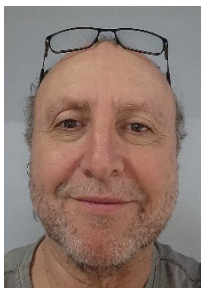
LEUKODYSTROPHY .org.au AUSTRALIA

# newsletter / **spring 2023**



The year is flying and I hope all individuals and families impacted by Leukodystrophy, together with their supporters and stakeholders are safe and well.

It has been 7 months since the passing of the Leukodystrophy Australia founder Sister Julie Thomas CSB OAM. Julie was a great friend and supporter to countless individuals, and I know many of us who would either pick up the phone to call Julie, or visit her, feel the loss of not being able to have a laugh with her, share a thought, or seek Julie's wise words. However, I know we can clearly visualise and hear Julie by just closing our eyes and thinking back to lovely memories.

Sadly, due to other commitments, our wonderful Social Work Mentor and previous Family Advocate, Dr Kylie Agllias has stepped down from the committee. Kylie has been instrumental in supporting our Family Advocate program and contributing great professionalism to the operations of the organisation. We wish Kylie all the very best for her future and thank her for her exceptional contribution to Leukodystrophy Australia.

**Continued on page 2**

## Upcoming Events

| | |
|---|---|
| **Friday 6 October** | **The Charity Challenge — Leukodystrophy Australia Golf Day**<br>Long Reef Golf Club, Anzac Avenue, Collaroy |
| **Saturday 14 October** | **Leukodystrophy Australia Annual General Meeting**<br>Online |
| **Sunday 29 October** | **Family Gathering South Australia**<br>The Watershed Café, Mawson Lakes, Salisbury |
| **24-26 November** | **Safe in Sound Music Festival**<br>Northcote, Victoria |
| **Wednesday 6 December** | **The Charity Challenge – Leukodystrophy Australia Golf Day**<br>Woodlands Golf Club, 109 White St, Mordialloc |

## Contents

## President's Report cont.

We are excited to inform you that our previous Family Advocate of seven years, Anne Patrica, has recently returned to support Leukodystrophy Australia in the capacity of Committee Member. As many of you will know, Anne Patricia has extensive experience and skills in directly supporting individuals and families living with leukodystrophy conditions and the expertise she continues to offer our organisation and committee is invaluable and will greatly enhance and inform our future strategic and operational planning. Anne Patricia will also support our Family Advocate Program as a Social Work mentor to our current Family Advocate Jane O'Neill.

Our Family Advocate program has been operating on a full-time basis since 22 August 2022. During this period, we have been able to significantly increase our contacts with individuals and families living with leukodystrophy and our members across Australia to offer support, advice and assistance. I encourage anyone experiencing any difficulties to please reach out to Jane—we are here to help and provide support in any way we possibly can.

We have had many enquiries around when we will be recommencing our Family Gatherings now that the COVID risks have become somewhat more manageable. We understand that these events are such a valuable means for you to give and receive support and to share and glean information and advice. We are very happy to advise that we have now scheduled a South Australian Family Gathering in Adelaide on 29 October 2023. Jane is in the process of contacting all of our members and families in South Australia to see if you may be interested in attending this.

The Leukodystrophy Australia Annual General Meeting is scheduled for Saturday 14th October @ 2pm via Zoom. We have sent information out to our members regarding this. If you would like to attend our AGM, please RSVP via the email notice or contact our Family Advocate Jane (contact details below).

The Leukodystrophy Australia Sydney Charity Challenge Golf Day is to be held on Friday 6th October at Long Reef Golf Club. For all those interested in participating at this event, please use the following registration link: **https://tiny.cc/sydneygolf** (or you can call me on 0477550977 for more info). For those interested in the equivalent Melbourne Golf Day at Woodlands Golf Club on Wednesday 6th December, please contact me and we will provide the registration details a little closer to the event (the Melbourne event also doubles as a Leukodystrophy member get together/lunch).

Please all take care.

**Earl Schonberger**

## This is Your Space

Do you have a story to tell, or a contribution to make, to Leukodystrophy Australia's Facebook page or Newsletter?

