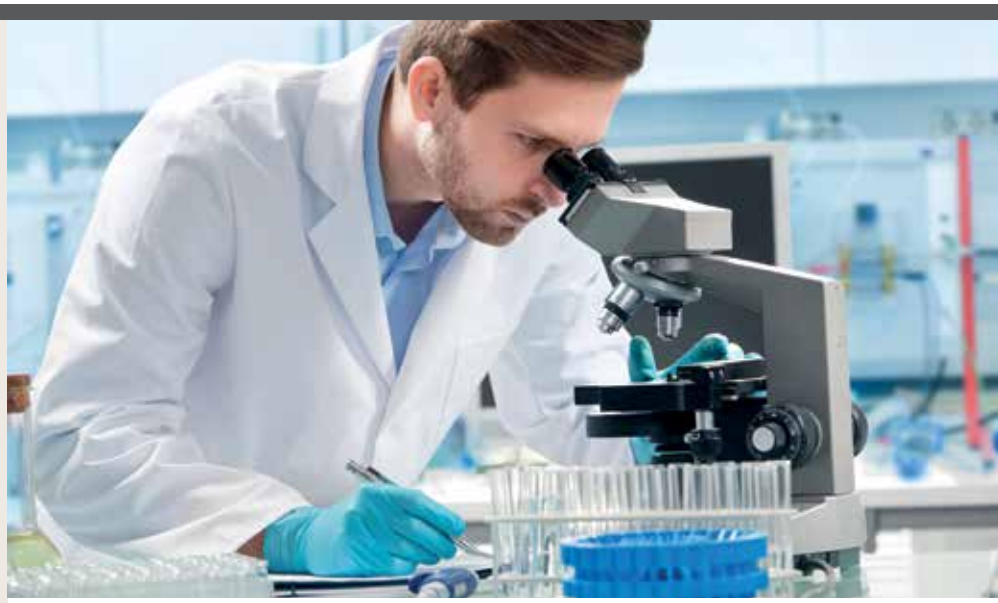
genetic support network of victoria

AUTUMN-WINTER 2016

empowering * connecting * supporting

CONTENTS



The involvement of patient organisations in rare disease research: an Australian study

A recent study has examined the role of Australian patient organisations in rare disease research.

By Ms. Deirdre Pinto, Dr. Dominique Martin and Dr. Richard Chenhall

Deirdre Pinto conducted the study as part of her Master of Philosophy (Population Health). Together with her academic supervisors, Dr. Dominique Martin and Dr. Richard Chenhall at The University of Melbourne's School of Population and Global Health, Deirdre has published a paper in the Orphanet Journal of Rare Diseases. The authors highlight patient organisations' valuable contributions while also discussing the difficulties their leaders face when trying to support research.

Organisations led by patients and/or their families are understood to be playing increasingly important roles in rare disease research. However, despite examples of rare disease patient organisations (RDPOs) that have facilitated significant advances in understanding and treating specific diseases, there is little information about the research-related goals, activities and experiences of RDPOs generally. In this study, we examined how Australian RDPOs are involved in research. Organisations eligible for the study were non-profit, non-government groups with a legal status (such as incorporation) in Australia, and which focus on a disease or closely related set of diseases with a community prevalence of 1 in 2000 or less.

Using a series of online searches, 117 Australian RDPOs were identified. Ms. Pinto analysed 112 RDPO websites, pilot tested a questionnaire with the leaders of three groups, and invited the remaining 114 RDPOs to participate in an online survey. 61 RDPO leaders completed the survey, giving a response rate of 53%. Ten RDPO leaders and two key informants participated in face-to-face interviews with Ms. Pinto. More than 90% of the surveyed RDPOs had

Cont. page 7

GSNV Murdoch Childrens Research Institute, Royal Children's Hospital, Flemington Road, Parkville, VIC 3052

P (03) 8341 6315 | **E** info@gsnv.org.au

www.gsnv.org.au

genetic support
network of victoria

Committee of Management

President	Monica Ferrie
Vice President	Maree Maxfield
Treasurer	Rachel Pope-Couston
Secretary	Anna Jarmolowicz

General Committee Members
Abbie Kinniburgh
Christine Williams
Catherine Beard
Emily Higgs
Bill Ellerton

Committee Meeting Dates

Tuesday 12 July
Tuesday 9 August
Tuesday 13 September
Tuesday 11 October
Tuesday 8 November
Tuesday 13 December

The GSNV quarterly newsletter is published by The Genetic Support Network of Victoria Inc. The opinions expressed are not necessarily those of the GSNV staff or committee. The views and opinions expressed in this newsletter are those of the individual authors. The material in this newsletter is for information purposes and does not constitute medical advice. GSNV articles maybe sourced and reproduced by support groups and other forums, but the GSNV requests written advice beforehand and written acknowledgement of the source is provided, once permission is given.

GSNV

Message from the Committee

I am thrilled to be writing to you as the new President of the GSNV – you can check out my profile on our website. I want to talk about what's happening and going to happen over the next 12 months.

It's an exciting time in genetics, from every angle and on every level something is happening. At a state level we are awaiting the release of the Melbourne Genomics Alliance discussion paper on a new genomics framework and a Victorian Government broad review of governance paper. The release of these papers will inform the development of the new GSNV strategic plan for 2016-2018.

We will also be in the fortunate position to utilise all the international intelligence and information gathered by our Group Leader, Louisa Di Pietro while in Italy, including the conferences and meetings she has been involved with. As a Committee, we are committed to the GSNV vision of Purpose, Dignity and Choice for people with genetic conditions. Our focus will be on serving the community – people who are impacted by genetic and rare conditions and those who serve them – support groups and clinicians.

The Committee will work closely over the coming year with our key stakeholders including the Department of Health and Human Services, to ensure that our energies are in synergy with Victorian and National policies and funding priorities, and the needs of our stakeholders and the broader community. We also intend to work hard to secure additional funding sources.

The AGM delivered a significant change to the GSNV Committee going forward and we are excited about bringing a different range of skills and experience to the GSNV Committee of Management in the year ahead. I welcome those who have joined us. New profiles can be viewed on the GSNV website.

I would also like to sincerely thank the outgoing Committee members who have served the GSNV with dedication and commitment.

Through this newsletter, the Committee will provide updates that include a review of our meetings, confirmation of areas of focus, how we are travelling against our strategic and business objectives, as well as keeping you informed about what's happening in the world of genetics and how it is impacting. We are looking forward to engaging with you over the year.

Monica Ferrie,
President of GSNV

GSNV MEMBERSHIP

Anyone can become a member of the GSNV, and we'd love to have you on board. Our current members include support groups, individuals and families affected by genetic conditions, health professionals, students, and members of the community.

Membership entitles you to:

- Newsletter and eNews
- Genetic support
- Peer connection
- Information and education
- Free Peer Support Training
- Advocacy on behalf of members

More information: **gsnv.org.au/become-a-memberdonate.aspx**

Your Feedback

I received the Summer edition of the newsletter this morning, great read!

– Sarah Burns, Project Coordinator, Programs & Support Services, Cystic Fibrosis Victoria.

GSNV

Message from the team

Dear members and the genetics community, This is the last time I will be writing to you from my desk abroad, and its truly hard to believe that over a year has gone by. Living and working overseas has been wonderful, and as I reflect on my time here in beautiful Italy, it's true to say, there is no place like home.

I have come to appreciate the Australian way of doing things, and although ours is not a perfect society, nor does it have the perfect health system and bureaucracy, it's definitely not too shabby compared to a lot of what I have seen around me. Over the year I have reported my personal journey in seeking treatment and the hoops that I have had to go through in doing so. I've decided that no matter how beautiful the country, the history, the food and the lifestyle of a particular place, your quality of life is undermined if you can't get what you need readily. In some ways, I've come closer to understanding how the rare diseases community feel and the frustration and isolation that they experience in trying to secure basic services and support. When it comes to health care and your quality of life on a daily basis in living with a genetic and/or rare condition, no one should be made to feel like an alien on their own planet.

