NEWSLETTER

Summer 2024 Edition

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At a glance:

President's Report

Photo: Andreas & Julia Mickler (Secretary), Jeremy & Amber Rundle (President), Vanessa Anderson (Public Officer), Kim Griffiths, Sherilyn & Harry Partridge at the ASX Refinitiv Charity Foundation Imagination Gala Ball, 23rd February 2024.

Welcome to our first newsletter for 2024. I'd like to start by acknowledging that today is Rare Disease Day!
Rare Disease Day is a global movement focused on advancing equity in healthcare, social opportunity and access to effective diagnosis and treatment for people living with a rare disease. The national peak body for the estimated 2 million Australians living with a rare disease is Rare Voices Australia If you'd like to learn more or get involved please visit either of these links.

Last Friday, 23rd February, a small group of past and present board members had the privilege of attending the 2024 ASX Refinitiv Charity Foundation Imagination Gala Ball. It was a fantastic night of fierce fundraising, fine food and wine and fabulous entertainment all for the purpose of bringing financial and corporate Australia together to raise much needed funds for charity.

BDSRA Australia had three auction items in the silent auction and raised $1680 from the sale of these items. We'd like to give special mention and thanks to The Monogram Mode, Welcome Hospitality and Pandora Jewellery for kindly donating the auction items.

There is just one week left to get your ASX Refinitiv Charity Foundation raffle tickets so if this has been on your to do list, now is your time to be in the running! Further details are outlined in this newsletter.

With the exciting announcement that Australia has been selected as the destination for the 19th International Congress on Neuronal Ceroid Lipofuscinoses we are now investigating how we

might align our next Family retreat with this conference. By doing this we aim to give our families the opportunity to access and get the latest advances in basic science, translational and clinical research in the Battens field on an international scale. We will keep you updated on our progress as we learn more from the organisers.

I want to acknowledge that the last few months may have been tough on many of our families as we have learned about some disappointing developments for a number of clinical programs across different CLN subtypes. Dr Ineka Whiteman has expanded on these developments in the Research Updates sections below. We know that the biotech sector is particularly vulnerable to the global economic challenges we have seen in recent years and I want to assure our families that BDSRA Australia, together with our team of international experts and other global patient bodies, have been keeping abreast of the changes, and continue to apply 'diplomatic pressure' on our industry partners to ensure they remain cognisant of the pressing needs and voices of our Batten community. BDSRA Australia will continue to keep our families informed of any program developments as more information comes to light.

We are here to support you, provide clarity and answer any questions you may have. Our Facebook page, these newsletters, and our website are also valuable resources for the latest updates and, as always, our team is here should you need to reach out to us directly anytime.

Warmest regards,
Amber

Research Highlight

CONGRATULATIONS to our 2023-24 BDSRA Australia Research Grant Winners!

In December, we were delighted to announce the winners of our 2023-24 Research Grants, with a total of over $88,000 awarded to three projects. Congratulations to our Chief Investigators:

Prof. Stephanie Hughes from the **Brain Health Research Centre - Otago in New Zealand** who has been awarded $35,000 for work developing a personalised medicine approach to CLN1 Batten disease; with a special thanks to Mila's Miracle Foundation for partnering with us to support this project;

Dr Lotta Oikari from **QIMR Berghofer Medical Research Institute QLD** awarded $24,961 for continued work investigating repurposed drug candidates for the treatment of Batten disease; and

Dr Jenna Ziebell from **Wicking Dementia Centre, University of Tasmania** awarded $28,290 for research exploring the role of microglia in CLN3 Batten disease.

We are extremely proud to support this world-class, promising research right here in our region. Further details on the successful projects and research teams are now posted on our website.

A special thanks also to our international Grant Review Panel for their time and expertise, and to all of our wonderful donors and supporters - without your generosity, this research would not be possible.

To help us continue funding research towards treatments and cures for Batten disease, please consider making a tax-free donation today by visiting our website:
https://bdsraaustralia.org/index-donate.php

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

Events

Upcoming

Virtual Coffee catchup - General Catch up
Join us for a relaxed chat, an opportunity to meet other parents from around Australia and NZ, to discuss topics that attendees raise.
Thursday, 14th March - 11:00 AM - 12:00 PM (AEDT)
Register your RSVP here.