We would love to hear from you!! It is really important that we represent and share the range of experiences and requirements of our members.

If you would like to contribute, please contact our Family Advocate Jane O'Neill on:

**Phone: 0418 790 059** | **Email: advocate@leuko.org.au**

## Family Advocate's Report

Hi everyone, my first twelve months with Leukodystrophy Australia has absolutely flown by!

I feel so grateful to have been able to spend time with and learn from our dear patron and mentor Sr Julie Thomas before she sadly passed away in early February this year. The beautiful messages of love, gratitude and sadness we and Sr Julie's Brigidine friends received from so many people whose lives she enriched were heartwarming and a testament to the very special, selfless and caring person she was. We all continue to miss Sr Julie greatly, and she remains in the forefront of our minds in every aspect of our work to support, assist and advocate for people affected by leukodystrophy conditions.

To those of you I have already been fortunate to meet to introduce myself and check in with you, thank you for welcoming me into your lives, sharing your personal stories and for your warm engagement with me. We have undertaken some amazing teamwork together as we have navigated systems of support and information across Australia and internationally to source the most helpful resources and assistance for you and/or your loved ones.

Thank you to everyone who has provided me with valuable information on resources, support organisations, opportunities and activities within your communities in the spirit of helping and informing all of our Leukodystrophy Australia members and families.

Your strength, courage, sense of hope, resilience, and advocacy on behalf of yourself, your loved ones and other LA members has touched my heart and so enriched my learning and understanding of what it means to be living your lives affected by leukodystrophy.

To those of you I haven't yet been able to connect with, I very much look forward to meeting you during the remainder of this year. In the meantime, please don't hesitate to reach out to me at any time for support, advice or assistance, a debrief or to just say "Hi". I will be very happy to hear from you.

If you have changed your contact details since your last contact with us, please let us know via email, text or a phone call so we are able to stay in touch with you.

**Our Family Advocate Program hours of operation:**
Monday to Friday 9am-5pm (NSW time)

(Hours can be flexible at times to ensure we can meet your needs in different time zones across Australia)

**Phone: 0418 790 059** or
**Email: advocate@leuko.org.au**

# Family Advocate Story

## Supporting siblings of children/young people with a life limiting condition

In the course of my work with Leukodystrophy Australia, I have often been asked for advice on how families can best support the siblings of children and young people with a life limiting condition. I can only appreciate how both very rewarding and challenging it must be as a brother or sister of a child or young person with leukodystrophy.

Siblings will often meet the definition of being a 'young carer', because of the emotional and practical support they are likely to be providing to their unwell sibling and to their family. Siblings may have to grow up faster and spend time helping to care for their sibling with special needs and with household chores.

It's natural for siblings to have many and varied feelings and experiences about having a sister or brother with special needs. These will vary from person to person and at different stages of life.

Some children and young people may be very keen to talk about their feelings and experiences and others may prefer not to talk. Some siblings may not share their thoughts, worries and feelings with their adult family members because they do not want to add to their challenges.

Siblings often have unique bonds with each other, and the illness of a brother or sister may represent the loss of a friend they have shared many life experiences with. Just like adults, siblings will experience a grieving or adjustment process throughout the course of their brother or sister's illness and can show different responses to these.

There can be many positive benefits for siblings as documented in research and literature such as increased empathy, greater tolerance for individual differences, increased confidence, independence and resilience.

Sibling experiences may also involve some negative consequences like feeling isolated, angry or resentful, feeling guilty that they are healthy, experiencing stress at seeing their parents/carers and family members upset, and feeling worried about the future.

Here are some simple suggestions from experts in the area of support for siblings of children with special needs which you may find helpful to consider.

### Encouraging Communication

Some of the most helpful and important ways of supporting siblings include giving them as much information as possible about their brother or sisters' illness, listening to them and helping them express their feelings.

Children can sometimes ask questions that can be challenging for parents to answer but it is important to answer their questions as honestly as possible. Acknowledging the question your child or young person has asked makes them feel heard and provides a safe environment to express their fears or concerns.

