

NEWSLETTER

Winter 2024 Edition



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President's Report



Photo: Group photo of Beach2Beach participants in 'Alara's Army' team

Last weekend the annual Beach to Beach fun run was held between Dee Why beach and Newport beach on Sydney's stunning northern beaches. We had an overwhelming response to this year's event and would like to say a huge thank you to Alara's army and all who participated and raised an incredible \$8700. Alex Mickler also made us all proud raising funds and running for his sister in the recent City to Surf.

While we are recognising such dedicated supporters I'd also like to back track to June and highlight another successful International Batten Disease Awareness Day and sincerely thank all those who got on board with the Bounce for Batten campaign posting photos on social media across the country to build awareness. A special mention must be made to our families and communities who organised dedicated fundraising events which you can read more about in this newsletter.

To find out about further events, family and educational support or fundraising please see our Events, Fundraising and Family Support sections below for plenty of upcoming activities in the next couple of months including securing your ASX Refinitiv Charity Foundation [raffle tickets](#)!!

In research news, as part of our strategy to collaborate on a more global scale we have been working for some time to expand our Australian and New Zealand Grants Program. In July at the BDSRA Foundation Family Conference in the USA, BDSRA Australia in partnership with the BDSRA Foundation USA, BDSRA Canada and the BDFA UK, launched the inaugural [Batten Disease Global Research Initiative](#) of which I am pleased to report is well underway and now accepting grant applications until THIS FRIDAY 30th. Further information about the grants initiative can be found in Ineka's report, but I'd like to say a huge thank you and acknowledge the team who have worked tirelessly to bring this initiative to fruition lead by Ineka Whiteman together with Amy Fenton Parker, Lori Brown, Bev Maxim, Liz Brownnutt, Joanna Nightingale, Jon Cooper and their supporting teams.

We look forward to expanding our reach and unearthing some promising new research leads to accelerate therapeutic development, improve diagnosis and standards of care, and enhance the quality of life and outcomes for all individuals affected by Batten disease.

Finally, it's so pleasing to be able to share some really **positive news on the clinical trial** front. In addition to Neurogene's CLN5 disease gene therapy program hitting a new milestone, we are delighted to share straight off the back of last night's launch announcement that REGENXBIO's investigational gene therapy programs for CLN2 disease have been acquired by the experienced and passionate team at Tern Therapeutics. You can read more about this exciting development below in our Clinical Program Updates.

Warm regards,
Amber

Annual General Meeting

This year's AGM will be held on **Sunday 10th November** online via zoom at **11 am AEDT**. Further information for you to register should you wish to attend will be provided closer to the date.

CALL FOR BOARD NOMINATIONS

The BDSRA Australia Board of Directors is now accepting applications for new board members. We have two available board positions.

Applicants who are encouraged to apply will preferably be associated or familiar with Batten disease, rare disease or genetic diseases, have some experience or knowledge of working in the NFP sector, will be required to be involved in fundraising initiatives and advocacy and/or will be able to contribute other skills and expertise in areas such as governance, strategy, grant applications, fundraising, marketing, finance or law.

To apply or express your interest in joining the board please email our secretary Julia Mickler at secretary@bdsraaustralia.org and we will respond to discuss the process and requirements with you.

All applications will be considered and applicants may have a phone or zoom interview. Elections will be held at the AGM on Sunday 10th November and applicants up for election will be expected to attend the online AGM.

For further information or any questions you have about joining the board and the positions available please email our secretary Julia Mickler at secretary@bdsraaustralia.org

Research Highlights



BATTEN DISEASE

Global Research Initiative

Announcing the Batten Disease Global Research Initiative launch and Expressions of Interest open until 30th August

On July 13th, BDSRA Australia was thrilled to be part of the unveiling of the brand new [Batten Disease Global Research Initiative](#) (BDGRI), a consortium of leading patient advocacy organizations worldwide that share the common goal of advancing research for all forms of Batten disease. Together, with our founding partners BDSRA Foundation, BDSRA Canada and BDFA UK, our mission is to foster and collectively invest in the most promising research worldwide that seeks to address key research questions and areas of unmet need in Batten disease.