This was all on my mind, as I headed off to the European Meeting on the Psychosocial Aspects of Genetics (EMPAG) and the European Society of Human Genetics (ESHG) meetings in Barcelona in May. In particular, was thinking about where we are up to globally with, and how are we moving forward in changing lives with genetic and genomic medicine. Overall, I think I can say it's all happening slowly. Research translation is still the frustration of the genetics community worldwide (as it's just not happening fast enough) and developing best clinical practices in genomics is still very much an academic exercise. There is some good evidence of what can be achieved, but it's very much a case of good collaboration and research rigour. The European Human Genetics Conference (now in its 49th year) is a forum for all professionals in human and medical genetics to review advances and develop research collaborations. The conference has become one of the premier events in the field of human genetics with over 3,000 delegates, more than 215 oral presentations, 18 workshops, 8 educational sessions, and over 150 exhibiting companies. The ESHG conference is where the latest developments in human genetics are discussed, and where professionals from all parts of human genetics meet. 2016 marked another joint congress with the European Meeting on Psychosocial Aspects of Genetics. The European Society of Human Genetics promotes research in basic and applied human and medical genetics and facilitates contact between all persons who share these aims. As a seasoned EMPAG/ESHG delegate I am pleased to report that this year was an outstanding meeting and the organisation, ambiance and professionalism in Barcelona notable. Withstanding the practical elements of the conference that were all rated highly, the conference content this year was very much about genomics and the use of new technologies and how they may impact clinical and research outcomes. In the EMPAG conference, which is where I spent most of my time, it was terrific to see the work of the Melbourne Genomics Health Alliance (MGHA) flagship projects profiled on a number of occasions, and received very favourably by the international audience. The work of the MGHA is highly regarded and has placed Victoria (and indeed Australia) squarely in the top ranked genomics hubs around the world.

EMPAG is always interesting as the sessions get to the very heart of genetics and genomics medicine, and the issues we need to be thinking about from a reflective practice and patient point of view. On the first day of the conference (Sat May 21) a joint ESHG and EMPAG session explored genetic privacy and data sharing and got right down to the ethical issues around privacy. 'The hitchhikers guide to data sharing' was an interesting session looking at the era of ubiquitous genetic information for research, clinical care and personal curiosity, but referenced all that against the need to protect the genetic privacy of the data originators. The presentation provided an interesting 'technical map' outlining potential threats to genetic privacy. The presenter took a different approach to the standard privacy versus utility argument and hypothesised trust-enabling techniques designed to create solid partnerships between researchers and participants. On day two (Sun May 22) of EMPAG, the educational session 'Direct to Consumer (DTC) Testing: Empowering patients, caring for consumers' looked at the impact of DTC testing on genetic healthcare delivery and the shift toward a more empowered patient, taking charge of their own health. The presentation explored the challenges of DTC testing and the mixed reactions and 'concerns' expressed by medical professionals and researchers around DTC testing. The underlying theme of this presentation was that, it is the public's right to access personal genetic information, when it is delivered in a responsible manner, and this can be supported by health professionals. Key principles of DTC testing delivered by private companies, is that if there is respect for consumer rights and privacy, if reports are written at an appropriate and comprehensible level, if there is educational content to support consumers, using valid test methods, and there is compliance with local regulation and policy, DTC testing and research can support the consumers ability to comprehend their personal genetic information without evidence of psychological harm. I think the general opinion of the room was that we still have a long way to go in providing this in the private domain and in general. Physicians are currently not prepared to deal with DTC test results presented by their patients. This lack or preparedness may have a negative impact on the patient - clinician dynamic. I think an evidence-based assessment of this is important and although DTC testing companies are working hard to engage and educate health professionals about DTC testing, it's still a rocky road of acceptance.

The following session on patient 'empowerment' and DTC testing looked at the use of this buzzword to support the idea that consumers should have direct access to genetic risk information without the involvement of health professionals. The session explored three theories on the concept of empowerment in the DTC space (such as empowerment as an expression of individual autonomy) and looked at how we can avoid unduly 'simplistic and binary assessments' of whether or not a technological practice such as DTC testing empowers or disempowers consumers.

The joint ESHG/EMPAG Symposia on 'The Future Lies in Uncertainty' provided excellent discussion on the clinical significance of findings through new genetic technologies, and that often the significance is unknown until much more evidence (bioinformatic or clinical) can be gathered. The phenotype to genotype approach represents a quantitative as well as a qualitative leap in clinical practice. Overall, this has important implications on how consent is obtained, for example with whole genome sequencing. With new technologies gathering lots more data (often with unknown

Cont. page 5

GSNV Volunteer Profile:

Elicia Pettirosso

My name is Elicia and I am currently studying a Bachelor of Science, majoring in Genetics. I'm really interested in the ways that recent enhancements in genetic knowledge and technology are being applied to the healthcare arena. I hope to one day enter this fascinating field, so volunteering with the GSNV seemed like the perfect opportunity to gain firsthand insight into various genetic disorders and the support networks that exist to assist people living with these conditions.

I'm currently volunteering with Australian Disorders of the Corpus Callosum (AusDoCC), a support group that assists individuals with complete or partial absence of the corpus callosum (the band of nerve fibres that connects the two brain hemispheres) and their families.

Being a rare condition with different variations, symptoms and clinical presentations, the support group is vital for individuals and their families to meet, make friends and work together towards promoting the disorder so that better diagnoses and treatments can be made. In my position I have been involved with the development of a member database, working on a symptoms list so others can better understand the condition, and assisting with a volunteer grant application.

I will also be helping with preparations for the next conference to be held in May 2017. It is so inspiring to see how passionate and hardworking this group is. I feel privileged to work with AusDoCC and I thank them and the GSNV for affording me this opportunity. ■

Drug trial for achondroplasia

By Tiffany O'Brien

My name is Tiffany O'Brien. I have completed a BSc in Biomedicine and a PgD in Genetics. I have recently begun working as an intake assistant at the Monash Familial Cancer Centre. I am currently a first year student in the MSc Genetic Counselling degree at the University of Melbourne, and am volunteering with the Genetic Support Network of Victoria.

Achondroplasia is the most common form of dwarfism and affects approximately 96,000 people worldwide. The condition can be associated with serious health complications and children with achondroplasia can require surgery. It is caused by mutations in the FGFR3 gene, which occur randomly in the majority of cases. The FGFR3 gene ordinarily functions to regulate bone growth, but in achondroplasia this gene is overactive and subsequently stops new bone cells from forming. This means that cartilage which would normally become bone is no longer able to be converted, leading to short stature. Complications such as back pain, scoliosis, skull abnormalities, and joint pain, may also be present.

Previously there had been no successful treatment for achondroplasia, despite growing knowledge of the condition over the last 20 years. However, a small molecule called CNP (C-type natriuretic peptide) has been found to be an important bone growth regulator and has been identified as an exciting target for therapy. CNP has the ability to block the FGFR3 pathway, which is overactive in achondroplasia, thereby allowing normal bone growth. Initial studies using injections of CNP in mice with the condition were found to reverse the decrease in bone growth. A pharmaceutical company, BioMarin, has since developed a modified CNP molecule for use in humans. Initial studies using monkeys, and later healthy adult male volunteers, showed promising results. The CNP protein, called BMN-111, is administered by a daily subcutaneous injection, similar to insulin injections for diabetics.

Professor Ravi Savarirayan, an expert in genetic bone disorders, has been the driving force in bringing the drug trial to Melbourne. The Melbourne site, located at the Murdoch Children's Research Institute and Royal Children's Hospital, began recruiting children in 2012 and is currently the largest centre for this trial. Professor Savarirayan has enrolled 24 children in the study thus far.

So far the results of the drug trial have been promising, with the latest results showing a 50% increase in bone growth velocity in the first 6 months of the treatment. The drug has shown to have no serious adverse effects to date and results from the trial are thought to be 'encouraging'.

They hope the trial will decrease the need for surgeries in patients and lead to better spinal and skull bone growth. Professor Savarirayan anticipates that the trial will be extended for a further 5 years, and may open up to include younger children with the condition in the near future. This drug trial provides an exciting opportunity for the treatment of achondroplasia which has the potential to transform the lives of children with this condition. ■

REFERENCES AND FURTHER READING:

1. Gene Reviews: www.ncbi.nlm.nih.gov/books/NBK1152
2. BioMarin Release: investors.bmrn.com/releasedetail.cfm?releaseid=918431
3. The Conversation article: theconversation.com/new-dwarfism-drug-shows-how-innovation-can-be-done-well-50527
4. The Courier Mail article: goo.gl/jgxWFG
5. Short Statured People of Australia Support Group: www.sspa.org.au

MyGene2
Uniting families for sharing and discovery

A global registry joint project by the University of Washington and the Garvan Institute of Medical Research, where families with rare genetic conditions who are interested in sharing their health and genetic information can connect with other families, clinicians, and researchers, to help identify others with similar genetic changes.