Children's understanding of illness and disability varies depending on their age and stage of development. Your child may ask questions like, 'Did I cause it?', 'Will it go away?' or 'Will I catch it?' Answering their questions openly and in a way they can understand at their age can help them to learn about what is happening to their brother or sister and help to reduce feelings of worry and anxiety. When children and young people understand their sibling's condition, it can help them connect and strengthen their relationship.

It can help to talk openly with your child about what it feels like to have a brother or sister with a serious illness. Explain the illness or disability, talking honestly about the effects on the family and the family's responsibilities, and keep them regularly updated with new information.

When children feel listened to, understood, and informed they have a greater capacity to cope with stress and worry. Listen to what they say and help them understand that their feelings are normal. Give them permission to be embarrassed, guilty, angry, resentful, or whatever they are feeling. Once children know that it is okay to experience the feelings they have, you can then talk together about ways they might feel most comfortable to express and manage these feelings. Sometimes it might help to seek professional advice around strategies to help your child or young person to communicate and deal with their feelings.

Adult family members can also give children and young people 'permission' to express their feelings by showing it is ok to share difficult feelings. By continuing to talk with your child about their feelings and your own, you can send the message that it's OK for them to ask questions and to have different types of feelings about their sibling and their situation. Through open discussion, they can learn and understand that they don't need to cope on their own and that it's OK to ask for help when they need to.

### Prepare siblings for the reactions of others

Your family may experience that people stare, tease and ask questions about your child with special needs. You can support your other children by giving them information to help them understand their sibling's condition and some responses they can give others. Planning and modelling appropriate responses with your child and addressing bullying situations if they arise is a great way to help them manage these situations.

**Continued on page 4**

## Family Advocate Story cont.

### Ensure some special time with each of your children

It is easy for a sibling to perceive that your child with special needs is more important and more loved because of the extra attention they require. Try to make some time for each sibling one-on-one eg. sharing a bedtime story or a special activity together outside of the home. It gives you the chance to chat, listen and understand what's happening for them and in their lives outside of home.

We can appreciate how very busy and at times stressful your life can be with all that is involved in caring for a child with special needs. It might help to remember that spending quality time with your other children can happen anytime and anywhere, in the middle of ordinary days and situations and when you have limited time. You may share a game, story or a laugh when you're bathing your toddler or in driving in the car with your child. It's all about giving your child your full attention in these everyday moments which can help them understand just how important and loved they also are.

Acknowledge the achievements and strengths of all children and discuss when and why there may be a need for differences in parenting styles with the sibling and your child with special needs. This will help them understand more about their brother or sister's condition and needs and may help to resolve any sense of unfairness they may be feeling.

Use available respite care opportunities, including offers of help from relatives and friends, to enable you to spend time with your other children, engage in activities with them inside and outside of the home and as important down time for yourself.

### Acknowledge and value the care given by a sibling

When a sibling helps out at home, it is important to acknowledge their contribution. For instance, playing with their brother or sister is a contribution that is often overlooked. Not only does it give parents a break to do other things, it also can provide a fun learning experience for your child with a disability, and an opportunity to strengthen the sibling relationship. This will help all children in your family to feel and understand they are special and valued.

### Encourage independence in a sibling

Siblings sometimes may need help and encouragement to develop goals for themselves and move towards independence. You can do this by supporting your child to have their own space at times and chatting to them about their thoughts, ideas, dreams and needs.

Encouraging activities and interests outside the family can help them establish their own social networks and independence. Some children and young people may also find attending online or local face to face support groups with others in similar situations to themselves can be helpful to understand more about their sibling with special needs and their own feelings and experiences.

At Leukodystrophy Australia we appreciate just how very difficult it can be to balance the challenges of caring for your child with special needs and their siblings, juggling family life, work and your own self-care needs. There is no right or wrong way to manage these challenges and the approach you take will really depend on what feels right for your and your family.

We hope that this article and the resources below can help you appreciate how much you are already doing to support the siblings in your family and perhaps provide you some new ideas to help you manage these life challenges together. Please don't hesitate to reach out to me if you would like further information, advice or to be connected with professionals with expertise in supporting siblings.