[Expressions of Interest are now open until Friday 30th August.](#)

Grants of up to USD \$50,000 will be awarded for one year, with the potential of renewal based on outcomes in the first year. Funding will be provided by the Global Research Initiative, with financial contributions from BDSRA Foundation, BDSRA Canada, and BDSRA Australia, and in-kind support from BDFA.

In addition to this core funding, support is also being sought from other interested parties who may wish to partner on selected, subtype-specific projects, as our 'GRI Funding Partners'. We are grateful to have already received interest from several potential Funding Partners including NCL Stiftung (Germany), Beyond Batten Disease Foundation, ForeBatten Foundation and Noah's Hope-Hope4Bridget Foundation.

To find out more about the Program and how to apply, please visit our site at battenresearch.org

Best wishes to all our Australian and New Zealand applicants!

Dr Ineka Whiteman

Head of Research and Medical Affairs

For further **Research Updates**, please see sections below!

Upcoming



Virtual Coffee catchup - Life with Late-Infantile Batten disease

A parent discussion on day-to-day life with an affected child/ren with Late-infantile Batten disease, CLN2, 5 - 7 etc. An opportunity for peer support, share the struggles, the joys and the practical how to manage paperwork, siblings, self-care and everything in between.

Tuesday, 3rd September - 1:00 PM - 2:00 PM (AEST) [Register here.](#)



Virtual Coffee catchup - Life with Juvenile Batten disease

A parent discussion on day-to-day life with an affected child/ren with Juvenile Batten disease, CLN3, late onset CLN2 etc. An opportunity for peer support, share the struggles, the joys and the practical how to manage paperwork, siblings, self-care and everything in between.

Thursday, 5th September - 1:00 PM - 2:00 PM (AEST) [Register here.](#)



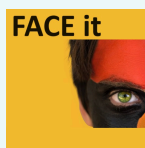
State of Childhood Dementia 2024

Parliamentary Friends of Dementia and Childhood Dementia Initiative invite support to launch the State of Childhood Dementia 2024 report.

Location: Enid Lyons Alcove, Parliament House, Canberra

Monday, 9th September - 4:30 PM to 6:00 PM (AEST)

If you are interested in attending this event, contact [BDSRA Australia](#)



Childhood Dementia Day

The FACE it campaign encourages people to make their face colourful and then to share a photo of it on Childhood Dementia Day, bringing a spotlight and awareness to childhood dementias like Batten Disease.

Wednesday, 18th September - on social media

#childhooddementia #FACEit Find out more [here.](#)



National Carers Week Morning Tea (Carers NSW)

Recognised yearly, National Carers Week 2024 is held during the dates **Sunday 13 to Saturday 19 October**. To celebrate, BDSRA Australia is organising a much deserved gathering for our caregivers in greater Sydney. Last year, 8 mums came together for morning tea at Eden Gardens for informal peer support, to chat, laugh and enjoy some delicious food! Keep an eye out - more information coming soon!



Lysosomal Disease Summit & Lysosomal Disease Patient Conference

Melbourne, Australia

18-20 October 2024 (only **Saturday 19th** for the Patient conference)

Proudly supported by BDSRA Australia, come and say 'hi' at our table

For more details and rego: [LD Summit](#) OR [Patient Conference](#)

Save the date - 2025



NCL2025 (the bi-annual International Congress on NCLs)

Novotel Twin Waters Resort on the Sunshine Coast

28 October - 1 November, 2025



BDSRA Australia Family Conference

Novotel Twin Waters Resort on the Sunshine Coast

Friday **31st October** to Sunday **2nd November, 2025**

Past



International Batten Disease Awareness Day & Bounce4Batten

Another wonderful year of creating awareness and fundraising.

Each year on 9th June - **next year is Monday 9th June 2025**

See more about what happened this year under Fundraising



Funrun Mania!

City2Surf in Sydney on **Sunday, 11th August 2024**

Beach2Beach in Sydney on **Sunday, 25th August 2024**

Thank you to those who participated and fundraised. Further updates with the amazing effort in our Fundraising section.