See the website **www.mygene2.org**

Message from the team Cont. from page 3

clinical significance) we can no longer expect that patients are able to consider 'all the possible outcomes' from such testing during the consent process. The session focused therefore on the issue of how much we can tolerate 'uncertainty' in the clinic. The following session followed on with a discussion on personal genomic testing and how individuals understand and respond to genetic risk information for a range of conditions. The main topics addressed public understanding and attitudes regarding testing options, the challenges in communicating test results the psychological effects of genetic risk, and health behaviour changes following testing.

The EMPAG session on day three (Mon May 23) looked at incidental findings and consent, and continued the discussion on genetic testing and incidental findings but focused more specifically on broad consent, consent models and 'pre and post' disclosure attitudes toward the return of test results. In this symposium Ivan Macciocca of VCGS, gave an excellent presentation on the Melbourne Genomics Health Alliance 'shared' clinical exome sequencing consent form which was developed for clinical services and testing laboratories working with the ten member organisations of the Alliance (e.g. VCGS, MCRI, Melbourne Health etc.). The aim was to establish a simple and standardised consent form that could be used across multiple health facilities and ultimately enable data sharing in an ethical manner.

The third presentation in this symposium was excellent and aptly considered professional and family ethics in the era of unsolicited findings. The session was entitles 'who is my families keeper' and moved the audience to think seriously about the ethical dilemma of protecting patients' privacy and potentially providing lifesaving information to relatives. During this session the presenter successfully got me thinking about 'who is responsible for conveying genetic risk information to family members?' The current debate on incidental findings draws some attention to the patient as a kind of moral agent co-responsible for their children's or siblings health. The conclusion drawn in my mind is that there indeed a moral basis for the claim that both patients and health professionals have a duty to warn relatives that are at risk of hereditary diseases, particular where early intervention or clinical intervention is needed. New strategies for genetic disclosure and shared responsibility are indeed needed moving into the genomics future.

Family and patient communication was an important theme throughout EMPAG/ESHG but also very prominent in the satellite conference for the Transnational Alliance for Genetic Counseling (TAGC). Here I had the wonderful opportunity of co-facilitating a TAGC session on 'Facilitating family communication about genetic risk development and the use of novel interventions to assist that process'. The session was conducted by myself, Allison Metcalfe (UK), Diana Scotcher (UK) and Margaret Sahhar (VCGS, Melbourne). We worked toward developing a review of the current state of interventions in promoting family communication. Small group exercises were develop to assist participants to develop techniques to support family communication in their own professional practice. The session was concluded with a facilitated discussion of the benefits and the challenges of introducing family communication techniques into genetic counselling curricula, with reference to cultural implications. The basis for the session was founded on the fact that there is sufficient evidence suggesting that many families experience difficulty in talking about an inherited genetic condition that affects one or more of them. There has been work in the UK, developing interventions in the in psychiatric setting, that has shown promising results in multi-family discussion group settings that could be applied to other scenarios such as talking about genetic risk. Often parents want more support from health professionals about managing inherited genetic risk within the family, and advice about talking to their children. There is however few appropriate opportunities to do so, and genetic counsellors (GC) are uncertain about how involved they should be in helping families to communicate risk information. Current practice focuses predominantly on the support of the individual affected or at risk, often to the exclusion of the wider family unit. Alison Metcalfe and team in the UK, have conducted the design of an intervention that will assist parents and children in talking about, and coping with the genetic risk affecting their family. Following some initial research and consultation with senior family therapists, GC leaders, patient group representatives and researchers, it was agreed that a multi-family discussion group (MFDG) intervention might be the most suitable mode in the genetics context. Their work has been outstanding and at this stage they are now working on the further evaluation and testing of the effectiveness and economic viability of their intervention, before it is integrated into genetic counseling practice.

Excluding the stunning location, the tapas, the weather and excellent collegial company in Barcelona, the ESHG/EMPAG/TAGC conferences were again a highlight for me and perhaps provided a fantastic finale to a year of conferencing, workshopping, networking and learning. After just over a year abroad I look forward to returning to my regular desk at the GSNV and continuing to work to support the genetics community in Victoria. Post AGM we have a number of new committee members and a new President, and Vice President. I welcome Monica Ferrie, Maree Maxfield, Abbie Kinniburgh, Christine Williams, Bill Ellerton, Catherine Beard, Emily Higgs and Anna Jarmolowicz to the GSNV team, and know that together we can work successfully to achieve outcomes that make a difference to those we serve. See you all July 18!

Louisa Di Pietro
Group Leader, GSNV ■

Health Professionals Attitudes to Using Whole Genome Sequencing in Newborn Screening Programs In Australia

You are invited to participate in a 15 minute anonymous online questionnaire designed to explore health professionals' attitudes towards the use of new genetic technologies in newborn screening programs.

Specifically, University of Sydney researchers are asking for your opinion, by the 31July, on the potential use and ethical issues pertaining to whole genome sequencing (whereby the whole genome is analysed) in newborn screening.

For more information or to complete a survey, please go to: **https://www.surveymonkey.com/r/HPsWGSinNBS** ■

Quality of Life for People with Intellectual and Developmental Disabilities

Researchers from Macquarie University are conducting a research project about quality of life for people with intellectual and developmental disabilities between the ages of 30 and 50.

The project involves a survey that will take approximately 30 minutes, the survey can be done by an individual with or without help, or on behalf of the individual. If you would like to get involved or would like more information email **peta.ryan@mq.edu.au** ■

RESEARCH

GUIDELINES FOR RESEARCH PROJECT INCLUSION IN THIS SECTION:

If you would like your research project advertised in this section, please send us a brief project description with your contact details as well as documentation of the ethics approval for the project.

Australian Patients and Families' Perspectives on Genome Sequencing

Genetic Alliance Australia (GA) invites you to be involved in a project which is the first of its kind in Australia.

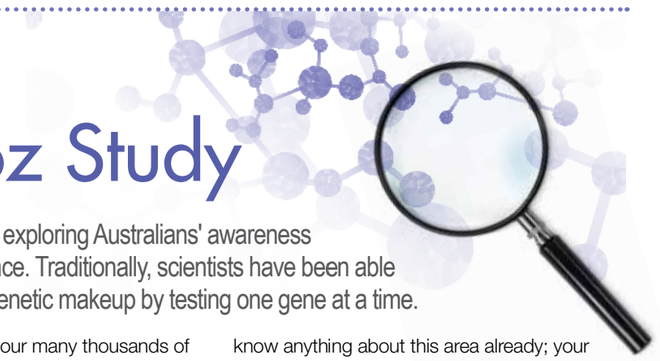
The project launched by GA is called "Australian patients and families' perspectives on genome sequencing". The aim of the project is to produce an "Australian Patient Charter on Genome Sequencing". This will outline the views and opinions of patients and families regarding genome sequencing in the healthcare system. This will become an important reference guideline for policy and decision-makers when incorporating genome sequencing into the Australian healthcare system.

The project will involve an anonymous online survey for individuals and families affected by genetic conditions, and aims to gather their views and opinions on the use of genome sequencing in a clinical setting. By participating in the survey you will have access to a video "Whole Genome Sequencing & You" which will serve as an excellent education resource.

There are various ways to access the survey:

1. Invitation PDF Advert: **http://www.geneticalliance.org.au/cmsAdmin/uploads/genome-sequencing-advert-v0-8.pdf**
2. Information on GA Website: **www.geneticalliance.org.au/genome.php?1**
3. Direct link to the survey: **www.surveymonkey.com/r/genomesurvey** ■

The Genioz Study



The Genioz study is exploring Australians' awareness of new genetic science. Traditionally, scientists have been able to look at people's genetic makeup by testing one gene at a time.

Now, we can test all of our many thousands of genes at the same time, and generate our own personal genetic profile to give healthy people info about their own genetic makeup! Called 'personal genomics', this broad group of genetic tests can be used in many ways, including ancestry, paternity, sporting ability, and health. We want to learn what people think about the topic – it doesn't matter if you feel you don't know anything about this area already; your opinions and views are valuable to us and our research partners.

Genioz is collecting data via an online survey, until the end of 2016. Anyone, regardless of their level of knowledge, can participate. Have your say here: **www.genioz.net.au/page/survey** ■

RESEARCH

The involvement of patient organisations in rare disease research: an Australian study

By Ms. Deirdre Pinto, Dr. Dominique Martin and Dr. Richard Chenhall Cont. from page 1

a goal to support research. Although most are small volunteer-based groups, 59% of RDPOs had provided funding to researchers in the previous five years and 77% had given non-financial support, such as helping to identify participants for clinical trials or studies (56%). Other forms of engagement included disseminating research information (79%), contributing to researchers' patient registries or biobanks (41%), lobbying authorities (36%), providing information or counselling to assist research participants (34%), conducting the RDPO's own studies (26%), and participating in committees established by government or research institutions (20%). Nearly all RDPOs (95%) had conducted at least one of these activities in the five years before the survey. The funds, logistic support and expertise provided by some RDPOs had contributed to advances in scientific knowledge.