### Helpful Resources – information, advice and support for siblings and parents/caregivers

**The Rare Sibling Experience – NORD Webinar**
**https://www.youtube.com/watch?v=Mzk03GiwCOU**

**https://raisingchildren.net.au/disability/family-life/siblings/supporting-siblings**

**https://siblingsaustralia.org.au/services/sibwise/**

**https://www.littledreamers.org.au/programs/**

**Loie's Disease – a children's book about Leukodystrophy (by Maria Kefalas)**
**https://www.amazon.com.au/Loies-Disease-Book-About-Leukodystrophy/dp/1665715677**

---

#### Article Reference Resources


**Carers Australia – Supporting Siblings**
https://www.carersaustralia.com.au/wp-content/uploads/2020/08/Supporting-Siblings.pdf

**Siblings Australia Inc Website**
https://siblingsaustralia.org.au/parents/

**The University of Queensland Faculty of Medicine – Palliative Care of Children Often Involves Treating the Whole Family**
https://medicine.uq.edu.au/article/2017/10/palliative-care-children-often-involves-treating-whole%C2%A0family

**NSW Paediatric Palliative Care Program – Supporting Siblings:**
https://www.nswppcprogramme.com.au/tabid/5472/Default.aspx

**Raising Children Website – Siblings of Children with Disability: How to support them**
https://raisingchildren.net.au/disability/family-life/siblings/supporting-siblings#siblings-of-children-with-disability-feelings-nav-title

**Learning Links – Helping Kids Learn – Information Sheet 22 p10 – written by S Samuel, Family Counsellor**
https://www.ideas.org.au/uploads/resources/699/Siblings%20of%20Children%20with%20special%20needs.pdf

**Rainbow Trust – Full Report – See Us, Hear Us, Notice Us (2018)**
https://www.rainbowtrust.org.uk/uploads/other/pdfs/Full_Report_See_Us_Hear_Us_Notice_Us.pdf



**Jane O'Neill**
**Family Advocate**

# Meet our Families:

## Happy birthday to Tyler who turned 12 in January

Your Disney themed party looked so much fun Tyler and we hope you had a great time celebrating with your family and friends.



## Happy 10th birthday wishes to April

April celebrated her 10th birthday in April with a beautiful unicorn themed party with her friends and family.

We can see how much fun you had at your party April through your big happy smiles.



## Happy 7th birthday to Rylan

Happy birthday to Rylan who celebrated his 7th birthday in June with his family and friends at his Superhero party. Way to go Batman!

You and Tyler and your Superhero Mum's look fabulous.

# Our Work with the Neurological Alliance Australia (NAA)

NEUROLOGICAL ALLIANCE AUSTRALIA

Leukodystrophy Australia is a proud member of the NAA (Neurological Alliance Australia), a collective of eighteen national not-for-profit peak organisations to represent adults and children living with progressive neurological or neuro-muscular conditions in Australia. The Alliance works collaboratively to identify and systemically advocate for opportunities that will promote improved quality of life for people living with these conditions and funding to support research.

As a member of this Alliance, we continue to work on Position Statements, submissions and responses to Government hearings and consultation papers related to issues affecting people living with neurological conditions in Australia.

You can find out more about the important work of the NAA through visiting this website:

**https://neurologicalalliance.org.au/**

# News and Events

## Big “Ride for Nathan” 2023

**Nathan will begin his third Big “Ride for Nathan” at Woorim (Bribie Island) on Monday 18th September and will arrive at Victoria Point on Saturday 23rd September 2023.**

**Follow along with his video updates on Facebook, as he trains around in his local area for his Big “Ride for Nathan” and give him a thumbs up if you see him, or comment for encouragement!**

### Nathan’s story

Nathan is 33 years old, has Cerebral Palsy and was diagnosed with Pelizaeus- Merzbacher disease at the age of 17. Pelizaeus-Merzbacher disease (PMD) is a rare X-linked genetic disorder affecting the central nervous system that is associated with abnormalities of the white matter of the brain and spinal cord. It is one of the leukodystrophies in which disease is due to abnormal development of one or more components (predominantly fats or proteins) that make up the white matter (myelin sheath) of the brain. The myelin sheath is the protective covering of the nerve and nerves cannot function normally without it. In PMD, many areas of the central nervous system may be affected, including the deep portions of the cerebrum (subcortical), cerebellum, brain stem and spinal cord. Signs may include the impaired ability to coordinate movement (ataxia), involuntary muscle spasms (spasticity) that result in slow, stiff movements of the legs, delays in reaching developmental milestones, late onset loss of motor abilities, and progressive deterioration of intellectual function. The neurologic signs of PMD are usually slowly progressive. **https://rarediseases.org/rarediseases/pelizaeus-merzbacher-disease/**