Virtual Coffee catchup - Bereaved

In the evening of Thursday 13th June, a group of our bereaved parents caught up via zoom from around Australia. We had regulars and first timers to a bereaved catchup, from recently bereaved to 10+ years. Some old connections were re-established as we talked over topics such as life updates, back to work, latest treatments, how to respond to hard questions, reminiscing, holiday/break plans and funruns.

Donations

[Donate Now](#)

Thank you so much for your support!

It's so hard to believe we are about to begin the final quarter of the year!

We would like to say a huge THANK YOU to those participating in the City2Surf and Beach2Beach for your efforts and fundraising! These incredible people have almost reached the \$9000 mark in [fundraising for BDSRA Australia](#). The amazing effort had us reach #3 on the [Beach2Beach Fundraising leader board](#).

We would also like to extend a heartfelt thank you to everyone who made recent donations. We had a wonderful response to the EOFY drive, and knowing what a busy time of year it is, makes us all the more grateful to those who took the time to donate. Thank you also to those who made recent **bereavement donations** in honour of their loved ones. We truly appreciate your thoughtful and generous gifts.

We continue to be on the recruiting drive for a **fundraising committee**. If this is something that you may be able to spare some time to help with, we would love to hear from you! Please email expressions of interest to treasurer@bdsraaustralia.org. It is an area we would love to expand on, but cannot achieve this without extra hands on deck, so please reach out if this is something that you are passionate about and able to assist with.

Amanda Gilpin
Treasurer

'More Perils of Motorcycling' by Alanna Gayko

Alanna Gayko was inspired by the Batten disease cause in 2000, so embarked on a solo journey around Oz on her trusty motorbike. Not only did she raise awareness and funds for BDSRA Australia but she wrote and published a book in 2010 to capture her adventure. All copies of that book sold out and proceeds were donated entirely to BDSRA Australia.

Alanna now has another book published and we were lucky enough to be at Alanna Gayko's recent book launch of her 2nd edition of the series, pictured below.

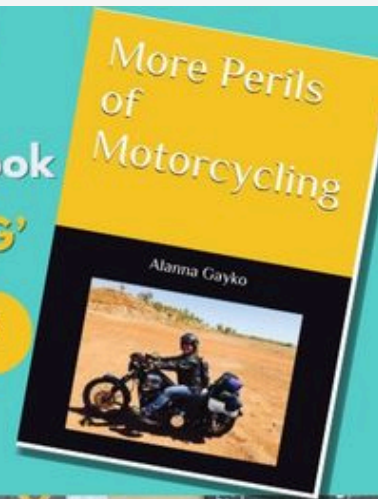
Alanna is once again generously **donating all proceeds of her book sales to BDSRA Australia**. Please get behind Alanna's momentous mission and get your own copy [here](#).

Support BDSRA Australia and purchase yourself a copy of Alanna Gayko's wonderful new book

'MORE PERILS OF MOTORCYCLING'

Order your copy from **amazon** now!

All proceeds directly benefit
BDSRA Australia.



Fundraising



International Batten Disease Awareness Day and Bounce4Batten (B4B)

We had a wonderful year creating awareness and fundraising for Batten disease.

- **online** Bounce4Batten campaign with reels and pictures of people bouncing
- **VIC** - Epsom Primary School (Spark family)
- **SA** - Battle in the Hills 2 - Mt Barker (McWaters family)
- **QLD** - Little Scholars Stapylton, Rise Early Learning Surfers Paradise (Theo's family)
St Mary's School & Laidley (Emily's family)
- **WA** - Spencer Park Education Support Centre (Gilpin family)

Here is a gallery of photos from the various local events [here](#)

Funrun Photos!

A big **THANK YOU** again to all who participated and donated to BDSRA Australia in Sydney's **City2Surf** and **Beach2Beach** events in August. Here is our photo gallery with some quotes from participants.



For more information on upcoming Funruns, please see our [website here](#).

If you are interested in participating, putting in a team and fundraising in an upcoming funrun event around Australia, please let us know and we can assist in setting up the charity page for you!