Several RDPO leaders proactively facilitate connections between researchers working in different research institutions and/or disciplines, thereby promoting the collaborations now recognised as critical to successful therapy development. Other RDPO leaders described how their involvement had helped align research agendas with the needs of patient communities or protected the interests of patients who participate as research "subjects".

Notwithstanding these important roles, RDPO leaders identified considerable challenges relating to their research involvement. The difficulty of raising funds for research was most frequently mentioned, but research-engaged leaders are confronted with many other challenges. For example, they may struggle to direct RDPO funding in ways which best meet the goals of the organisation, or they may lack the expertise and power to assert their knowledge and ideas when collaborating with researchers. Many leaders also identified aspects of the research environment – such as a lack of government and industry funding for or "interest" in their diseases – as barriers to their RDPOs' involvement in research. In our Orphanet journal paper, we discuss the difficulties reported by RDPO leaders in terms of two systemic issues: the proliferation of and lack of collaboration between RDPOs, and the lack of specific government policies and resources supporting rare disease research and patient advocacy in Australia.

Internationally, there is growing interest in strengthening collaborations between RDPOs and researchers. This is underpinned by the need to alleviate the personal and societal burdens of diseases that collectively affect an estimated 6–8 % of the population. Based on initiatives implemented in other countries, and the findings of our study, we suggest a number of measures to strengthen Australian RDPOs' involvement in research. We believe that such initiatives – ideally implemented as part of a national rare diseases plan – could support Australian research and the aspirations of under-served communities. ■

FOR MORE INFORMATION:
The research paper can be accessed at: www.ojrd.com/content/11/1/2
If you would like more information about the study please email Deirdre Pinto, deirdre.pinto@hotmail.com

REFERENCE:
Pinto D, Martin D, and Chenhall R. The involvement of patient organisations in rare disease research: a mixed methods study in Australia. Orphanet J Rare Dis. 2016;11(1):2.

Ms. Deirdre Pinto (pictured) has worked in health policy and program management for many years, and prior to 2013 was employed at the Victorian Department of Health. She is involved with a United States-based patient organisation for Rasmussen's encephalitis, a rare neurological disease.

Dr. Dominique Martin is a Lecturer in Health Ethics in the Centre for Health Equity. Previously practicing as a medical doctor, Dominique conducts and supervises research into a range of bioethics issues. She is actively involved in international policy development in the area of organ trafficking and "transplant tourism".

Dr. Richard Chenhall is an Associate Professor in Medical Anthropology in the Centre for Health Equity. He is currently working on a number of projects focusing on the health of Aboriginal and Torres Strait Islander Peoples, and is also conducting research related to alcoholism and self-help groups.

RESEARCH

HEALTHY PARTICIPANTS NEEDED

A new Murdoch Childrens Research Institute study is looking into the lung function of children with Cystic Fibrosis, which is the most common life-threatening genetic condition in Australia. But we can't do it by ourselves! We're looking for healthy preschool children between 3 – 5 years to have a lung function test in at the Murdoch Childrens. If you can help us for an hour or so on three occasions over a year, we'd be so grateful – for more information please contact Natalie on 9936 6273.

The Royal Children's Hospital are looking for kids to participate in an exciting research study, the first of its kind in the world! The project, led by Prof. Tony Penington, is examining the head shape of children and the changes that occur during childhood. The 3D Imaging Centre at the RCH will take a 3D photograph of your child's head using 18 specialised cameras. It takes less than a second and is not harmful. Each participant will receive a copy of the 3D photograph on a CD. They are looking for children, especially under five, who do not have a medical condition that affects their growth and bones for this project. Find out more at bit.ly/22I76lz or email **robert.reitmaier@rch.org.au** to arrange a time to have your child's photograph taken.

Carer Wellbeing Research Project

Researchers at Institute for Health and Ageing are exploring the physical activity and mental wellbeing of older carers. Participants complete a 15-minute questionnaire and enter a draw to win a $100 Coles gift card.

Complete the questionnaire online at **www.tinyurl.com/carersurvey** or request a paper copy by calling 0411 098 286.

SUPPORT GROUPS

GSNV Small Grants recipients:

The GSNV is keen to support the vital work of support groups who are often under resourced and in need of a helping hand.

The GSNV Small Grants Program lets us help the community with a small but nonetheless helpful grant that may get a project or much needed purchase under way. Small Grants may be used for a once-off project or activity that an individual or a group wishes to undertake that will benefit the community. The GSNV will be accepting application for the next round of grants in late 2016, dates to be advised.

ORGANISATION/GROUP	DESCRIPTION OF PROJECT
Lymphoedema Association of Victoria	Seeking funding/support for postage to send out the newsletter each quarter
Australian Pompe's Association	Funding to cover printing, stationary, mailing costs to add Pompe's to NBS register
Australia Alopecia Areata Foundation Inc.	Purchasing of 500 window stickers to distribute to hairdressing boutiques and salons
Aspergers Victoria	Safe Talk Workshop upskilling volunteers, depression and anxiety peer support
SIDS and Kids Limited	Administrative and financial support to run Difficult Decisions support group
UsherKids Australia	Videos for website and promotional flyers
CHARGE Syndrome Association of Australia	Cover costs of support group meetings
Hearts In Mind	Home office expenses for two facilitators
People With Multiple Sclerosis Vic Inc.	Provide taxi vouchers and petrol vouchers to assist people who may experience difficulty in attending because of the costs associated with special transport as well as accommodation costs

SAVE THE DATE
30 JULY 2016

COMMUNITY CONFERENCE
30 July 2016
CYSTIC FIBROSIS Victoria

CFV'S ANNUAL COMMUNITY CONFERENCE

The Cystic Fibrosis Victoria (CFV) Community Conference brings people from the Victorian cystic fibrosis (CF) community up-to-date with latest developments and to provide opportunities for discussion with medical and health experts and those affected by CF.

When: Saturday, July 30 2016
Where: Augustine Centre, 2 Minona Street Hawthorn
Cost: Free for members and $30 for non-members

For more information or to register your attendance to the 2016 conference, visit **www.cysticfibrosis.org.au/vic/community-conference**
Registrations close Thursday, July 21.

SUPPORT GROUPS

From left to right: Adrienne Sexton, Karni Liddell, Hannah Pennington, Professor Ingrid Winship

The Neuromuscular Support Group for Young Adults second informal meeting

By Hannah Pennington

The Second Neuromuscular Support Group for Young Adults was held at VicHealth, Carlton on Thursday 28 January, 2016.

The evening began with an introduction from me, Hannah Pennington – welcoming my guests and thanking them for attending. My genetic counsellor, Adrienne Sexton (a genetic counsellor from the Royal Melbourne Hospital) introduced our first speaker, Professor Ingrid Winship (a Professor of Genetics and Executive Director of Research with Melbourne Health) who gave us an insightful talk on the services and testing options that are offered within the Royal Melbourne Hospital for young adults who suffer from neuromuscular conditions. She also outlined the obvious gap in medical services for those particular individuals.

Our second speaker for the evening was the vivacious Karni Liddell. Karni was flown in from Brisbane to attend our meeting, as she and I have both been unable to attain a conclusive diagnosis for our neuromuscular conditions. Karni spoke about her personal experience with spinal muscular atrophy, and her struggles as a child and young adolescent to adult with the condition. Karni is also a former Paralympic swimming competitor, winning a bronze medal at both the 1996 Atlanta Games and the 2000 Sydney Games.

Next to speak was my friend, Fin Kelly. I met Fin through Adrienne Sexton at the Royal Melbourne Hospital in 2014. Adrienne put us in contact after she suggested that I chat with other individuals with similar symptoms and age to myself. Fin suffers from ARSACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay) and has been experiencing symptoms for his entire life; they include difficulty coordinating movements and muscle wastage, which we have in common. Fin wrote a poem to express his feelings in regards to his condition; it was incredibly beautiful and moving. Our final speaker for the evening was Emma Bradhurst. Emma and I met during the first Neuromuscular Support Group for Young Adults in 2015. Emma was put in touch with the support group as she also sees a geneticist at the Royal Melbourne Hospital as she suffers from Cerebral Ataxia. Emma's condition affects her gait and muscle coordination, which are also symptoms that I share.