Virtual Coffee catchup - General Catch up
Join us for a relaxed chat, an opportunity to meet other parents from around Australia and NZ, to discuss topics that attendees raise.
Wednesday, 27th March - 8:00 PM - 9:00 PM (AEDT)
Register your RSVP here.

International Batten Disease Awareness Day & Bounce4Batten
Let's raise awareness and take the opportunity to fundraise.
Please note the date in your diary!
Sunday, 9th June 2024

Past

Research Year In Review webinar
On December 14, BDSRA Australia co-hosted the '2023 Research in Review', along with our friends at the BDSRA Foundation. Panellists Ineka Whiteman, David Pearce, Jonathan Cooper, Kourtney Santucci, and Tony Cook discussed the major NCL research takeouts and highlights for 2023, followed by Q&A. Watch a recording of the event here: 2023 Batten Disease Research in Review Webinar (youtube.com)

REGENXBIO CLN2 gene therapy update - webinar
On December 1, BDSRA Australia's Head of Research & Medical Affairs, Dr. Ineka Whiteman, provided the latest news on REGENXBIO's decision to halt their gene therapy program for CLN2 disease. The webinar also included further updates on all other CLN2 studies, followed by a Q&A. Ineka was joined by panelists BDSRA Foundation (USA) board members Dr. David Pearce and Dr. Fernanda Leal-Pardinas. You can watch a recording of the event here: REGENXBIO Webinar | BDSRA Foundation - YouTube

Virtual Coffee catchup - Bereaved Catchup
In the evening of Wednesday 6th December, a group of our bereaved parents caught up via zoom from around Australia and NZ. Each meeting is a unique mixture of people and we go with the flow. It's an opportunity to connect, share and support each other.

Donations

Donate Now

Thank you so much for your support!

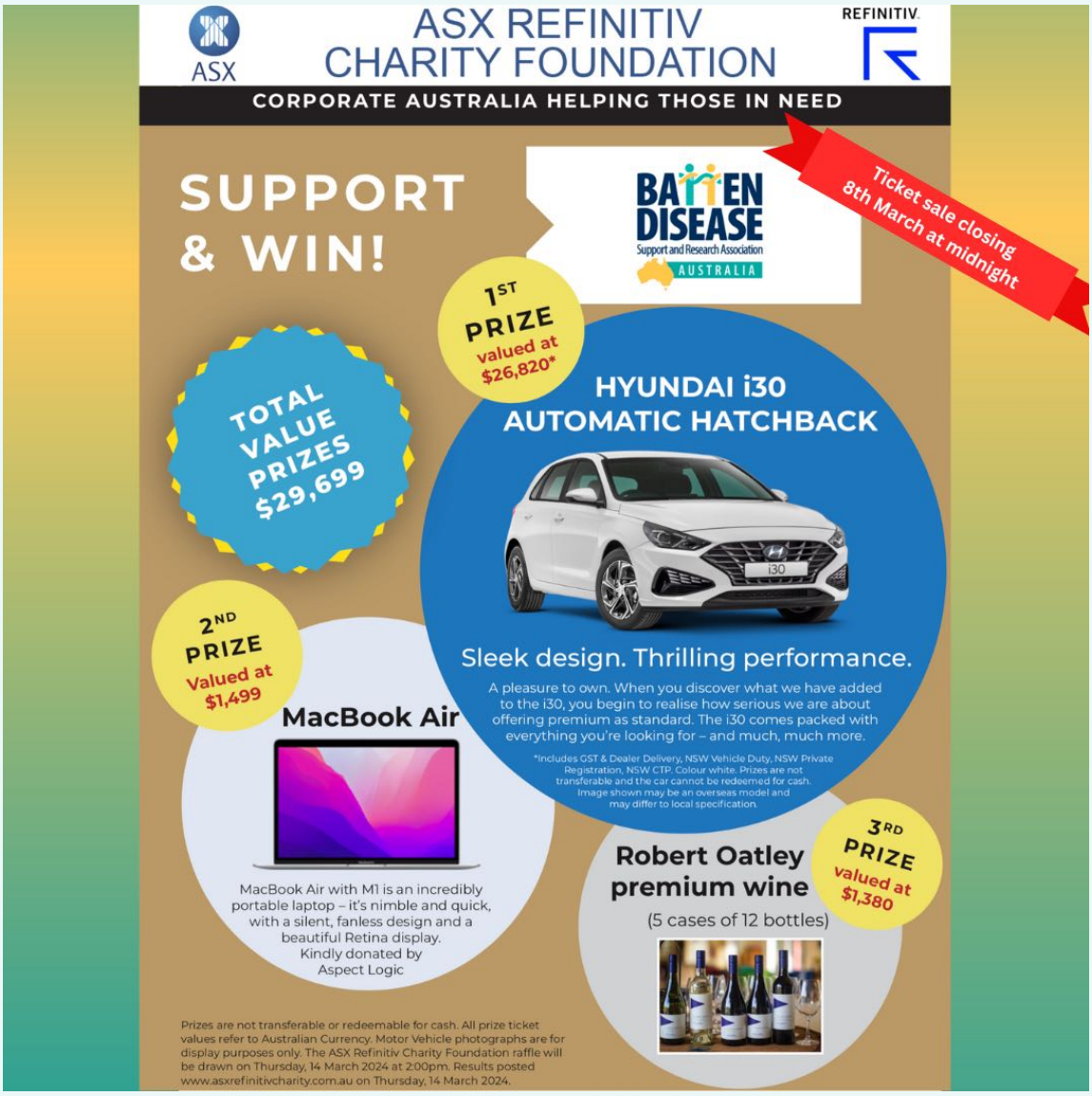
How is it almost March already!