ASX Refinitiv Charity Foundation

As one of the benefiting charities for the 2024-25 round of the ASX Refinitiv Charity Foundation program. BDSRA Australia will continue to receive funds from this wonderful organisation and work alongside them supporting their annual fundraising events.

The **ASX Refinitiv Charity Foundation Raffle** is now live and we are committed to selling \$10,000 worth of tickets. Last year, one of our very own Batten supporters won 1st prize - a new car, next time, this could be you! We are calling on our loyal families and friends to purchase and share with your own networks to promote this raffle as all tickets sold go towards the sum of the annual donation we receive from the Foundation. This year's prizes include:

1st prize: MAZDA2 Automatic Hatchback – valued at \$27,000

2nd prize: MacBook Air - Valued at \$1,599

3rd Prize: Apple Watch Series 9 – valued at \$809

Please click on the link below to **purchase your raffle tickets today and share this direct link** with your family and friends.

[Batten Disease Support and Research Association Australia \(asxrcfau.com\)](https://www.asxrcfau.com).

The next ASX event is the Financial Markets & Media Charity Sailing Regatta which will be held on Friday 29th November at Middle Harbour Yacht Club in Sydney. We are now seeking volunteers for this event. If you are interested in volunteering your time for a fabulous day of sailing excitement, please get in touch with Nikki Hopkins at info@bdsraustralia.org

Family Support

The town that turns PINK for Emily

Jessica Lyon's daughter Emily was diagnosed with CLN6 in 2021, being the only known child in Australia with this variant until very recently.

Their hometown of Laidley in QLD have backed Emily and her family to "Paint the town Pink" in support of Batten disease awareness. Jessica says "It means so much to know our community cares, their support is amazing"

For more of the story and photos, read [here](#).



We would love to hear from you!

To share your family's experience in the newsletter or for further information on how we can support you, please contact Liz at support@bdsraustralia.org

Lysosomal Disease Patient Conference - Melbourne

BDSRA Australia proudly supports the Lysosomal Disease Summit hosted by Fabry Australia this October 19-20 at the **Pullman Hotel Melbourne**. In conjunction with the main Summit, a **one-day Patient Conference** will be held on **Saturday 19th October** with FREE registration for patients and families.

We encourage Batten families to come along! There will be a variety of Batten-specific and other relevant talks for families throughout the day. You will have the opportunity to learn about lysosomal disease (LD) science, clinical research and healthcare policy, and connect with other families in the LD community.

BDSRA Australia's Dr Ineka Whiteman (Head of Research & Medical Affairs) and Liz Sanson (Family Support Coordinator) will be presenting and hosting an exhibition table, so come along and say hello!

For further events details including the preliminary program and link to register head to [Lysosomal Disease Patient Conference](#)



Honouring our recent Angels



Luke & Curtis Halley

Luke Halley gained his angel wings aged 26 on the 14th March. Luke's cheeky smile, infectious spirit and zest for life will be missed by all that knew him. He really was a large character in their tight-knit family. Memories are filled with the outings and adventures the family did together.



Just a few weeks after his younger brother, Curtis Ray Halley departed aged 28, to be reunited with Luke. The pair lived life to the fullest, two peas in a pod as they bravely battled Juvenile Batten disease. Curtis Ray had the biggest smile and warmest heart, all will miss his beautiful spirit, kindness and his love of music and animals.

We send our love and wishes for comfort amidst the immense grief to their parents Sara and Jeff, their brothers Lee and Ryan and all the extended family.



Steve Waugh Foundation

Supporting children and young adults with Rare Diseases.

"We take a holistic approach in supporting children and young adults (0-25yrs) with the rarest diseases. We work to improve their quality of life by providing support and opportunities to connect with others through: [individual grants](#), [respite camps](#), and raising awareness."

[Steve Waugh Foundation](#) has supported families affected by Batten disease over many years when there was no one else to turn to.

[Grant Guidelines](#) and Application [Requirements](#) explain where they can help and what you need to provide.