Emma spoke about her diagnosis as an adult, and how it was unknown to her throughout her childhood and early stages of adulthood, that she has a neurological condition. She also spoke very passionately about her belief in her faith, which assists her in the daily struggles of her disorder. The purpose of this particular meeting, previous and (hopefully) future meetings are to promote the importance of supporting one another through our difficulties as young adults living with neurological conditions. It is my hope that we continue to strive to ensure others feel supported and cared for, regardless of their disability.

Children's Tumour Foundation UPDATE

By Natalie McLean, Support Coordinator (Victoria/ Tasmania), Children's Tumour Foundation | NF Australia

SAVE THE DATE
27 AUGUST 2016

Melbourne's next NF1 Paediatric Seminar

Hear from speakers involved in many aspects of management of NF1 in children, learn more about the NF Clinic @ RCH's progress.

A great opportunity to meet other families in the Melbourne and Victoria region who are on the NF journey like you. More details to follow. Spaces will be limited. Registrations will open in July.

Hobart Information Seminar a Success!

CTF's first ever information seminar in Tasmania was held in Hobart on the evening of Wednesday 20 April.

Professor David Amor from the Victorian Clinical Genetics Service who travels to Tasmania regularly for clinics spoke about the diagnosis, management and in brief detail on the genetics of NF1. We had a good turnout, some even travelling from the north west of the State! Those who attended indicated they found benefit from the event.This event was not recorded.

Should you require information about doctors in your area, resources and support please contact Natalie McLean on 03 9936 6268 or **Natalie.mclean@ctf.org.au** if you are located in Victoria/ Tasmania, and Sally Maspero 02 9713 6111 or **Sally.maspero@ctf.org.au** for the rest of the country.

Albury/Wodonga May Support Group Meeting

The wonderful support group located in Albury/ Wodonga and run by Lana Hanssens met for May awareness month on Saturday 7 of May in Wodonga.

Six families attended the event as did Natalie McLean from CTF and Michelle Sproule from the Victorian Clinical Genetics Service.

SUPPORT GROUPS

Cystic Fibrosis Victoria update

By Sarah Burns, Project Coordinator, Programs and Support Services Team, Cystic Fibrosis Victoria

Cystic fibrosis carrier screening

For people living with cystic fibrosis (CF), the month of May is also known as 65 Roses month, a national campaign raising much needed awareness and funds for those living with CF and their families.

During 65 Roses month Cystic Fibrosis Victoria (CFV) and the CF community participated in a number of fundraising and awareness raising activities including; 65 challenges, tin rattling, community fundraisers and selling merchandise and roses. The month long event came to a successful close after more than 60 volunteers braved the cold and wet weather in Melbourne's CBD on the 27 and 28 May to raise just under $10,000.

However, it wasn't all about the dollars and was equally about raising awareness of cystic fibrosis in the community, so CFV took the opportunity to launch their much anticipated CF carrier screening video campaign. A set of informative and innovative videos was developed to apprise the Victorian community of CF and the availability of CF carrier screening. After all, on average one in 25 people carry the CF gene - most of whom are completely unaware that they are carriers.

Making the decision to know your CF carrier status is personal choice, however almost all children born with CF are born to parents who have no family history. Meaning, even people with no family history of CF could be a carrier. For a child to be born with CF both parents must be carriers of the CF gene change. Carriers of the CF gene change do not have CF nor show any symptoms of CF. Every four days a child is born with CF in Australia. CFV is dedicated to raising awareness of how you too could be a carrier of the gene change that causes CF.

CF Carrier screening (genetic testing) services are available in Australia to help identify if you are a carrier of the CF gene change. This test can be done by giving a blood or saliva sample. Carrier screening is available to those people 18 years and over and is the only way to find out if you are a carrier of the gene change that causes CF.

CF Carrier screening can be ordered prior to pregnancy or early in a pregnancy by your GP, obstetrician or a genetic counsellor.

If you are thinking about having a family or are just interested in finding out your CF carrier status, then consider speaking to your GP about CF carrier screening.

To watch CFV's CF carrier screening videos or to read more visit **www.cfscreening.com.au** ■

The CF Carrier Screening Education Campaign is supported by the Rotary Club of Balwyn.

Personal story: Renae Prendergast

Prior to Kari being diagnosed with CF my family had no known history of CF. My husband has a cousin with CF, however, his family were never made aware of the implications that this could pose for them in their future plans for a family. Prior to meeting my husband there was a falling out, meaning I was not aware of the implications either and had little knowledge of CF.

After Kari was diagnosed with CF, we both decided to be screened to confirm the diagnosis and the particular CF gene change. I would highly recommend couples be screened for CF so they can make informed choices about their pregnancy. Especially if there is a high chance they could have a child with CF.

From our experience I wouldn't want any parent to have to face what we do on a daily basis. I'm so happy to have our gorgeous girl in our lives, however, CF is a relentless, unforgiving, life shortening, debilitating illness and an emotional roller coaster. As a parent you do not want to see your child battle this horrible disease. ■

The opinions expressed in this article are the author's own and do not necessarily reflect the view of the GSNV. We respect the diversity of opinion in our communications and acknowledge the personal experiences conveyed in personal stories are based on the author's perception of the reality around them.

SUPPORT GROUPS

Alpha-1 Association of Australia update

The Alpha-1 Association of Australia (AAA) is a support group for people with Alpha-1 Antitrypsin Deficiency (Alpha-1), their families and their carers. Alpha-1 is a genetically inherited condition that can affect the liver and lungs and can affect people of all age groups. By Jenni Nankervis

ALPHA-1 REGISTRY

The AAA is in the process of establishing a National Alpha-1 Register for Australia. The Murdoch Children's Research Institute has agreed to develop and host this register. The AAA and Medical University of South Carolina are collaborating on this project.

The AAA is currently looking for a Project coordinator / Research Assistant for this project. Interest from prospective sponsors is most welcome.

NEONATAL SCREENING FOR A1AT

The AAA is also working on adding Alpha-1 to the National Neonatal Screening Program. In 2015, one of our members wrote a 147-page Newborn Screening Application with reference to over 500 articles about Alpha-1. The services of a Health Economist are needed to investigate the cost of having Alpha-1 included in the newborn screening process.

The National Screening Program is due to be up and running in the first half of 2016. **www.genomics.health.wa.gov.au/nbspf**

ALPHA-1 CONFERENCES AND CONGRESSES

The 5th Alpha-1 Global Patient Congress in Barga, Italy, held April 9-11 2015, drew 200 people from 26 countries to hear renowned Alpha-1 scientists, clinicians, experts, industry partners, caregivers, patients and their family speak on the status of the latest research. See more at: **www.alpha-1global.org/2015-Patient-Congress/Home**

I was fortunate enough to attend this event with others representing Australia's Alpha-1 community. It is always a rewarding experience to meet the many other support group leaders and discuss what is happening in our countries. Discussion revolves around treatments, research and new ways to raise awareness and to provide support to our members. The next Alpha-1 Global Congress is planned for 2017. The venue for this event has not been announced as yet. In June 2016, Steven Knowles, President of the AAA, member of the Global Steering Committee for Alpha-1 Global, representing Australia and New Zealand, attended the 25th Alpha-1 National Educational Conference in Miami, USA. **www.alpha1.org/Alphas-Friends-Family/Education/National-Education-Conference**

AWARENESS

The Alpha-1 Association of Australia has been successful in having November declared as Alpha-1 Awareness Month.

SOCIAL MEDIA

We currently have a number of social media groups which provide support and information about Alpha-1. The AAA Forum, a chat group, the Facebook group Alpha-1 Association of Australia and its Twitter account are accessible via our website. See the link at top of article.

There are other Facebook Groups - Alpha-1 Oz and Alpha 1 Talk and Support which were started up by members of the AAA. Our social media presence has grown considerably over the past couple of years and because of this, our membership has grown.

For more information about Alpha-1 go to www.alpha1.org.au ■

Congenital adrenal hyperplasia for inclusion on the NBS

By Caron Brindley acting President of the CAHSGA

Congenital adrenal hyperplasia, or CAH, is a rare (1 in 15,000 children are born with this condition each year in Australia), genetic disorder that affects the adrenal glands.

The adrenal glands produce hormones, including sex hormones and cortisol and aldosterone. A person who has CAH does not make enough of the hormones cortisol and aldosterone, and makes too much androgen, which is a male sex hormone.

Girls are usually diagnosed at birth as the overproduction of androgens causes ambiguous genitalia. Boys are usually diagnosed within the first weeks of life when they are unable to produce the essential hormones cortisol and aldosterone and have an adrenal crisis, a life threatening medical emergency.