We are in the process of planning the year ahead. There are some fantastic fundraising opportunities coming up with the fun run season not far away! The Perth HBF Run for a Reason will be the first one off the mark in May. Each state has their own events, so jump on board, get fit, have fun, and make some awesome memories while raising crucial funds for BDSRA Australia.

We're forming a **Fundraising Sub-committee** and we are still looking for more volunteers who would like to put their creative ideas to good use for our incredibly worthy cause. If you can participate or are interested in facilitating the committee, we would love to hear from you! This is open to our Batten families and also the wider community of family, friends and supporters who want to make a difference.
If this is something you are interested in, email either treasurer@bdsraaustralia.org or info@bdsraaustralia.org.

Amanda Gilpin
Treasurer

Fundraising



ASX Refinitiv Charity Foundation Raffle

HURRY! There is just over a week left to get your tickets!! **Ticket sales close midnight Friday 8th March!!**
Draw Date: Thursday 14th March 2024 at 2pm
Profits from all tickets sold through our unique online link go directly to BDSRA Australia.

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)

Fun Runs!!

Last August, many members from our community enjoyed participating and fundraising at the City2Surf and Beach2Beach held in Sydney, the dates for 2024 have already been announced!

If you are interested in participating, or putting a team together and fundraising in an upcoming funrun event around Australia, please let us know and we can assist in setting up or connecting you to our charity page! Contact Nikki at info@bdsraaustralia.org

Check out the Biggest Fun Runs in Australia!

Here are just a few of the upcoming fun run events in 2024!

Sun 19 May - HBF Run for a Reason, CBD to Gloucester Park, Perth, WA
Sat 6-7 July - Gold Coast Marathon, Marine Parade, Southport, QLD
Sun 21 July - Run Melbourne, Melbourne, VIC
Sun 11 August - City2Surf Sydney, Hyde Park to Bondi Beach, Sydney, NSW
Sun 25 August - Beach2Beach, Dee Why Beach to Newport Beach, Sydney, NSW
Sun 8 Sept - Bridge to Brisbane, Gateway Bridge to Brisbane Showgrounds, QLD
Sun 15 Sept - City-Bay Fun Run, Festival Theatre, Colley Terrace, Adelaide, SA

..... But it doesn't have to stop at fun runs, there are many other sporting event opportunities such as; cycling, ocean swims, triathlons, marathons, car rallies, big walks and sailing regattas to name a few.

If you have family, friends or community members fundraising for BDSRA Australia and are happy to share with us for our next newsletter or on our social media, please let us know!

Bottles4Battens - Return and Earn

What is your New Years resolution??
Why not convert your litter reduction and recycling into dollars for BDSRA Australia.

Share our unique barcode below with family and friends, school and sporting groups and encourage them to donate all or some of their drink container refunds to support our charity.

Ready to recycle?

