz Sanson started in April this year as Family Support Coordinator for BDSRA Australia. Liz has been part of the Batten disease community for 5 years. Her Batten angel, Matilda, will be forever 9. Liz says:

"2021 has been an incredibly hard year for everyone with Covid19 outbreaks, restrictions & lockdowns. It certainly adds hardship to those who are already facing the everyday challenges that Batten disease brings. As the year comes to an end and we look forward to the year ahead, it can look daunting with the uncertainties, but there is hope with each new day. In this crazy holiday season, I encourage you to take opportunities to slow down and just be, to make the most of the little moments. Best wishes for a Merry Christmas & Happy New Year, Liz"



Please don't hesitate to contact Liz at support@bdsraustralia.org if you need support. She is a wonderful listener - and has a wealth of experience about Batten disease. Also, join us for a **holiday 'coffee catchup' for families** on 18 January 2022. Zoom details are available from Liz.

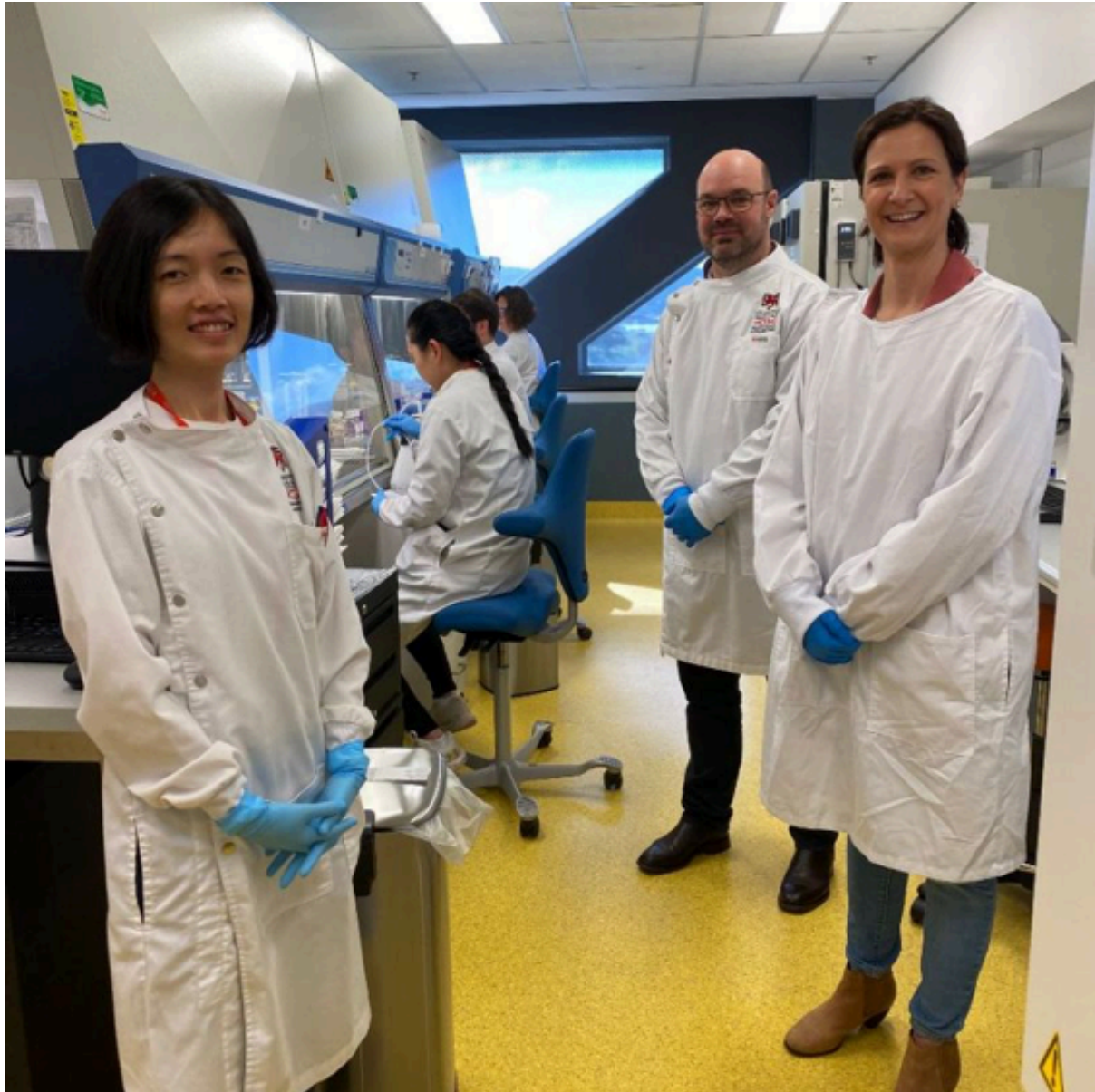
Meet the Gilpin family



In April 2021, Amanda and Craig Gilpin's world was turned upside down when their 6 year old daughter, Hollie, was diagnosed with CLN5. Amanda writes: *"Until that point, we had been living the Pilbara dream in remote WA with Hollie and her big sister Maddison. It is very overwhelming riding the waves of rare disease and the isolation we felt was suffocating. The most important conversation I had at that time was speaking with Liz at BDSRA and the support they have given us since is phenomenal. I was so scared to take that call because it made everything real but it empowered us, and we were welcomed into a new family of beautiful people who truly "got it", which is something we are very thankful for. Living thousands of km's away from any family support, we were so used to doing things independently, so it's been hard to adjust well ingrained habits and start asking for and accepting help offered. One app that was suggested and has been very helpful is 'Gather My Crew'. It allows us to post up things we need help with and*

people can offer the help if they are in the position to do so. We also began the "Hope for Hollie" private Facebook group to share our journey. Luckily we have also been blessed with a very supportive and incredibly proactive therapy team who go above and beyond. Without the incredible support of our friends, family and broader community who have completely embraced our family's situation we would not be able to tackle this journey in the way that we are, for which we will be forever thankful."

Research, Medical and Scientific Affairs update