The Congenital Adrenal Hyperplasia Support Group Australia (CAHSGA) was formed in 1985. We are working to improve knowledge and services for those affected by CAH. We are a small not for profit group run entirely by volunteers. Over the years we have had several campaigns to lobby federal and state health ministers to include CAH on the Newborn screening (NBS) program. NBS for CAH is still not routinely tested for in Australia. We are one of the only developed countries who do not test for CAH. Indeed, parts of the USA and New Zealand have been screening for CAH for more than 25 years. The Philippines has also recently introduced screening.

The mortality rate for boys with CAH is higher than that of girls. If CAH was on the NBS this would not be the case. Many newly

Cont. page 13

GENETIC SUPPORT & ADVOCACY

Rare Disease Day 2016 – The patient voice

By Keri Pereira

Rare Disease Day (RDD) was held on 29 February 2016 (a rare day!). RDD aims to raise awareness of the impact of living with a rare disease on individuals and families.

This day also aims to highlight some of the challenges and achievements that happen along their journey. It's an international day of celebration which recognises the experiences, triumphs, difficulties and the human faces of the rare diseases community worldwide. This year's slogan was "Join us in making the voice of rare diseases heard", which emphasised the patient voice and the importance of recognising the patient's experience. At the GSNV we use this day to bring together researchers, clinicians, and the community; and share information between these groups. This year we held a seminar with presentations from health professionals, researchers and support group leaders.

The GSNV was very lucky to have international researcher Dr. Kym Boycott presenting the work she has been conducting on rare diseases. Kym is a medical geneticist at the Children's Hospital of Eastern Ontario (CHEO) and clinician scientist at the CHEO Research Institute. She was Lead Investigator of the Genome Canada and CIHR funded 'Finding of Rare Disease Genes in Canada' (FORGE Canada) project, which investigated the molecular aetiology of rare paediatric diseases. She currently leads the Genome Canada and CIHR funded large-scale project 'Enhanced CARE for RARE Genetic Diseases in Canada', which is focused on improving the clinical care of patients and families by expanding and improving the diagnosis and treatment of rare diseases.

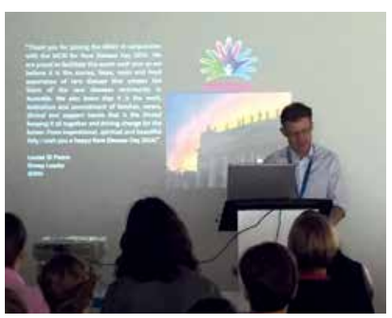
As her work suggests, Kym is highly interested in ensuring research into rare disease continues. As the Chair of the Diagnostics Committee of the International Rare Disease Research Consortium Kym stressed the importance of international collaboration as when working together, we are more likely to be successful in gaining a better understanding of rare diseases.

Professor Kathryn North, the Director of MCRI and Associate Professor Tiong Tan, Clinical Geneticist at VCGS both gave fantastic presentations on the institute's perspective on rare disease and the clinical and research initiatives that are under way. The MCRI is taking a leading role in progressing research and clinical management for people with rare disease through projects such as the Melbourne Genomic Health Alliance (MGHA) and the Australian Genomics Health Alliance.

We were extremely happy to have the founders of UsherKids Australia presenting as they drove home the theme of "the patient voice". They spoke eloquently about their children's diagnosis of Usher syndrome and their journey to starting the newly formed support group. The relationships that they have built with clinicians and researchers has seen them become part of the Deafness flagship with the MGHA, which is a big step forward for children diagnosed with hearing loss.

The overarching theme which came out of the day was the importance of collaboration between clinicians, research and the community, not only nationally but internationally as well. The sharing of knowledge and resources will only benefit the rare disease and medical community.

The GSNV would like to thank MCRI, the presenters, and all that attended the informative day. We would also like to acknowledge that Kym's visit to Australia was supported by the Royal College of Pathologists of Australasia and the GSNV would like to thank them for allowing Kym to speak at this event. ■



GENETIC SUPPORT & ADVOCACY

Your Say... Your Thoughts

In this section of the newsletter we ask for support group members to write about an issue that is important to them. We want to hear about the issues that are close to your heart, we value your contribution.

The McMahon Story

This story was provided by the Syndromes Without A Name support group.

Kate McMahon, shares her story about what it is like to receive a diagnosis and how they cope when the diagnosis is so rare.

I used to joke that my daughter had "Olivia Syndrome" when people asked me what was wrong with her. It seemed a better thing to say than "actually, we really don't know". Because for two and a half years, my husband Tim and I had no idea why our beautiful little ray of sunshine had so many issues. It is a huge relief to be able to now say to people "she has Kleefstra syndrome". There are only about 300 people in the world with "KS" and it is so rare not much is known about it. Typically, those with the syndrome have moderate physical and intellectual disabilities and experience developmental delays, but there is a large spectrum of abilities so we are still working out where we fit.

When I was asked to write about what it was like to get a diagnosis for Olivia, I guess the overwhelming feeling we have is that we are lucky. I think you always assume, until you learn differently, that doctors hold the answers to most ailments. We had done a lot of tests including an MRI and several X-rays and ultrasounds as well as blood tests and photographs, so it was sobering to be told by the genetics team at the Royal Children's Hospital that Olivia had an undiagnosed genetic syndrome and the likelihood was they would never be able to tell us what it was. Thankfully, in February this year, against the odds, we finally got our answer. Olivia had been put forward to participate in the Melbourne Genomic Health Alliance research project, which was a pilot to show the effectiveness of exome and genetic sequencing in diagnosing rare syndromes. We were warned in advance the chance of her exome sequence finding something was only 20 per cent, which had really haunted us. We wanted more children in the future, but were not prepared to shoulder the unknown risk of having another child with Olivia's issues.

We got a call about ten months after the test was sent away to come in for a chat with our geneticist, Dr. Zornitza Stark. She told us she had found the tiny "spelling mistake" in Olivia's genes which resulted in Kleefstra. There were a lot of tears and hugs.

I think every parent holds onto that tiny bit of hope that all of a sudden, their child may just "come good" until they are told otherwise, even when you know that is against every instinct. But reading the brochure on KS, it was like someone was perfectly describing our daughter. It was so overwhelming and inspiring to see other families talking about how their children were at school, walking and talking – milestones we were never sure until that moment if Olivia would be able to meet. But there are many obstacles we will have to deal with too. We have since linked in with some of the parents whose children have KS on Facebook and we know that there are many, many very tough challenges to come for Olivia and ourselves. Overall, however, we are so relieved to have the diagnosis. ■

Congenital adrenal hyperplasia for inclusion on the NBS

Cont. from page 11

diagnosed baby boys with CAH spend many weeks in Intensive Care Units after becoming critically unwell due to adrenal crisis. Experts suggest the lack of screening in Australia costs the life of one child every few years, with several children almost losing their lives every year. If CAH was on the NBS this would not be happening to our families. With early treatment, children with CAH enjoy a wonderful quality of life.

A delay in diagnosis can be fatal for children with more severe forms of CAH, but even for those with less severe CAH the impact of late diagnosis carries a unique burden.

Growth and pubertal development are affected, and children must then consult specialists and take expensive, unpleasant drugs for many years to ensure their development progresses as steadily as possible. CAHSGA is working with medical professionals and advocacy groups to ensure CAH is included on the NBS in Australia.

These campaigns have been in the form of:

- on-line petitions **www.change.org/p/introduce-newborn-screening-for-congenital-adrenal-hyperplasia-cah-in-australia**
- twitter campaign **twitter.com/AUSCAHscreening**
- personal correspondence and meetings with local and state members of parliament.

Support group website: **www.cah.org.au** ■

GENETIC SUPPORT & ADVOCACY

USHER KIDS AUSTRALIA

Bringing together families of Children with Usher Syndrome in Australia

Usher syndrome is a rare genetic condition characterised by hearing loss or deafness, the progressive loss of vision and in some cases, vestibular dysfunction. By Emily Shepard, UsherKids Australia Co-Founder

The loss of vision is caused by an eye disease called Retinitis Pigmentosa (RP), which affects the light sensitive area of tissue on the back of the eye (the retina) causing night blindness and tunnel vision. My son Louis was diagnosis with Usher syndrome when he was three and a half. After being born with a profound hearing loss, we knew there was something else wrong when he struggled to keep his head up, was late to sit, late to crawl and late to walk so we pushed for genetic testing.