Batten Disease Support and Research Association Australia

Scan your barcode at the machine

Family Support

Photo: Hollie receives her long-awaited modified bike

Hollie's bike

After waiting over 2 years, Hollie finally got her wheels earlier this month! This new bike has come all the way from Germany and with it's specific adaptations, means she can now ride just like all her family and friends!

The benefit of this piece of equipment goes further than just social inclusion and providing the opportunity to participate in typical childhood experiences. It has a list of therapeutic benefits and is able to adapt as support needs change, ensuring Hollie maintains her physical and cognitive function for as long as possible.

Read more about this heartwarming story here.

TODAY is Rare Disease Day
#RareDiseaseDay #LightUpForRare #ShareYourColour
Rare Disease Day occurs on the last day of February annually, this year, it falls on 29 February 2024 – the rarest day in the calendar!

Today we join forces globally to raise awareness and promote change for over 300 million people living with a rare disease. Here are some basic ways you can get involved: update your social media profile by downloading any of these banners, graphics, backgrounds or filters, or share a rare story using the above hashtags.

We've chosen the 2022 family retreat group photo, to show our own Rare and wonderful community. We come together through shared experiences, the impacts of Batten disease and the passion and advocacy to see change and a cure for those affected by Batten Disease.

In case you missed it!
Feeding Tube Awareness Week #FTAW2024

We want to increase awareness and understanding around feeding tubes both for our families and in the community. Feeding tubes will often become an option at some stage, to provide children and young people with Batten Disease another way to give medication, fluids and food when they battle swallowing or feeding challenges.

We want shine a light on this topic, encourage discussions and information sharing to address stigma, fear, and misinformation. It's important to highlight both the benefits of opting for this intervention for our loved ones as well as the day-to-day impact of tube feeding on individuals, carers and families.

Here are some links if you want to know more:
Feeding Tube Aware - official awareness website by ausEE
ChildFeeding.org - a lot you can investigate on it, from fact sheets to tips and tricks
The Blend - A magazine that can be purchased or read online, stories and info
TubieFun - just one of many shops and ways to get feeding tube accessories
for more information sharing or if you want to discuss your experiences, please chat to Liz our Family Support Coordinator.

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

Family Register - Reminder

Have you filled in the Family Register form?

Our new Family Register form, created in May 2023, is how BDSRA Australia keeps families informed of **clinical research opportunities**, and to better understand Batten disease in our region and the **needs of our families**. If you haven't completed it, we ask that you take the 2-3 minutes to fill it in. Your details are kept STRICTLY CONFIDENTIAL at all times. Fill in the form here!
BDSRA Australia Family Register form (google.com) 📋

Remembering Our Angels - Online Memorial

BDSRA Australia's Memorial gallery

BDSRA Australia has a Memorial gallery to remember our Batten Angels. Scroll past the family stories to see it here.

If you had a child/ren pass away from Batten Disease in Australia and New Zealand and wish to have them included in our online memorial, please contact Liz at support@bdsraaustralia.org

Resources

Starlight Children's Foundation

Starlight loves to bring some joy and happiness to sick kids, who going through a difficult time, including those diagnosed with Batten Disease.

Wishgranting - It's a once-in-a lifetime gift that gives these kids something to look forward to and create new memories to treasure forever.

Starlight Express Rooms - Found in major hospitals, they're filled with fun and laughter and feel a world away from the reality of hospital.

Livewire - Starlight's dedicated program for teenagers living with a serious illness, disability or chronic health condition.

Siblings Australia inc

In 2024 Siblings Australia celebrates 25 years of support and advocacy for siblings of people with disability and chronic illness.

The Sibling Australia website has a range of information, resources and services for siblings, parents and professionals.

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.

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

Dr Ineka Whiteman, Head of Research and Medical Affairs

Clinical Program Updates

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, for all NCLs or by subtype. Check it out here: Resources and Support for patients and families (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)

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

FEATURE: NZ Researchers Play Pivotal Role In Bringing Gene Therapy To Clinical Trial

For more than 10 years, New Zealand-based researcher Dr Nadia Mitchell has dedicated her career to understanding Batten disease and potential treatments through extensive work in sheep models of CLN5 and CLN6 disease. Along with longtime NCL researcher and collaborator Prof. David Palmer and postdoctoral researcher Dr Samantha Murray, Mitchell's team at Lincoln University has focused on brain- and eye-directed gene therapies in two naturally occurring Batten disease sheep models, and on translating gene therapy products for CLN5 and CLN6 disease from those sheep into human clinical trials.

