



**Summer Newsletter 2018**



**Kellie Panton, President**

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Once again, Christmas is upon us and another year almost at an end. It has been a year that for many brought loss and our hearts go out to you all.

As President, there has been many highlights - the family gathering in Adelaide at The Watershed on the Wetlands, and the Charity Challenge Golf day in Melbourne, where I had the opportunity to attend both these wonderful events. I also attended the Charity Challenge Gala Ball & Silent Auction in Sydney and really enjoyed meeting family members I would not normally have the chance to do. My son came with me, and spending the weekend together is always a great thing to do. The generosity of people wanting to donate and bid in the Auction is always amazing.

I would like to wish all our families, supporters and committee members a wonderful and safe Christmas and hope to connect with you more next year.



**Bronwyn Byrne Office Manager**

**Toll free 1800 141 400 or 0418 755 994**

**Office reopens 7 January 2019**

**messages will be checked**

The close of 2018 has been dizzying in the many events and changes that have taken place by some incredible people, working exceedingly hard across multiple facets of life in the world of medicine and wellbeing. Now, here we land almost at Christmas, most I am sure looking forward to a slowing of pace before we all start over. Please find some time!

People caring for a family member or friend with a rare disease are some of the most remarkable people, as are the invaluable Volunteers and Committee Members of our organisation, all displaying persistence and resilience. Not always easy! My job gratefully allows me to interact with them and others such as the valuable connections we have fostered with Australian Genomics Health Alliance, Melbourne Genomics, Genetic Services Network Victoria, Mission Massimo Foundation, Neurological Alliance Australia, and Rare Voices Australia. Thanks to the tenacity and talent of these people, we have seen breakthroughs in the recognition of Government toward Rare Disease and Genomics, translating into large funding allocations in the health sector that will make a difference.

2019 can only get better and I look forward to working in this shifting environment with you, our families and friends. Our core activities of advocacy, family support and family gatherings, grant writing, supporting events and research will continue and a new website! Wishing you peace and happiness at Christmas and into the New Year, Bronwyn



*SA Family Gathering 2018 thanks to Edwards family, Greyhound Racing Australia; and Shirdi Sai Santhan Melbourne, who raised \$10K for LEUKOdystrophy Australia families. BOTH OUTSTANDING EVENTS!*



On 15 December 2018, the first anniversary of Massimo’s passing, the Federal Government funding for Massimo’s Mission was announced. \$3M over 3 years!

The funding will go towards closing the gap between genetic diagnoses and clinical treatments for Leukodystrophies, and other rare childhood brain disorders.

“It will allow researchers to understand disease mechanisms faster and test potential therapies in potentially life-changing time frames.” **Greg Hunt MP - Minister for Health.**



This announcement is a touching tribute to honour Massimo’s legacy and will support pioneering Australian research to finish the mission he started. Leukodystrophy Australia thanks Mission Massimo Foundation and all the researchers, medicos, donors and believers in this long haul to another epic journey, that of treatment and the ultimate, a cure.

## Melbourne Genomics Health Alliance

*Global knowledge. Individual care.*

Recently, Leukodystrophy Australia attended a fantastic workshop where there were about 40 attendees, along with guest speakers from many facets of health care. The topic - **Understanding Genomics: Empowering Patient Advocates.**

Should you be interested in genomics, on the Melbourne Genomics website, you can now find the “Introducing Genomics” presentation by Fran Maher as a pdf file, with notes for each page viewed from the icon at top left of screen. <https://www.melbournegenomics.org.au/patients/community-resources>

We also have links to podcasts and radio interviews. Please contact the Leukodystrophy Australia office for details.



*Did you know, you could call any genetic services network and speak with a genetic counsellor? Maybe not straight away, but they will call you back.*

- Six Things Everyone Should Know About Genetics  
[www.ashg.org/education/everyone\\_1.shtml](http://www.ashg.org/education/everyone_1.shtml)  
This American Society of Human Genetics website provides basic genetics information and resources.
- Garvan Institute resources  
[www.garvan.org.au/research/kinghorn-centre-for-clinicalgenomics/learn-about-genomics/dna-base/dna-and-disease](http://www.garvan.org.au/research/kinghorn-centre-for-clinicalgenomics/learn-about-genomics/dna-base/dna-and-disease)

The Centre for Genetics Education have a new fact sheet on Reproductive Carrier Screening - <http://www.genetics.edu.au/publications-and-resources/facts-sheets/FS65REPRODUCTIVECARRIERSCREENING>



## FAIR FOR RARE

A FAIR GO FOR AUSTRALIANS  
LIVING WITH RARE DISEASE

Rare Voices Australia's (RVA) 2018 National 'Fair for Rare' Summit was held on 16 and 17 November in Melbourne, and Leukodystrophy Australia attended, amongst many other stakeholders working together towards a common purpose to reflections on the fostering of true connection over the course of the two days. Delegates engaged in 46 sessions between the presentations, Q&A panels and workshops. The buzz and energy that drove the Summit, as well as the common vision shared by delegates to contribute towards effective patient policy that transforms lives, was truly remarkable and to be celebrated.