It was a very lonely time for our family as there was no support group to turn to, and many of our doctors knew very little about the condition. Since then I have been working with another parent in Melbourne to improve the diagnosis, education and support for families of children with Usher syndrome in Australia. We both had great difficulties navigating specialists, information and support for our sons after diagnosis and vowed to improve this path for families that follow us. Our support group, UsherKids Australia was officially formed in 2015 and our website **www.usherkidsaustralia.com** was launched in early 2016.

In the last 18 months we have managed to bring a greater awareness of Usher syndrome to our clinicians both in Melbourne and to the broader community, and continue to dedicate our time to the following areas:

- Working with Able Australia to improve a resource kit provided to newly diagnosed patients to ensure it is up to date and relevant for our children.
- Raising awareness through Early Intervention Centres to identify deaf children who may have gross motor delays consistent with vestibular dysfunction to refer for genetic testing.
- Working with Ophthalmologists to improve the clinical care protocol for children with Usher syndrome, including adding specialised retina photographs to routine checks to chart the natural history of the eye degeneration.
- Sharing lived experiences with newly diagnosed families.
- Holding events and public awareness campaigns to help identified children of all ages across Australia living with Usher syndrome to strengthen our growing community, including Usher Syndrome Awareness Day, articles in CHILD and Marie Claire magazines and presenting at GSNV Rare Disease Day.
- Lobbying research groups to fund Usher syndrome studies and treatment trials.
- Creating educational material for clinicians to provide to newly diagnosed families at the time of diagnosis.
- Providing an online resource for both families and medical professionals of all the latest Usher syndrome news, research and support services.
- Advocating for all Usher syndrome patients in Australia to register with both The Australian Inherited Retinal Disease Register and DNA Bank, and The International Usher Syndrome Registry to further accelerate global research.

You can contact us at **info@usherkidsaustralia.com** or through our website **www.usherkidsaustralia.com.**

SERVICES

50 YEARS OF VICTORIAN NEWBORN SCREENING:

Past, Present and Future Symposium

To celebrate the anniversary of Victoria's 50 years of Newborn screening, the providers of the test, the Victorian Clinical Genetics Service hosted a special one-day symposium on the 10 February.

Researchers, doctors and nurses from across the State who had worked on the program since its inception were joined by families whose children were diagnosed with conditions as a result of newborn screening, to mark the milestone together.

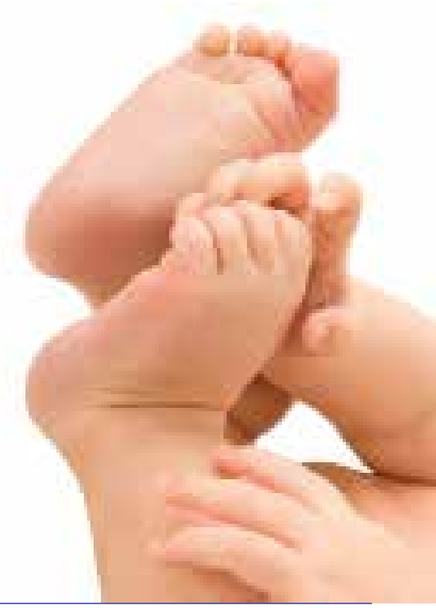
The newborn screening test is voluntary, with 98% of all parents in Victoria choosing to participate. A few drops of blood are collected via heel prick of a 48 to 72 hours old newborn, and soaked onto a special absorbent card (Guthrie Card). The cards are sent to VCGS and screened for 32 different serious conditions that benefit from early detection and treatment, such as phenylketonuria (PKU), hypothyroidism, and cystic fibrosis (CF).

Bridget Wilcken, from the Children's Hospital at Westmead spoke about the history of the test, which begun with PKU. PKU arises from an enzyme deficiency that leads to the accumulation of phenylalanine in the blood, which can cause brain damage. The stimulus for screening was the discovery of a dietary treatment in the 1950s. It was found that brain damage could be prevented if phenylalanine was removed from the diet, beginning in the first few weeks of life. By 1973, the screening test for PKU was universal in Victoria. In 1975, a thyroid stimulating hormone test for hypothyroidism was added. If treated before 3 months of ages, babies are prevented from developing significant intellectual disability. In the 1980s CF, biotinidase deficiency, hemoglobinopathies, and galactosemia were added. The 1990s saw a pivotal change with the introduction of Tandem Mass Spectroscopy, which separates compounds by mass and structure, to screen for a further 22 conditions. Since 1998 however, no new conditions have been added to the test, as currently, funding is not available to screen for additional disorders. Congenital Adrenal Hyperplasia, for example is not part of NBS in Victoria, but is screened for in New Zealand (see page 11 of this issue).

Research is currently underway at the Murdoch Childrens Research Institute (MCRI) to develop a new test for currently included disorders as well as additional conditions. A national policy framework is currently under development to improve the way new tests are assessed for inclusion in the NBS, as well as ensuring a uniform rollout across Australia. New tests currently being considered for addition to the screen, include Duchenne muscular dystrophy, Rett syndrome, fragile X, long QT, and whole genome, exome or panel sequencing. Discussion focused on the accuracy of these tests and how results will be interpreted. A major concern is also that, new additions to the screen may increase the risk of detecting cases that don't need any intervention.

A family story was also presented by Monique Cooper (see page 17 of the Summer 2015-2016 edition of the newsletter). Monique spoke about her experiences with newborn screening; finding out that her newborn son, Charlie was affected with PKU. When she first learnt about this condition, when he was born, she researched it and learnt that it could lead to severe intellectual disability, seizures, brain damage, and stunted growth. Monique also spoke about how she was overwhelmed by the number of doctors and nurses that her child needed to be seen by. She had many questions about his future, but was reassured that the symptoms of PKU can be controlled with diet and lifestyle changes. Monique expressed that she finally started to feel that Charlie would be okay when she was introduced to the Metabolic Dietary Disorders Association (MDDA), and saw that other children with the same condition were growing up healthily. Monique continues to work closely with the metabolic team at the RCH, who monitor Charlie with regular blood spot tests. Monique is now also president of MDDA, and has done great work advocating for and bringing together families affected with metabolic conditions.

A commemorative booklet was also produced, which is available for download along with the speakers' material. See: **www.mcri.edu.au/NBS2016-content**



SERVICES

The Melbourne Genomics Health Alliance

The huge potential of genomics in medicine is on the way to being realised sooner in Victoria through the work of the Melbourne Genomics Health Alliance. By Anna Jarmolowicz, Gemma Brett, Emma Creed, Ellie Prawer, Giulia Valente, Karen Meehan

Funded by its 10 members and a substantial contribution from the Victorian Government, Melbourne Genomics will over the next four years provide genomic sequencing to Victorian hospital patients with specific conditions and evaluate outcomes in comparison with usual medical care. The Alliance is also establishing systems and processes to support genomics within the Victorian healthcare system.

Originally formed in 2013, the Melbourne Genomics Health Alliance brings together the clinical, research and teaching strengths among Victoria's leading hospitals and research organisations to overcoming the challenges of delivering genomic medicine. The Alliance's vision is for Victoria to be a world leader in the use of genomics in healthcare.

The Alliance members are:

- The Royal Melbourne Hospital
- The Royal Children's Hospital
- The University of Melbourne
- The Walter and Eliza Hall Institute of Medical Research
- The Murdoch Children's Research Institute
- CSIRO
- The Australian Genome Research Facility
- Peter MacCallum Cancer Centre
- Austin Health
- Monash Health

Melbourne Genomics has recently employed three new genetic counsellors to join the existing two already undertaking work for the Alliance.

These genetic counsellors work with clinicians to identify which patients can benefit from exome sequencing, the optimal timing and circumstances for such testing, as well as providing support to individuals and families undergoing testing.

INTRODUCING:

Gemma Brett is a Genetic Counsellor at the Victorian Clinical Genetics Services, and is responsible for counselling families at The Royal Children's Hospital (RCH). She also coordinates and provides genetic counselling within the RCH Neurofibromatosis Clinic. After a completing a Bachelor of Medical Science, Gemma relocated to Germany to complete a Master of Molecular Bioscience (Developmental Biology). She has also undertaken laboratory research in human genetics within Heidelberg University Hospital (Germany) and Karolinska University Hospital (Sweden). Gemma completed her Master of Genetic Counselling in Melbourne, which included a thesis exploring genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice. Before taking up her current role, Gemma worked in international development in Nepal, establishing the Public Health Concern Trust Nepal as a national centre for research excellence.