Significantly, this research has prompted the approval of an Investigational New Drug (IND) application by the US FDA, which enabled the initiation of the first in-human gene therapy clinical trial for the treatment of CLN5 disease in 2022 (Clinical Trial ID NCT05228145), and dose-escalation and expansion of the study in 2023. This work has been celebrated in international and local media stories.

BDSRA Australia is immensely proud to have contributed over $110,000 toward this pivotal research in the past four years, with two BDSRA Australia Research Grants awarded to Dr Mitchell in 2020 and 2022. This month, Dr Mitchell submitted the final report for her most recent Grant, and we are delighted to share this summary of the project findings below. Impressively, this work has led to the publication of no less than five peer-reviewed research publications in the past 12 months, and links to those papers are also provided below.

Congratulations Dr Mitchell, Dr Murray and team, and thank you for the dedication and HOPE you bring to our families and community.

Project: Ongoing gene therapy and peripheral pathology studies in ovine (sheep) CLN5 and CLN6 Batten disease
2022-23 Grant: $ 54,545

Sheep with naturally occurring CLN5 and CLN6 Batten disease at Lincoln University have been instrumental in better understanding the disease and developing treatments. This study demonstrated the long-term safety of increasing doses of ovine and human CLN5 gene therapies in CLN5 affected sheep. Brain- and eye-directed gene therapies halted or slowed disease progression and vision loss, and extended survival.

Very high doses of the ovine CLN5 gene therapy product provided the most robust response, with sheep clinically normal at three years of age which was double their natural life expectancy. The human CLN5 gene therapy product was less effective in sheep but was still biologically active and safe. Our in-life and post mortem data from the sheep study supported the dose escalation and expansion of the current Phase 1/2 clinical trial of the human CLN5 gene therapy product (Clinical Trial identifier: NCT05228145).

However, as more treatments become available for Batten disease, there is a pressing need for effective ways to monitor disease progression and treatment efficacy. Our sheep study found that blood plasma levels of the neurofilament light chain (NfL) protein was an effective biomarker of neuronal damage, which could be used in parallel with functional and clinical scoring and brain MRI imaging in the clinic. Finally, most treatments for Batten disease have focused on the brain and the eye, so we continue to examine disease pathology in the peripheral tissues (heart, liver, lung, kidney, gastrointestinal tract etc) as we may need to also treat these organs into the future.

Publications during the grant period:

• Murray SJ, et al (2023). Efficacy of dual intracerebroventricular and intravitreal CLN5 gene therapy in sheep prompts the first clinical trial to treat CLN5 Batten disease. *Frontiers in Pharmacology* 14:1212235 https://doi.org/10.3389/fphar.2023.1212235

• Mitchell, NL et al. (2023). Long-term safety and dose escalation of intracerebroventricular CLN5 gene therapy in sheep support clinical translation for CLN5 Batten disease. *Frontiers in Genetics* 14:1212228. https://doi.org/ 10.3389/fgene.2023.1212228

• O'Leary C, et al (2023). Intraparenchymal convection enhanced delivery of AAV to treat Mucopolysaccharidosis IIIC. *Journal of Translational Medicine* 21(1):437. https://doi.org/10.1186/s12967-023-04208-1

• Mitchell NL, et al (2023). Characterisation of neuropathology in ovine CLN5 and CLN6 neuronal ceroid lipofuscinoses (Batten disease). *Developmental Neurobiology* 83:127-142. https://doi.org/10.1002/dneu.22918

• Murray SJ, et al (2023). Progressive MRI brain volume changes in ovine models of CLN5 and CLN6 neuronal ceroid lipofuscinosis. *Brain Communications* 5:fcac339. https://doi.org/10.1093/braincomms/fcac339

Photo: Dr Samantha Murray (L) and Dr Nadia Mitchell (R) with CLN5-affected sheep. Photo credit: Lincoln University

Elpida Therapeutics - CLN7 Gene Therapy Program Update

On January 18, Elpida Therapeutics held an online meeting for the CLN7 disease community to discuss the AAV-based gene therapy (AAV9/MFSD8) program currently in Phase 1 trial for the treatment of CLN7 disease (NCT04737460), and Eplida's **plans to move to the next phase of clinical trials**. The meeting was attended by families, doctors, scientists and patient advocacy groups from all over the world.