Nathan started walking late in his development, which only lasted a few years until he was confined to a wheelchair for the rest of his life as the Cerebral Palsy took hold. He is able to stand on his legs with the assistance of a walker for a very short period of time, however his leg muscles are very stiff and he’s unable to bend his legs easily. He lives with constant abdominal pain as a side-effect of how he must live with his disease, and everyday tasks are a mammoth effort for Nathan who requires constant support.

In spite of his daily challenges, Nathan is the one of the most charismatic, conversational, and friendly people you’d ever meet. Always charming anybody and everybody with his questions of life and desire to get to know you, and just a bit cheeky with a never-ending smile. Nathan loves to be with people and has time to chat with anyone as if they were a life-long friend. Nathan had a long-time goal to ride a long distance on his hand-cycle, and his desire was to raise funds to inspire research into PMD – Nathan would like to be cured of this insidious disease as anyone would be. Nathan achieved this dream in March 2021 when he rode his hand-cycle from Noosa to Redcliffe on selected cycle trails. Nathan gained local published and radio media coverage and trained with his key support worker Matt. He rode over 1000 km’s in the year leading up to his ride. He totalled 115 KM’s over six days in March 2021 with the help of his dedicated family, wonderful volunteers and the team from Holistic Horizons Support Services. It was truly an unbelievable effort against all odds. We are very proud of him.

In the 12 months before his Noosa to Redcliffe ride, Nathan’s father was diagnosed with cancer, which thankfully he survived. It was decided in view of this that funds raised should go towards Nathan’s future provision with 10% given to Leukodystrophy Australia as there is no research into PMD in Australia, and very little research world-wide.

The day Nathan rode into to Sutton’s beach he said he wanted to do it again. So, preparations began for a ride in 2022. It was much harder this time around even though the distance was shorter, and the ride was delayed. Nathan’s diseases are degenerative – He has had numerous specialist appointments, sickness, infections, a stay in hospital and Covid not to mention the extra weather events and flooding we experienced that year. Yet Nathan was determined & achieved his goal a second time Kingaroy to Redcliffe on selected trails – 85 kms over 4 days.

It was then proposed that the “Ride for Nathan” be run annually, for as long as Nathan can ride, and then continued as a legacy to Nathan for other differing ability participants to continue. Nathan, his family, and the team are especially keen to have greater research into PMD become a reality. Nathan’s immediate need is his own modified van. Nathan finds it ever harder to get in and out of regular vehicles, and the risk of injury to family and support workers is real when helping him into and out of a vehicle. Any funds raised this year would assist with that need.

**https://gofund.me/49f73af5**


## FREE to Leukodystrophy Australia families

Leukodystrophy Australia have been proud to work with Safe in Sound since 2021. Safe in Sound is an exploratory music concert series for persons living with disability and their families/supporters. The Safe in Sound team focuses on creating music that explores sound to stimulate the listener.

Drawing on their experience in sound art and improvisation, they encourage focused listening and enjoyment for people with disabilities, family and carers. Artistic Director Robbie Avenaim said "We believe improvised music can stimulate joy and encourage mindfulness for the listener, and contribute to a well-balanced life through engaging with art. We aim to engage families and carers to both supervise and take part in developing new methods of stimulation, entertainment and communication".

If you would like to find out more about this wonderful program, you can view footage of the concert series at **https://youtu.be/4ryob6zi7GE**

Families interested in participating in 2023, please contact our Family Advocate to arrange your free sessions:

**advocate@leuko.org.au**

Mobile: **0418 790 059**

## Remember the Girls a website for X Linked Carriers

Taylor Kane is the founder and president of Remember the Girls, an international non-profit organization that unites, educates and empowers female carriers of x-linked genetic disorders—a group that is underrepresented and often overlooked by the medical profession **(rememberthegirls.org)**.