[How to Apply](#) Grant [Round 3](#): 1 - 30 September 2024



Managing behavioural challenges in Batten disease

Dementia Support Australia offers free, nationwide specialised Childhood Dementia Support with trained consultants who understand childhood dementias like Batten disease, its related behavioural and cognitive challenges, and the impacts these can have on the child, their family and carers.

No referral needed. Simply call the 24-hour helpline on 1800 699 799, fill out the contact form or chat with DSA online.

To learn more, visit [here](#).



'Max & Abby' book turns 2!

We're excited to celebrate the 2nd anniversary of our published children's book 'Max and Abby'!

A key resource to help young children, especially siblings and peers, understand Batten disease and the changes and challenges that occur in affected children over time.

Written by Dr Ineka Whiteman, developed by

BDSRA Australia in collaboration with Dementia Support Australia (DSA).

Read more about the book on our [webpage](#).

Order your copy now via Amazon

Purchase your copy for AUD \$16.66 (+P&H) via [Amazon.com.au: Books](https://www.amazon.com.au/Books). All proceeds go to BDSRA Australia's support and research initiatives.

Families! BDSRA Australia is delighted to provide one complimentary copy to all families in Australia with a child affected by Batten disease (currently caring for, or bereaved). Head to our website and fill in the order form: <https://bdsraaustralia.org/Max-Abby>.

Not on social media?

Keep up-to-date on BDSRA Australia's latest news and events on our website!

We frequently add items to our website [News page](#), so feel free to head over anytime to check out the latest in research and support news!

Research Updates

Compiled by Dr Ineka Whiteman, Head of Research and Medical Affairs



Photo: Dr Ineka Whiteman with Prof. Jon Cooper (left) at the Pediatric Storage Disease Lab in St Louis, Missouri USA.

Highlights from USA visit in July

In the first half of July, I was excited to visit two US Batten Disease Centers of Excellence, tour the Pediatric Storage Disorders Lab in St Louis Missouri, and attend the US BDSRA Foundation Annual Family Conference also in St Louis.

As part of the recently launched [US Clinical Centers of Excellence](#), for which I am proud to be Program Lead in my role with the BDSRA Foundation, I was invited to attend the **Batten Disease Clinic at Children's Hospital Colorado in Denver**, led by Directors Dr Kourtney Santucci MD and Dr Scott Demarest MD. It was a wonderful opportunity to see their multidisciplinary clinic in action, with Batten patients being seen by neurology, clinical genetics, paediatrics, neuropsychology, psychiatry, speech pathology, occupational therapy and physical therapy over 1-2 days as part of their 6-monthly visits.

Similarly, at the **Children's Hospital of Orange County (CHOC) in Los Angeles**, I was invited to meet with the Batten Disease Clinic Director, Dr Ray Wang MD, and the multidisciplinary team, and to present on the global prevalence of NCLs, international patient group initiatives and the Centers of Excellence program.

In St Louis, Missouri, I was honoured to tour the laboratory of NCL research legend Professor Jonathan Cooper PhD, to meet his team and observe their neuropathology research in action. **Prof. Cooper's Pediatric Storage Disorders Lab** is the leading international center for the morphological analysis of pathological changes in the central and peripheral nervous systems in NCL.

St Louis was also the location of this year's **BDSRA Foundation Annual Family Conference**. This is my third year in a row attending conference in person and can honestly say, it seems to grow bigger and better every year. I was absolutely blown away by the atmosphere, sense of community, robust science and more than 50 clinicians and scientists who attended in person, and we had nine different research studies that families could participate in on-site!

The conference program included the main **Research Session** with presentations covering Batten research advances 'from lab bench to bedside', latest **clinical trial updates**, the '**Ask-A-Doc**' panel with Centers of Excellence (CoE) Directors, concurrent sessions on '**Behavior, Cognition and Learning**', '**Palliative Care – Debunking the Stigmas**', and the '**Ask Me Anything: Scientists Edition**'. A number of these sessions were recorded. If you are interested in viewing any of these please reach out at research@bdsraaustralia.org



Photo: Children's Hospital Colorado Batten Disease Clinic team members with the Fries family. Dr Andrea Miele, Neuropsychologist (back right), Dr Kourtney Santucci, Co-Director (2nd from right) and Dr Scott Demarest, Co-Director (Front centre).