The primary aims of the meeting were (1) how to most **effectively identify and connect with CLN7 disease patients** and their families globally, and (2) to discuss the potential design, strategy and initiation of a retrospective and prospective **natural history study,** a vital clinical trial component for evaluating the efficacy of the investigational gene therapy. Elpida Therapeutics is working hard to develop a standardized data collection approach and will hold another meeting with families in the very near future.

For further information on the CLN7 gene therapy program and to stay up-to-date with program developments, please reach out to Souad Messahel, Head of Clinical Operations at Souad@elpidatx.com.

A copy of the presentation from the January 18 meeting can be downloaded **here**.

Neurogene - CLN5 Gene Therapy Program Updates

On December 19, 2023, Neurogene released the following announcement:
"We are thrilled to share that the merger that we announced in July of this year between Neurogene and Neoleukin Therapeutics is now complete, which secures the funding we need to advance our mission of bringing life-changing genetic medicines to patients and families impacted by devastating neurological diseases. In connection with this merger, we are moving from a privately held company to a public traded company, and will begin trading today on the NASDAQ under the ticker 'NGNE'. "

Further to this, on January 5, Neurogene announced the **completion of enrollment of Cohorts 1 and 2 in the ongoing Phase 1/2 clinical trial** for CLN5 Batten disease (NCT05228145), with **interim clinical data** expected in the second half of 2024. Neurogene is **currently enrolling a final higher dose cohort** for this study.
In the fourth quarter of 2023, Neurogene completed a positive meeting with the FDA regarding the future potency assay for batches of the 'NGN-101' study drug. The FDA accepted Neurogene's proposed potency assay strategy and provided alignment with the testing approach, which will allow release of all future NGN-101 batches.

To enable advancement into a registration study, Neurogene is **collecting and analyzing natural history data** for CLN5 Batten disease and planning to request a clinical/regulatory strategy meeting with the FDA in the second half of 2024. The focus of this meeting will be to align with the FDA on the expected clinical requirements to support a streamlined registration pathway, which will be necessary **to move this program forward into a pivotal clinical trial.**

Read the full press release **here**.

CLN5 disease natural history study

In relation to these CLN5 gene therapy program updates, the BDSRA Australia would like to remind our community that the natural history study for CLN5 disease, mentioned above, **is still open for recruitment and seeking participants**. This is an international study, open to all individuals with a genetic diagnosis of CLN5 disease and where disease onset occurred at the age of 5 years or younger. **Current and bereaved** families affected by CLN5 disease are encouraged to participate.

For further information, please get in touch with the recruitment site University of Rochester Batten Center (URBC), NY , or reach out to Dr Ineka Whiteman, Head of Research and Medical Affairs via email.

Amicus returns CLN3 and CLN6 clinical gene therapy programs to Nationwide Children's Hospital

As many of you know, we have been awaiting further news regarding the future of the CLN3 and CLN6 gene therapy clinical programs led by Amicus Therapeutics. On February 6, BDSRA Australia along with its partner organizations around the world, released a Joint Statement to the Global Batten Disease Community regarding the following important updates relating to both programs:

In a Letter to the Community, Amicus advised that it has decided to return the rights to all its Batten programs to The Abigail Wexner Research Institute at Nationwide Children's Hospital (NCH), which includes the CLN3 and CLN6 clinical programs, and the CLN8 preclinical program. As such, NCH is now responsible for determining the next steps in developing those programs and for all follow-up with the CLN3 and CLN6 clinical trial participants and their families.

NCH has reassured our community that it remains committed to moving its CLN3 and CLN6 programs forward. Dr. Emily de los Reyes at NCH has provided further details on those plans in a separate Community Letter, which can be viewed here.

Patient dosed with 'TSH-118' gene therapy for treatment of CLN1 disease

On February 15, important news broke regarding progress in the CLN1 gene therapy program 'TSHA-118', currently in Taysha Gene Therapies' clinical development pipeline. In a statement, Taysha Gene Therapies announced that it had provided investigational clinical trial material for TSHA-118 to support the investigator-initiated trial (IIT) under an individual-patient Investigational New Drug (IND) at RUSH University Medical Center in Chicago, Illinois.

BDSRA Australia along with its partner organizations around the world, were quick to respond, issuing a Joint Statement to the Global Batten Disease Community that afternoon. A copy of the Joint Statement, including further details on the study, can be viewed here.