**Remember the Girls Podcast**

## The Nathan Centre

Monique Garcia is the Founder and Director of a wonderful organisation called The Nathan Centre. The organisation aims to support families managing rare disease. Monique has created three Facebook groups that could be of interest to your members:

### Rare Disease Kids Australia

**https://www.facebook.com/groups/332689378281950/about**

### Rare Disease Palliative Care Australia

**https://www.facebook.com/groups/739138833866072**

### Rare Disease Angels Australia

**https://www.facebook.com/groups/711530736855502**

**http://www.thenathancentre.com/**

## Continuation of Victorian Public Transport Concessions for Carer Card Holders

Carers Victoria has announced that after their ongoing advocacy efforts, the Victorian Government has now confirmed it will continue current access to public transport concessions for Carer Card holders.

**Well done Carers Victoria!**

You can find out more about transport concessions for Carers in Victoria and how to apply for a Carer Card through visiting this website:

**Public transport travel discounts**

**Carer Card Program Victoria**

# SNUG Retreats – Family Action Centre, University of Newcastle

The SNUG project provides residential retreats for families caring for a child with a rare health condition. The whole family attends the retreat together and a program of activities is designed so that EVERY attendee can participate. All accommodation, meals and activities are provided. If you care for a child with a rare condition, or you work with a family who might be interested in these special retreats, please submit an expression of interest through visiting the home page for Snug Retreats: **https://www.newcastle.edu.au/research/centre/fac/outreach/snug**

SNUG is coordinated through the Family Action Centre at The University of Newcastle and is supported by the **Steve Waugh Foundation** whose goal is to be 'somewhere to turn' for families of children with rare diseases.

The retreats aim to create a relaxed environment for families to build on their strengths, improve resilience, and develop friendships and support networks with others who are experiencing similar challenges.

The retreats:

- Focus on identifying and celebrating the skills and strengths of ALL family members – parents, children with special needs, and their siblings.
- Provide a wide range of activities over 4-5 days which are designed so that all can participate – including music therapy, art therapy, physical activity and outdoor recreation, craft, sensory play, and parent discussion.
- Are supported by a team of trained volunteers.
- Educate, train and support our volunteers (mainly undergraduate students of the University of Newcastle) in understanding the unique issues faced by families caring for children with rare medical conditions through our volunteer program.
- Are held approximately 6 times every year at Sport and Recreation Centre facilities in Lake Macquarie, NSW.
- Are the only retreats of this kind in Australia.

SNUG Retreats Connecting Families

STEVE WAUGH FOUNDATION

Australian Government

Carer Gateway

# Carer Gateway Support

## About the Australian Government Carer Gateway

Do you provide care or support for someone close to you?

Across Australia there are more than 2.65 million unpaid carers, that's 1 in 9 people!

Unpaid carers look after someone who lives with disability, a medical condition, has a mental health condition or is frail due to age.

You can become a carer at any time. Many people don't consider themselves a carer. They are simply children, parents, partners, relatives, or friends supporting someone close to them.

A carer might look after someone 24 hours a day and help with daily activities. Or, they might look after someone who only needs a little bit of help.

Responsibility, care, compassion, and love are all reasons why someone will step forward to provide unpaid care. Most unpaid carers primarily help with tasks like shopping, housework, cooking, home maintenance and providing transportation.

Carer Gateway is here to support you. Carer Gateway was designed by carers for carers.

We offer a national approach to providing reliable services, support and advice to carers in Australia.

Services available include counselling, peer support, coaching, skills courses, tailored support packages (including planned respite), access to emergency respite, information and practical advice.

Call Carer Gateway on **1800 422 737**
Monday to Friday, 8am to 5pm
or go to **CarerGateway.gov.au**
to find out what we can do for you.