Batten Disease Research Facility Visit - University of Tasmania

Head of Research, Medical and Scientific Affairs, Dr Ineka Whiteman (right), travelled to Hobart in September to meet with Research Grant recipients Associate Prof. Tony Cook (middle), Sueanne Chear (left) and their research team at the Wicking Dementia Centre, University of Tasmania.

Dr Whiteman was given a "VIP" tour of the Wicking Centre laboratories and observed some of the exciting work being conducted in stem cell models of CLN3 disease. "How impressive it is to see firsthand the world-class facilities and cutting-edge experimental techniques being used by Prof. Cook's research team," Dr Whiteman said. "They have been able to measure some of the cellular changes that occur as a result of different CLN3 gene mutations, using neurons that have been derived from human stem cells. Soon the team will be testing a whole range of drugs and compounds to see if the CLN3-associated damage can be rescued in these neurons." It is hoped that this research may lead to identification of new

therapeutic drugs for treatment of CLN3 disease. To read more about this project, head to: <https://bdsraaustralia.org/previous-grants>.

Clinical Research Update for Families - webinar series

We recently held research update webinars summarising the current clinical trial landscape for CLN3 disease and CLN2 disease. The sessions included a general overview on drug development, the clinical trial process, patient registries and natural history studies, and discussed a number of investigational therapies currently in development for CLN3 and CLN2 disease. To request a copy of the recorded presentations, please email Ineka at research@bdsraaustralia.org.

Batten Disease and Childhood Dementia to feature in University's Bachelor degree program

During her recent visit to the Wicking Dementia Centre, University of Tasmania in Hobart, Ineka took part in an extended interview to discuss Batten disease and childhood dementia, their prevalence, causes, treatments and family support mechanisms. This content will form part of the syllabus for the University's Bachelor and Diploma of Dementia Care from 2022. We are so encouraged to see the growing awareness and education around Batten disease and childhood dementia!

If you have any questions about Batten disease research, including clinical trials, please contact Ineka at research@bdsraaustralia.org. Further research updates and medical & scientific information sessions will be held throughout 2022. To register your interest, please email info@bdsraaustralia.org.

Our new angels

(WARNING: Aboriginal and Torres Strait Islander readers are advised that the following story contains an image of someone who has passed.)