*** Please also see our note to families below regarding Natural History Studies for CLN1 disease.**

JCR Pharmaceuticals – CLN1 Enzyme Replacement Therapy Program Update

During WORLDSymposium earlier this month, JCR Pharmaceuticals held a small, in-person meeting with patient advocate groups and families affected by CLN1 disease. President Amy

Fenton Parker attended in person, while I joined the group online.
During the meeting an update was shared relating to JCR's investigational Enzyme Replacement Therapy candidate 'ATG-194', an intravenously-administered enzyme that is shown to cross the blood-brain barrier. ATG-194 has been previously studied in an 'n of 1' investigator-led study in a child with CLN1 disease under compassionate use in Germany (learn more here), with additional research and development ongoing.
The main purpose of the meeting was to bring experts together to discuss the initiation of a natural history study for CLN1 disease, and how we might all work together – families, patient groups, clinicians and industry – to advance robust collection and documentation of clinical data from CLN1 patients around the world. This study will lay an important foundation for the future development of therapies for the treatment of CLN1 disease. We look forward to sharing further updates on this program in the future.

*** Please also see our note to families below regarding Natural History Studies for CLN1 disease.**

Photo: Leaders from JCR Pharmaceuticals and MEDIPAL along with CLN1 families and patient advocates attended the San Diego meeting in person and online earlier this month.

Collaboration Pharmaceuticals – CLN1 Enzyme Replacement Therapy Program Update

Earlier this month, Collaboration Pharmaceuticals hosted a virtual meeting with patient advocate groups and CLN1 families from around the world. The purpose of the meeting was to share program updates relating to their lead pipeline product – a recombinant human enzyme replacement therapy delivered directly to the brain (intracerebroventricular or ICV) for the treatment of CLN1. Currently in preclinical development, the efficacy of this ERT has been described in mouse and sheep studies conducted by Prof. Jonathan Cooper and colleagues (learn more here). Collaboration Pharmaceuticals is currently manufacturing the protein in preparation for IND-enabling toxicity testing.

Discussion also focused largely on natural history studies for CLN1 disease to advance robust collection and documentation of clinical data from CLN1 patients around the world. We look forward to sharing further updates on this program in the future.

*** Please also see our note to families below regarding Natural History Studies for CLN1 disease.**

Therapeutic development for CLN1 disease: the importance of natural history studies

Natural history studies gather information of how a disease affects a person over a lifetime. Natural history data gathered in a robust and systematic way can help inform our understanding of disease, improve quality of care, and is essential for the design and execution of clinical trials of potential new therapies, particularly in rare diseases like all forms of Batten disease.

With ongoing development of multiple programs for treatment of CLN1 disease as described above, the BDSRA Australia and other patient advocacy groups are well-placed to work together with families, clinicians, researchers and industry to help facilitate and drive natural history studies for CLN1 disease. To enable us to support the advancement of all of these programs, we encourage all families who are affected by CLN1 disease (both currently caring or bereaved) to fill in the BDSRA Australia Family Register form (for families in Australia/NZ), or the BDSRA Foundation Family Register form (for families outside Australia/NZ) .

These Registers are of course not limited to CLN1 families only. We encourage families affected by all forms of Batten disease to fill in this form if you have not already.

CLN2 gene therapy program preliminary data presented at WORLDSymposium

As REGENXBIO continues to actively explore partnership opportunities to enable the ongoing development of its CLN2 gene therapy programs (see previous announcements on this here), it was encouraging to see preliminary data from the RGX-381 (ocular) and RGX-181 (CNS) gene therapy programs presented at WORLDSymposium in San Diego earlier this month. The presentations were:

RGX-381: Interim results from the first-in-human clinical trial of an investigational gene therapy for the treatment of ocular manifestations of CLN2 Batten disease
Presented by Dr Christina Ohnsman, REGENXBIO

Interim results from the first-in-human intracisternal dosing of RGX-181 investigational AAV9 gene therapy in a child with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2)
Presented by Dr Carolina Fischinger Moura de Souza, Hospital de Clínicas de Porto Alegre, Brazil

We are pleased to advise that REGENXBIO have agreed to participate in a 'WORLDSymposium Highlights' webinar in the coming weeks, where they will present these preliminary results and participate in a live Q&A. The webinar will be hosted by the BDSRA Foundation and Dr Ineka Whiteman, Head of Research & Medical Affairs.