# Changes to medical prescriptions

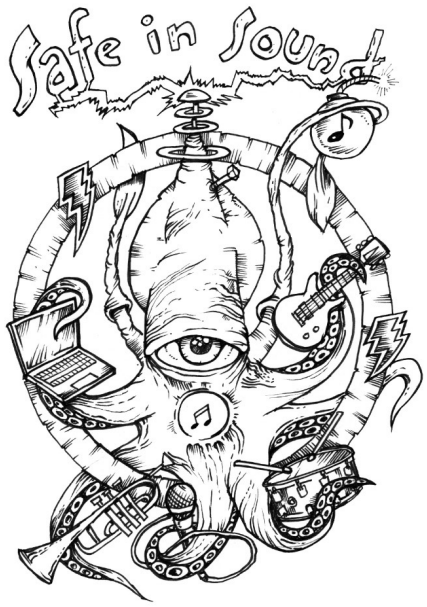
There has been much misinformation about the changes to medical prescriptions taking place in 2023.

Consumers' Health Forum has produced an information package which you might like to distribute to your family, friends, clients, patients and others.

**https://www.60dayscripts.com.au/**

# Safe in Sound Music Festival

## 24-26 November 2023
## Northcote Victoria




Legendary WHATISMUSIC? organiser Robbie Avenaim returns to refresh and reshape the frontiers of Australian sound art performance with his new music festival effort: Safe In Sound! Avenaim's ability to conjure up the most profoundly uncomfortable, and uncomfortably profound, often wildly unhinged live experiences has created countless mythical moments still discussed decades later in the Aussie underground - and he now turns his unparalleled creative gaze to a new, relatively untapped, reserve of genius potentialities. For over five years, the Safe in Sound program has made experimental music accessible to people with disability via in-home concerts. Now, several of those participant-performers are eager to share their unbridled creativity, freedom and joy with the wider world.

Across November 24-26 JOLT Arts **(jolted.art)** Northcote will host three days of truly liberated sonic generation, featuring these relatively new improvisers paired with some of Australia's greatest musical explorers - as well as divergent dancers, iconoclastic kids and other uniquely skilled audial creators with disability from throughout the local community.

The festival will culminate in a symposium featuring performers of all experience levels and the curators openly discussing and detailing the benefits and broken boundaries of this work.

The Safe In Sound festival promises to help reinvigorate the Australian avant-garde at large, shaking loose many of the unspoken limitations on a music that takes pride in calling itself free and providing all new mythical musical moments for aeons to come.

# Manly Adolescent and Young Adults Hospice

The AYAH is a Public NSW Health Facility and is the first of its kind in Australia, offering respite care, symptom management and end of life care to adolescents and young adults aged 16-30 years with life limiting illness. The upper age has just been increased from 24 to 30 years which is wonderful news!

This specialised care extends to patients' families, including bereavement support, counselling and accommodation.

Having opened their doors to patients in February 2023, the AYAH have already had the opportunity to see the positive impact their service has on families who are going through very difficult times.

AYAH's holistic approach ensures the patients clinical needs are met, while also ensuring their stay is as enjoyable as possible within their physical and cognitive abilities.

You can find out more about the Hospice through reviewing the following links or through contacting the hospice directly via email:

Email address:
**NSLHD-AYAHMANLY@health.nsw.gov.au**

Facebook:
**www.facebook.com/ayahmanly**

Instagram:
**Instagram.com/ayahmanly**

Website address:
**www.nslhd.health.nsw.gov.au/manly/Pages/default.aspx**

# Have you forgotten to initiate or renew your LA membership?

Members are the essence of our organisation. Leukodystrophy Australia works with its members to achieve goals that benefit all people living with Leukodystrophy, with a specific focus on individuals and families living their best lives.

Our membership numbers are so important to enable us to raise awareness of leukodystrophy, apply for grants and make policy submissions, and in keeping our organization relevant.

Membership for people who are directly affected by leukodystrophy conditions is **free**.

We would be very grateful if you could please initiate or renew your membership for **2023-24** today.

Applications can be found on our website:
**https://www.leuko.org.au/support-us/membership/**

Or contact our Family Advocate Jane directly and she will forward it on to you.