Photo: Children's Hospital Orange County (CHOC) Batten Disease Clinic and Neurology/Metabolic team members with Clinic Director Dr Ray Wang (seated right) and Dr Ineka Whiteman (seated left).



Photo: The Pediatric Storage Disorders Lab team (L to R): Sophie Wang, Jon Cooper (Principal Investigator), Ineka Whiteman (BDSRA), Beth Eultgen, Ewa Ziółkowska, Bart Olszowy and Karl Jackiewicz.



Thank you for helping to shape the future of Batten disease research

Over the last two months, we were so encouraged by the global response to the two-part Research Priority Setting community survey initiative conducted by the BDSRA Foundation and Kennedy Krieger Institute in Baltimore, Maryland USA.

To our knowledge, this is the first time this type of validated research priority setting method has been undertaken for Batten disease.

Through this process, almost 200 participants in Survey 1 posed over 400 research questions that they felt were vital for the Batten disease research field. Those questions were collated and summarized into a ‘shortlist’ of popular research questions and in Survey 2, participants were asked to rank the ‘Top 10’ research questions important to them.

The results of this two-part survey determined the ‘Top Priority Research’ questions that are now a critical part of the [Batten Disease Global Research Initiative Research Strategy](#). This will now help to drive future investment in Batten disease and ensure we are focused on fostering research that pragmatically addresses the topics of highest importance to the Batten community.

Whether you’re a parent, clinician, researcher, industry representative, policy maker, educator,

allied health or support worker, THANK YOU TO EVERYONE who participated in this important initiative!

Clinical Program Updates



Tern Therapeutics and REGENXBIO - Gene Therapy for CLN2 Batten disease

We are tremendously heartened to share that on August 27, Tern Therapeutics announced its launch and acquisition of the investigational gene therapy programs RGX-381 and RGX-181 from REGENXBIO, for the treatment of CLN2 disease.

In its [press release](#), Tern announced it has entered into a global licensing agreement with REGENXBIO for both gene therapy products, now designated TTX-381 and TTX-181, respectively, to form its initial therapeutic pipeline. TTX-381 and TTX-181 are novel one-time gene therapy products being developed for the treatment of ocular and central nervous system (CNS) manifestations CLN2 Batten disease, respectively. Tern also advised its closing of a \$15 million funding round to accelerate ongoing clinical testing of TTX-381 and to advance its pipeline.

In a show of unity, REGENXBIO simultaneously released [a letter to the Batten community](#) expressing their support for these new developments and the team at Tern Therapeutics.

In recent years, we have seen highs and lows in the development of these CLN2 gene therapy programs, as outlined in the timeline below. BDSRA Australia, along with our global patient advocacy partners, has worked diligently behind the scenes to support the process and ensure our CLN2 community had a consistent voice at the table. We look forward to continuing the collaboration with Tern to ensure these programs continue advancing as swiftly and effectively as possible.

Read Tern's full press release [here](#), which includes statements from Dr Ineka Whiteman and BDSRA Foundation President, Amy Fenton Parker.

For more information, visit the Tern Therapeutics website (<https://www.terntx.com>) or contact patientadvocacy@terntx.com

THE YEAR IN REVIEW: Timeline of events for the CLN2 gene therapy programs

- **August 2, 2023:** RGX-381 (ocular) program update – First patient dosed in Phase I/II trial at Great Ormond Street Hospital (GOSH), London
- **August 31, 2023:** RGX-181 (CNS) update – Initial interim data from the single patient study (investigator-initiated trial or 'IIT') presented at the SSIEM Annual Symposium in Jerusalem