Further details on this event will be posted soon, so keep an eye out on your inbox and our social media pages.

Update on the Beyond Batten Disease Foundation (BBDF) Batten-1 Phase III clinical trial

You may have seen the recent Theranexus press release. Theranexus has run into some unanticipated delays. Options are currently being considered and BBDF will schedule a family research call to follow up as soon as we have more definitive information and timelines to

share with you. If you would like to be included on BBDF's email list for important updates and information, please email info@beyondbatten.org

Conference news!

NCL2025 International Congress

Held every two years, the international Congress on Neuronal Ceroid Lipofuscinosis ('NCL Congress') is **the premier conference for NCL basic, translational and clinical research**. This meeting is primarily geared toward researchers, clinicians, industry partners and patient groups, however international families are also warmly invited to attend.

As many of you already know, **NCL2025 will be held in Australia in late-2025**, with details to be announced in the coming months. To assist with planning, the local Congress Organising Committee invites all potential attendees (researchers, clinicians, industry partners, patient groups and families) to fill in this Expression of Interest survey at your earliest convenience.

Research Opportunities

SPEECH & LANGUAGE STUDY – CLN2 and CLN3 disease

YOUR FINAL CHANCE TO PARTICIPATE!

Thank you to all those families who have already signed-up and participated in the **world-first research study** into the **characterization of speech and language in individuals with Batten disease** (CLN2 and CLN3 disease).

BDSRA Australia is proudly supporting this important study and would like to encourage our families in Australia, New Zealand and worldwide to participate before the enrolment period **closes on March 28, 2024**.

The team is seeking **10 more families** to enrol and complete the study (five CLN2 and five CLN3 families). The online assessment is reasonably brief (and enjoyable!), and Lottie is wonderful with our young participants! See one of our 'control subjects' below taking part.

To get involved, download the flyer via here or contact the team at Murdoch Children's Research Institute directly at geneticsofspeech@mcri.edu.au

Photo: Study participants take part in speech and language assessments over zoom with MCRI's research and speech pathologist Lottie Morison.

Research Publications - Selected

A selection of recent research publications in Batten disease are provided below. Read summaries and links to each article here.

🔬 First in man study of intravitreal tripeptidyl peptidase 1 for CLN2 retinopathy

🔬 REVIEW - Intravitreal enzyme replacement for inherited retinal diseases

🔬 Developmental skills and neurorehabilitation for children with Batten disease: A Retrospective chart review of a comprehensive Batten clinic

🔬 Safety and efficacy of cerliponase alfa in children with neuronal ceroid lipofuscinosis type 2 (CLN2 disease): an open-label extension study

🔬 REVIEW - Cerliponase alfa and neuronal ceroid lipofuscinosis type 2: long-term outcomes and lessons for future research

🔬 Assessing the integrity of auditory sensory memory processing in CLN3 disease (Juvenile Neuronal Ceroid Lipofuscinosis (Batten disease)): an auditory evoked potential study of the duration-evoked mismatch negativity (MMN)

🔬 RELATED MEDIA ARTICLE - Researchers find possible neuromarker for ‘juvenile-onset’ Batten disease

🔬 Characterization of two human induced pluripotent stem cell lines derived from Batten disease patient fibroblasts harbouring CLN5 mutations

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🔬 CLN3 deficiency leads to neurological and metabolic perturbations during early development

🔬 Mechanisms regulating the intracellular trafficking and release of CLN5 and CTSD

🔬 CLN2 disease resulting from a novel homozygous deep intronic splice variant in TPP1 discovered using long-read sequencing

🔬 Loss of Depalmitoylation Disrupts Homeostatic Plasticity of AMPARs in a Mouse Model of Infantile Neuronal Ceroid Lipofuscinosis.

🔬 Mechanistic Insights into S-Depalmitolyse Activity of Cln5 Protein Linked to Neurodegeneration and Batten Disease: A QM/MM Study

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@bdsraaustralia.org

Batten Disease Support and Research Association Australia is a registered charity.
Australian Registered Body Number: 073 435 202
Australian Business Number (ABN): 56 056 138 99
Our mailing address is: 74 Mclachlan Ave Shelly Beach NSW 226