RVA has been leading the call for a **National Rare Disease Framework** since the organisation’s inception in 2012. A timely and welcome announcement by Minister Hunt who opened the Summit was the government’s commitment to a National Rare Disease Framework, the development of which will be led by RVA. It was a perfect opening for the Summit and a truly amazing and momentous policy decision. The Summit also provided a wonderful opportunity to start this Framework consultation and collaboration. The themes of the Summit included the 6 Strategic Priorities, but also incorporated a focus on mental health/wellness, the youth perspective and supporting the rare disease workforce. You can view the 2018 Summit Report by clicking <http://www.rarevoices.org.au/news/3209/2018-national-rare-disease-summit-report>



The theme for Rare Disease Day 2019 is '**Bridging health and social care**'.

For most people living with a rare disease, as well as their family members or carers, the reality of daily life can include any combination of the following: collecting and taking medicines, attending appointments, participating in physical therapy, using specialist equipment and accessing various social and community support services and respite care. Managing these care-related tasks alongside their usual daily activities such as work, school and leisure time can be challenging.

Rare Disease Day 2019 is an opportunity to be part of a global call on policy makers, healthcare professionals, and care services, to better co-ordinate all aspects, of care for people living with a rare disease.

Due to its success in 2018, the [#ShowYourRare](#) social media campaign continues for 2019. People around the world will paint their faces and show their rare for **Rare Disease Day – Thursday 28 February 2019**. **Get involved for Leukodystrophy** <https://www.rare diseaseday.org/> or email [info@leuko.org.au](mailto:info@leuko.org.au)



The parents and individuals who share their stories help us feel less alone on this journey, if you've got a story to share, please get in touch we have a whole country of people who would love to hear your story!



Hi! This is Ethyn and he is 12, turning the big 13 in June. He has a little sister, Charlee, who is 9 and loves him to bits. We live in a little town called Penola, South Australia. I am a carrier (Stacey) and have the Cerebral AMN so am on disability pension. My beautiful husband is my Carer.

Ethyn has Cerebral Adrenoleukodystrophy, a terminal disease and Addison's disease that is treated with tablets and is deadly. We have the best Neurological team and medical staff working with us. They truly are amazing. At the age of 8 his ALD became symptomatic. Luckily, for us there was a treatment at this very same time in Boston, Massachusetts USA, and we were invited to take this rare opportunity of Gene Therapy.

Our community friends and family rallied together to help us as a family get to Boston, stay for nearly 6 months, as well as keep our home and bills up to date. It was a full on experience. Ethyn was a trooper. Through countless needles, 5 rounds of Chemo, side effects, sleepless nights, tears and nightmares and so much more, we all kept smiling and supporting each other.

It is hard, to really put this story into words for anyone else to get the full impact. But what really matters is our son is still alive and thriving and our family unit is stronger than ever.

Ethyn loves playing basketball. He and his team won the grand final last year. We never thought he could ever achieve such a thing in the early days. Basketball shoes can be expensive and sometime we need a bit of help. Via a grant to Leukodystrophy Australia, Coopers Brewery Foundation stepped in and gave us money to buy his shoes this year! Also very grateful to them for paying to send our boy on school camp. As you can imagine it is still very hard to let go of the apron strings but it's so important Ethyn learns to cope away from us too. He had a ball!! We cannot thank Coopers Brewery Foundation and Leukodystrophy Australia enough for the on-going support.



*Thank you again from all of us and remember to Keep Shining 🌟🌟🌟🌟🌟*



**Family Advocate**

0418 790 059

Email: [advocate@leuko.org.au](mailto:advocate@leuko.org.au)

Hello everyone, what a joy it was to meet so many new and current clients at the Charity Challenge Ball this year. It was a night, which allowed all of us to have fun, get to know each other and listen to wonderful stories of love and hope. This fundraising event is stupendous; those such as our committee member Earl Schonberger who put their time and effort into the event are gems. I remain in awe of the generosity of considerate donors. I hope I will have the opportunity to attend next year and meet other wonderful people.

The end of 2018 has snuck up on me very quickly. Throughout 2018, I have again been very blessed to once again share many family stories and walk beside those who wish to have a chat, and those who required support, advocacy and assistance. Thank you to all the wonderful families I have worked alongside, to Bronwyn who continually works closely with me assisting in creating a service where families are welcomed and supported, to a supportive committee and to all others who make my role pleasurable.

I have been fortunate to be involved with **Robbie Avenaim's** project **'Safe in Sound'** and attend outreach visits to families in NSW with Robbie and his colleague **Jim Denley**. The sessions are already, well received by several of our families in NSW and Victoria. I loved observing and listening to families producing fabulous chaotic sounds with diverse instruments, all assisted by Robbie and his staff. Families 'letting go' and being 'in the moment' having fun and enjoying themselves was a wonderful space to sit in. During the debriefing sessions everyone had a smile on their faces and I walked away thinking what an awesome atmosphere I had been privileged to witness. I am looking forward to outreach visits with Robbie, his team and the families in 2019. My hope is Robbie may secure more funding in the future, so we may offer this wonderful experience to our families nationwide.