- **September 30, 2023:** RGX-381 (ocular) program update – Phase 1/2 safety data presented at International Congress on NCL in Hamburg Germany
- **November 8, 2023:** REGENXBIO releases its third-quarter earnings release, announcing a corporate restructuring that included the decision to halt the development of multiple investigational AAV gene therapies, including the RGX-181 and RGX-381 programs
- **November 8, 2023:** REGENXBIO releases a letter to the Batten community noting that the dosed patients will continue to be followed and that new patients will not be enrolled in the two trials. REGENXBIO also assures commitment to identifying a partner to advance the investigational therapies to patients
- **November 9, 2023:** Global Batten disease patient organizations release a joint statement assuring commitment to working with REGENXBIO and their potential partners to ensure the programs advance
- **November 21, 2023:** BDSRA meets with now-former REGENXBIO founding President and Chief Executive Officer Ken Mills to discuss strategic plans for progressing RGX-181 and RGX-381 and explore a range of options
- **November 2023-August 2024:** BDSRA remains in contact with REGENXBIO and potential partners
- **February 7-8, 2024:** Interim results for RGX-181 and RGX-381 presented at WORLDSymposium in San Diego, CA.
- **April 9, 2024:** Interim results for RGX-181 and RGX-381 presented during BDSRA Ask-An-Expert webinar.
- **August 27, 2024:** Tern Therapeutics announces acquisition of RGX-381 and RGX-181 (now designated TTX-381 and 181, respectively) CLN2 gene therapy programs.



Neurogene Inc. - Gene Therapy for CLN5 Batten disease

Phase 1/2 clinical trial now fully enrolled

On Friday, August 9, Neurogene Inc. announced enrolment is now complete for its Phase 1/2 investigational trial of NGN-101 Gene Therapy for the treatment of CLN5 Batten disease, with a total of six patients enrolled (Clinical trial ID: [NCT05228145](#)). Neurogene unveiled this news in its Second Quarter 2024 Financial Results and Highlights Recent Updates.

According to the report, Neurogene plans to provide interim clinical data and a regulatory update in the first quarter of 2025. Read our global update [here](#).



Beyond Batten Disease Foundation/Theranexus - Batten-1 Program for CLN3 Disease

18 month data from Phase I/II trial of Batten-1

Together with Theranexus, the Beyond Batten Disease Foundation is pleased to share the latest press release reporting positive efficacy and safety data after 18 months of treatment for CLN3 patients in the Phase I/II Batten-1 clinical study. These results include reduced levels of a recognized neurodegeneration biomarker, consistent with the absence of marked motor symptom progression. Read more here:

[Theranexus_PR_Results_18_Nf_biomarkersL_V153.pdf \(beyondbatten.org\)](#)

BioMarin Pharmaceutical - BRINEURA® (cerliponase alfa) for CLN2 Disease

BRINEURA® label expansion in USA: now approved for children under 3 years with CLN2 disease

In 2018, the enzyme replacement therapy Brineura (Cerliponase alfa) was approved in Australia for the treatment of CLN2 disease, with no age restrictions. In the USA however, Brineura was approved (in 2017) for treatment in symptomatic children 3 years of age and older with CLN2 disease. On July 24, in a significant advancement for the treatment of US children with CLN2 disease, the FDA announced its approval of an expanded label, now approving Brineura for children of all ages with CLN2 disease, regardless of whether they are symptomatic or presymptomatic.

In the BioMarin press release, BDSRA's Dr Ineka Whiteman commented: "The opportunity to start Brineura treatment earlier (in the US), even before the onset of symptoms, provides newfound hope for the families impacted by this rapidly progressive disease. Importantly, this expanded indication provides further impetus for early diagnosis of CLN2 disease, as we continue advocating for inclusion of CLN2 disease on the RUSP (Recommended Uniform Screening Panel) for newborn screening (in the USA)."

The full press release from BioMarin can be read [here](#).

Clinical trial updates - useful resources

The BDSRA Australia website provides an interactive search tool that enables users to locate current clinical trials listed on [Clinicaltrials.gov](https://clinicaltrials.gov), for all NCLs or by subtype. Check it out here: [Resources and Support for patients and families \(bdsraaustralia.org\)](https://bdsraaustralia.org).