Matilda (top)

Matilda Analee Sanson was diagnosed with CLN2 in June 2015 and passed away on the evening of Saturday 30th January 2021, aged 9. Matilda was a very determined and loving girly girl. She enjoyed so much about life, from animals and friends to being out & about at school and events, despite her ever-changing health and physical regression. We will miss Matilda and her beautiful smile.

Damilia (left)

Beautiful Damilia took flight with the desert breeze, moving and returning to Country with her ancestors. Our hearts are full of her love, song and dreams beyond this world. Fly high, sweet angel. Damilia passed 31st October, 2021, age 15. Our love and thoughts are with Angela and Damilia's incredible circle of family and friends.

Lachie (right)

Lachlan Fitchett was nearly 23 when he passed away on Thursday 5th August 2021. Lachie, also known as "Shaggy Rogers", was remembered for his determination, his passion for life and his smile. He was a huge fan of Scooby Doo and Guns & Roses. We all send our love to Sharon and the whole family.



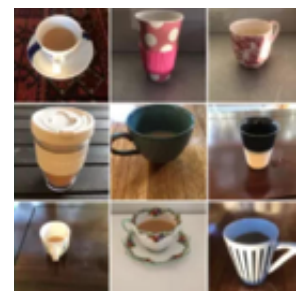


Special (virtual) coffee catchup for bereaved families

In November, we held our first virtual catch up for bereaved parents in Australia & New Zealand. It was facilitated by our guest Meg Chin, a Family Support Practitioner at Very Special Kids, a children's hospice and family support organisation in Melbourne.

Bereaved Batten parents had the platform to meet and share with others who understand the unique challenges & stages of caring for a child affected by Batten Disease and experience of grief & loss when their child passes. The feedback from those who were able to attend, was that they were so thankful for the group, it was a comforting experience, they felt understood and it really helped how they were feeling.

If you have lost a child (of any age) to Batten disease and would like to be involved in future opportunities to connect with other bereaved parents, including but not limited to future zoom catch ups, please let Liz know at support@bdsraustralia.org



Fundraising and donations



This year, there have been a number of very special fundraisers for BDSRA Australia to honour friends and family members affected by Batten disease.

We would like to say a special thank you to:

- **Wilson Hickey** (pictured above), who raised over \$7000 through sponsorship for walking, riding and swimming over 100km on Lord Howe Island during the school holidays in honour of his cousin Tayla Riddle.
- **Violet Garrido**, who raised \$2111 and **Kai Riddle**, who raised \$1100, by selling lolly bags and merchandise at their schools on Bounce4Batten Day in honour of Mia Murchison.
- The kids at **Complete Kids Early Learning Centre**, which held various fundraisers in honour of their friend Harper, raising over \$2500.
- The **Mickler family**, who continued their ongoing fundraising through recycling Bottles for Battens; and
- **Naomi Foster** and **Bobbie Riddle**, who raised over \$1600 by selling Burn for Batten candles. These beautiful candles have sold out again this year but will be available to buy at next year's retreat.

These fundraisers help BDSRA Australia to continue its important work. If you'd like to hold a fundraiser please reach out to Meg at fundraising@bdsraaustralia.org for ideas or help.

We would also like to thank everybody who donated to BDSRA Australia in 2021. Without your support, we would not exist. You can support BDSRA Australia by giving at <https://bdsraaustralia.org/index-donate.php>

ASX Refinitiv Charity Foundation Raffle 2021

We are extremely fortunate to again be a beneficiary of the ASX Refinitiv Charity Foundation. Please don't forget to go online and buy your raffle tickets - with all proceeds for tickets sold via this link going directly to BDSRA Australia [https://www.asxrcfau.com/console/rafflepages/2/The-Batten-Disease-Support-and-Research-Association?](https://www.asxrcfau.com/console/rafflepages/2/The-Batten-Disease-Support-and-Research-Association?fbclid=IwAR3IQEgxRdiLRCILASipYIP4r8_ZcNZrXYsvyduHGwDS6YWYily2txxJvNw)

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Last year, BDSRA Australia ticket holders won first and second prize in this raffle – you've got to be in it to win it!

Merry Christmas from BDSRA Australia

Rose, sister to Batten angel, Matilda



Personal Details Update: For all our families, so we may keep you informed and up to date, if your circumstances or contact details have changed, please inform Nikki Hopkins at info@bdsraustralia.org.au



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