Christmas has varying meanings for all and whilst it brings joy, happiness and festivities it may bring its challenges. May you feel a sense of peace, find a quiet space for reflection and surround yourself with those who love and support you. While my normal availability will be reduced while the LA office is closed, I am still around if you wish to have a chat.

Whatever the Christmas Season means to you religious or otherwise, please stay safe and embrace what the World has to offer in the form of hope and love. The Christmas period for me means spending quality time with my family in a quiet loving space, attending midnight Mass at our beautiful Newcastle Cathedral and paying visits to residents in a nearby Residential Care facility.



Love and respect, Anne Patricia.

**I will be on annual leave from 2 January 2019 and will return on 9 January 2019**

My usual working hours are Tuesdays and Wednesdays 8.30am to 5pm.



LA offers our condolences to all those in the Leukodystrophy community who have been touched by bereavement this year, all missed so dearly. Love and prayers to you.

- Michael Quattrone 15 June 2018
- Samuel Moorhouse 28 June 2018
- Jackson Daff 1 December 2018

❤️ Many donations were received throughout the year – Thank you!

Donations of \$1000 or more were also gratefully received from -

Anonymous, All Souls' Opportunity Shop, Energy Consult, Greyhound Racing South Australia, Hala Raghieb, Hughes O'Dea Corredig, Insurance Advisernet Foundation, Jardine Lloyd Thompson, J & T Clancy, Lions Club of Geelong Corio Bay, Measaval Pty Ltd, Shirdi Sai Sansthan Melbourne, The Charity Challenge.

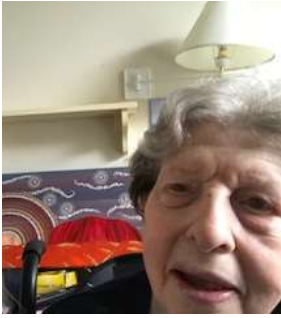


The Charity Challenge Leukodystrophy Australia Golf Day 2018 @ Woodlands was such a great day



The Long Reef Golf Day <https://drive.google.com/open?id=1dLG6B0ocr4jDBKuat0odI9szwqOi87M->

The video made for The Charity Ball [https://drive.google.com/open?id=1H5M7EukLoPEvfqgx4frh8WXG7pfW\\_2i](https://drive.google.com/open?id=1H5M7EukLoPEvfqgx4frh8WXG7pfW_2i)

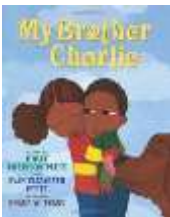


Greetings to all. I just want to say, I do hope you are all feeling as well as possible. I am very aware that for many, this year could have been a little kinder, but I do think many show much courage and cheerfulness.

May those who are having a “holiday” or not, have a happy, quiet time, filled with much love.

Most of us know the wonderful work our team does. A sincere thank you for so much generosity. May 2019 fill with good things for all.

Peace, joy and happiness, Julie csb OAM



**Books for siblings of children with disabilities:** Siblings of children with special needs are amazing human beings. Their lives are often very different to that of their friends and they may sometimes need a bit of help while navigating their journey. Here are some great books, which provide tips and advice, share the stories of others going through similar things and tales to help them understand their sibling better. <https://www.publishersweekly.com/978-0-545-09466-5>



**Books for all children 4 – 7: Willow Willpower** has big dreams. Willow inspires children of all ages to dream big, all while whisking them around the globe. Discover some of the most famous places in the world alongside Willow as she takes you high in the sky. [@willowwillpower](https://twitter.com/willowwillpower)



Leukodystrophy Australia wishes to give a big thank you to our volunteers Lucas Mitchell, Amy Ruscigno, Jacklyn Vick, Tracy Lyons, Julie Merrett and Leonie Allen who have been integral to our core business in assisting the Office Manager on a number of tasks both one-off and ongoing. Such a pleasure having you on board.



A big thank you to Genetic Services Network Victoria (GSNV) for your outstanding support in your Supporting The Support Groups program. We have made some wonderful connections, sharing common goals, pooling resources, and importantly, we really do feel supported. We are looking forward to collaborating next year in your goals.



Thank you MS, Colin Crumb, Senior Facilities Manager for their continued support in office space, technical support and priceless camaraderie of staff, including Andrew Giles MS Australia and the Neurological Alliance Australia.



Thank you to Tonic The Agency – John Epta - for donating our Locus Database and the ongoing support with Ewan Duckworth [www.tonic.com.au](http://www.tonic.com.au) You have really made a huge difference to us.



For information on Leukodystrophy Australia [www.leuko.org.au](http://www.leuko.org.au), events, volunteering, joining Committee, please contact Office Manager, Bronwyn Byrne 0418 755 994, or email [info@leuko.org.au](mailto:info@leuko.org.au)