In addition, a current summary and updates on NCL clinical programs has been compiled into an interactive table, in collaboration with the BDSRA Foundation. Visit this page for latest updates: [Clinical Studies – Batten Disease Support & Research Association \(bdsrafoundation.org\)](https://bdsrafoundation.org).

For any questions on any of these programs, feel free to reach out at research@bdsraaustralia.org



2nd Annual Lysosomal Disease Summit

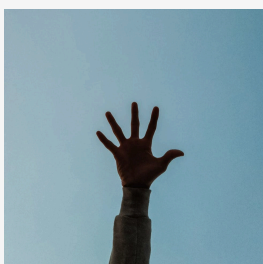
[Pullman & Mercure Melbourne Albert Park](#), Melbourne, Australia, 18 – 20 October, 2024

The Lysosomal Disease Summit will bring together metabolic and lysosomal disease healthcare practitioners and researchers from Australia and the surrounding Asia-Pacific region, to address the current challenges facing lysosomal disease clinicians and researchers, discuss the latest research updates, and identify opportunities for improvements in the diagnosis and treatment of lysosomal diseases.

The concurrent Patient Conference and Lysosomal Disease Summit will be an important opportunity for clinicians and researchers to come together to advance the field of lysosomal disease research and treatment. The Summit will also provide an opportunity for attendees to network, find collaborators, advance careers, and build relationships.

We encourage you to **share this message** with anyone within the Lysosomal Disease community, whether they are affected individuals or involved in related research endeavors.

[Click here for more information](#). **Early Registration** ends 16 September, 2024. [Register Today!](#)



BDSRA Australia Family Register –
please consider joining!

We've had a great response to our previous calls for parents and carers to join the BDSRA Australia Family Register, but we know there are many current and bereaved families who have not been captured on this important database.

The Family Register is a vital tool that enables us to keep you informed of ongoing Batten disease research, including future clinical research opportunities.

The Register also enables the BDSRA Australia to better understand the prevalence of Batten disease in our region, including the different subtypes and geographical locations. This helps us tailor our education and support activities according to the needs of our families.

The Register is open to all current **and bereaved** families.

The information collected in this form is kept STRICTLY CONFIDENTIAL. Your involvement in this survey is entirely voluntary, and you may request to be removed from the list at any time.

The form takes just a few minutes to complete and can be accessed here:

 [BDSRA Australia Family Register form](#)

Research Publications - Selected



A selection of recent research publications in Batten disease are provided below. Summaries and links to each article are [here](#).



Targeting autophagy impairment improves the phenotype of a novel CLN8 zebrafish model



Phenotypic/Genotypic Profile of Children with Neuronal Ceroid Lipofuscinosis in Southern Brazil



Mutations in CLCN6 as a Novel Genetic Cause of Neuronal Ceroid Lipofuscinosis



Intraventricular Cerliponase Alfa Treatment in a Patient with Advanced Neuronal Ceroid Lipofuscinosis Type 2



TRPML1 activation ameliorates lysosomal phenotypes in CLN3 deficient retinal pigment epithelial cells



Reduction of neuroinflammation and seizures in a mouse model of CLN1 batten disease using the small molecule enzyme mimetic, N-Tert-butyl hydroxylamine



Editorial: Neuronal ceroid lipofuscinosis: molecular genetics and epigenetics



Neuronal ceroid lipofuscinosis in a Schapendoes dog is caused by a missense variant in CLN6



Neuronal Ceroid Lipofuscinosis in a Mixed-Breed Dog with a Splice Site Variant in CLN6



OCT Biomarkers in Ocular CLN2 Disease in Patients Treated With Intravitreal Enzyme Replacement Therapy



Ceroid lipofuscinosis type 2 disease: Effective presymptomatic therapy-Oldest case of a presymptomatic enzyme therapy

Summaries and links to each article above are [here](#).

Personal Details Update: To ensure you are kept informed and up to date on all our news and events, please be sure to let us know if your circumstances or contact details have changed.

Please email Nikki Hopkins at info@bdsraustralia.org



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Australian Registered Body Number: 073 435 202

Australian Business Number (ABN): 56 056 138 99

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