Thank you

# Research update

## Research for newborn screening

### Information from Pathology Queensland and University of NSW – newborn screening for leukodystrophies

**Our research**

Pathology Queensland and the UNSW are researching a genomic newborn screening test that identifies over 100 serious genetic conditions with treatment, including 5 leukodystrophies (X-Linked Adrenoleukodystrophy, Krabbe Disease, Metachromatic Leukodystrophy, Fucosidosis, Cerebrotendinous Xanthomatosis). When treatment for other conditions is developed they can be added to the test.

This year 10,000 newborns will be screened, their parents will be interviewed about their experience of the test and the cost effectiveness of the testing will be calculated to see if this new test could be made available as part of newborn screening for all Australians.

---

## Pelizaeus-Merzbacher Disease (PMD) natural history study – Ionis Pharmaceuticals Inc.

The Royal Children's Hospital and the Murdoch Children's Research Institute in Melbourne is currently recruiting patients with PMD to participate in a natural history study which commenced in August 2023. This study may help scientists and researchers learn more about PMD to help develop treatments for this disease.

### Your child or a loved one may be able to participate if they:

- have classic PMD
- are 6 months to 10 years old
- has had genetic testing and has a duplication in the PLP1 gene*

*If you are unsure of the mutation type, please contact your doctor

### What is involved?

- Participants visit the study doctor up to 3 times over 2 years for the study
- Study participants receive at no cost:
  - Study health assessments
  - Study support and monitoring by a healthcare team
  - The opportunity to help advance PMD research

If you would like more information or would like to be involved, please contact the study coordinator **Eloise Uebergang** on **(03) 8341 6382** or **Eloise.uebergang@mcri.edu.au**

## Women with X-ALD

### Opportunity to participate in an international online study

Adrenoleukodystrophy (ALD) is a rare genetic defect that mostly affects men due to the X-linked inheritance pattern. It is now known that women with ALD often develop neurological symptoms that are comparable in severity to those of men. The frequency of symptoms increases with age.

The **Leukodystrophy Outpatient Clinic at the University of Leipzig, Germany** is currently conducting an online study asking women with ALD worldwide about their quality of life. They would like to find out how many women with ALD develop symptoms over the course of their lives and how these affect their physical and psychological well-being.

This study will investigate how common symptoms are in women with ALD and how symptoms affect women's quality of life.

Participation in the study involves filling out questionnaires about your symptoms and how they affect your psychological well-being and quality of life in different areas. The questionnaires will be made available to you electronically on the online platform Leuconnect **(https://www.leuconnect.com/en-GB)**. Participation is possible in English, German, French, Italian and Spanish!

You can find more information about the study under the following link: **https://www.leuconnect.com/en-GB/Study/Card/3**

With your participation, you will be making an important contribution to research into ALD, particularly in affected women, and to the development of new treatment approaches tailored to the needs of women with ALD.

If you have any questions, please feel free to contact Dr Lisa Schäfer: **lisa.schaefer@medizin.uni-leipzig.de**

**Universitätsklinikum Leipzig**
**Dr. Dipl.-Psych. Lisa Schäfer**
**Leukodystrophie-Ambulanz**
**(Leitung: Dr. med. Wolfgang Köhler)**
**Klinik und Poliklinik für Neurologie**

# Research Study
# Murdoch Children's Research Institute & University of Melbourne

## Investigating Parent's Service Experiences of Genomic Testing

This study aims to harness the voice of parents to shape the delivery of complex genomic testing outside of genetic services. Project Co-ordinator **Erin Crellin** **erin.crellin@mcri.edu.au** is in the process of recruiting parents from across Australia to participate in online focus groups (1-1 interviews are also available if preferred).

Any parent whose child has had genomic testing (exome or genome sequencing) for a rare genetic condition in the past five years is eligible to participate.

**Has your child had genomic testing in the last 5 years for a genetic condition (excluding cancer)?**

We are keen to hear from parents with different service experiences.

For example, receiving genomic testing for your child from a

**genetic doctor or counsellor**

**general paediatrician or specialist**

**Your views will shape how genomic testing is delivered outside genetic services, a change which aims to help more families access accurate diagnoses more quickly.**

Contact Erin Crellin (crelline@student.unimelb.edu.au) if you are interested or scan the QR code to learn more.