Anna Jarmolowicz is a Genetic Counsellor at the Victorian Clinical Genetics Services, and is responsible for counselling families at The Royal Children's Hospital (RCH). Anna has a Bachelor of Health Science, majoring in Genetics and Public Health, and a Graduate Diploma in Counselling. Anna has previously worked as a Research Assistant at a not-for-profit organisation with a focus on neurodegenerative conditions. Anna completed her Master of Genetic Counselling in Melbourne, which included a thesis exploring how parents told their children that they were conceived using preimplantation genetic diagnosis. During this time she also worked at the Genetic Support Network of Victoria, providing support and information to individuals, families and support groups.

Emma Creed is the Genetic Counsellor responsible for counselling patients at The Royal Melbourne Hospital. Emma has a Bachelor of Science majoring in Genetics and a Bachelor of Arts majoring in Psychology with Honours. Emma previously worked as a Research Assistant with an organisational psychology research group at the University of Melbourne, specialising in negotiation research. Emma completed her Master of Genetic Counselling in Melbourne, including a thesis exploring the experiences of individuals who established a genetic support group in Victoria. She has returned to Melbourne after working as a Genetic Counsellor at the Familial Cancer Service at Westmead Hospital in NSW.

Ellie Prawer is the Genetic Counsellor responsible for counselling patients at Monash Medical Centre. Ellie has a Bachelor of Science majoring in Genetics from The University of Melbourne. Ellie previously worked as a Research Assistant for the Melbourne School of Global and Population Health. Ellie completed her Master of Genetic counselling in Melbourne, including a thesis exploring the lived experiences of female carriers of X-linked Adrenoleukodystrophy. Ellie has experience working in Familial Cancer and is currently working in general genetics.

Giulia Valente is the Genetic Counsellor responsible for counselling patients at Austin Hospital. Giulia has a Bachelor of Biomedicine majoring in Biochemistry and Molecular Biology. Giulia then completed her Master of Genetic Counselling in Melbourne, including a thesis exploring the attitudes, knowledge and practice patterns of Victorian health professionals in regards to population-based cystic fibrosis carrier screening. Giulia has previously worked as a Genetic Counsellor in Victoria.

SERVICES

WHAT IS GENOMIC MEDICINE?

Genomics is an area within medical science that examines the genome; your complete set of genetic information. Your genome contains instructions (or genes) for how your body functions and your physical characteristics. Your exome is a small portion (about 1%) of your genome that is thought to be most important for health. An exome sequencing test (commonly performed from a blood sample) aims to identify changes, called 'variants', in a person's exome. Currently, when a patient is referred to a genetic health service, the clinical team might have a genetic diagnosis in mind based on their symptoms. It may be possible to do a simple test to sequence (read) one or more genes to confirm this diagnosis. Alternatively, the case might be more complex, and there could be many potential diagnoses involving many different genes. In these complex cases, sequencing all the genes at the same time (exome sequencing) is more helpful. Once all the genes have been sequenced, it is possible to examine them one by one without having to perform a whole new test.

WHAT IS THE ROLE OF GENETIC COUNSELLORS IN THE MELBOURNE GENOMICS HEALTH ALLIANCE?

The role of genetic counsellors within Melbourne Genomics is to facilitate a patient's journey through sequencing the section of their genome that specifically contains the genes, which we call the 'exome'. Once a patient has been identified as eligible for an exome sequencing test, our initial discussions centre around the medical, psychosocial and insurance implications of having such a test. We discuss what 'targeted' exome sequencing is and the possible results and implications, both for the person being tested and for their family, in order to ensure they are making an informed choice about whether or not they would like to participate in the research. When the results become available, we discuss the nature of the findings with the patient and whether or not the test was successful in providing a diagnosis and whether there are implications of the result for other family members. We also provide ongoing support throughout the process.

HOW WILL GENOMICS HELP PATIENTS IN VICTORIA?

Exome sequencing may allow more individuals with genetic conditions to achieve a specific diagnosis. It can be very difficult to live with symptoms but without a diagnosis. Some individuals spend years on what has become known as the 'diagnostic odyssey'. Also, treatment or management options might become available once a genetic diagnosis is made, or the person may be eligible for other research projects in the future. Confirming a diagnosis genetically can sometimes allow other family members to access genetic testing. Through the work of the Melbourne Genomics Health Alliance and its 10 members, we hope to understand what it is like for patients and families to undergo exome sequencing. This information will be used to guide health professionals in offering exome sequencing as a clinical test in the future. To aid in this learning process, we ask participants in Alliance research projects to complete surveys or participate in focus groups so we can learn about their experience.

WHICH PATIENTS WILL BENEFIT?

The Melbourne Genomics Health Alliance is recruiting participants from The Royal Melbourne Hospital, The Royal Children's Hospital, The Peter MacCallum Cancer Centre, Austin Health and Monash Health. Genomic sequencing is currently being offered only to individuals with specific conditions, including health problems affecting the immune system, cardiac conditions, individuals with complex medical needs, severe congenital deafness and advanced cancer. Around 2,000 Victorians will receive access to genomic sequencing through Melbourne Genomics Health Alliance over the next four years. And many more Victorians will benefit in the long term from the Alliance's work in integrating genomics into everyday healthcare.

GLOSSARY

EXOME: a small portion (about 1%) of your genome that contains your genes.
GENOME: your complete set of genetic information.
SEQUENCING: reading the letters which make up your genetic code.
VARIANTS: changes in the genetic code.

VARIETY PROGRAMS

SMILE PROGRAM

Variety's Smile Program provides responsive financial and practical assistance and facilitating access to information, resources and services for the families and carers of children affected by a rare disease or condition.

Variety's Smile Medical Support Grant provides assistance to families with a child who has a rare disease or condition, to purchase minor items and services relating to their child's healthcare, over a 12 month period. Through this program Variety provides a grants of $1,000 for expenses relating to medical appointments, healthcare items, therapy, respite care, parking and fuel, and can be applied for once every 12 months.

The Case Management Service provides the service of a Case Manager for families affected by rare diseases and conditions. Variety's Smile Case Manager will be available to support families affected by rare disease, helping to find support networks and assistance (includes financial, emotional and physical support).

More info here: **www.variety.org.au/How-we-help/Variety-caring/Smile-Program**

CARING FOR KIDS PROGRAM

Through Variety's Caring for Kids Program, Variety helps parents care for their sick children at home by providing financial support for medical items and equipment.

More info here: **www.variety.org.au/nsw/how-we-help/individuals/caring-individual**

FUTURE KIDS PROGRAM

Through Variety's Future Kids Program, Variety helps children realise their potential by providing financial support and scholarships for education and communication needs.

More info here: **www.variety.org.au/nsw/how-we-help/individuals/future-individual**

SERVICES

FINANCIAL SUPPORT

FINANCIAL SUPPORT

THE STEVE WAUGH FOUNDATION

The SWF is committed to a co-ordinated approach to the service, identification, treatment and research of rare diseases to improve the quality of life of children and young adults affected by rare diseases (0-25 years of age).

SWF recognise due to resources and financial limits we will not be in a position to support all individuals, family and community needs in the area of rare diseases.

Our philosophy is to support the rarest diseases that are ineligible to get support from other sources.

GRANT ROUNDS – KEY DATES

The next round for applications for funding assistance is 1 September 2016 to 30 September 2016.

Please note: Applications for funding assistance may be submitted outside these dates. However, your application will not be reviewed until after the next grant round window closes, at which time all applications received in that round will be reviewed.

More info:
www.stevewaughfoundation.com.au/grants/

"What's that lab?"

Our regular segment, 'What's that lab?' aims to inform our members about the laboratory services of the Victorian Clinical Genetics Services (VCGS). In this edition Dr. Sebastian Lunke, the new Head of the Translational Genomics Unit (previously headed by Dr. Damien Bruno), tells us about the lab's role.

The Translational Genomics Unit's (TGU) main role is to develop and support genomic sequencing technologies for clinical testing and research projects. The genome is in effect the instruction manual for your body. The process of sequencing is like scanning through this instruction manual looking for spelling mistakes. Some spelling mistakes lead to disease because they change how the instructions are interpreted. Finding such a spelling mistake, helps us to more clearly understand the patient's disease, which may in turn lead to greater understanding of prognosis and possible treatments.

The lab consists of three main units:

1. The research genomics service – works closely with the MCRI researchers to explore cutting edge genomics technologies and make them available to the research community.
2. The clinical exome service – one of the first units offering NATA accredited clinical whole exome sequencing in Australia.
3. The clinical development unit – works on the development and optimisation of new clinical tests and technologies, and facilitates their translation from research use into clinical practice.

The interplay between these three units and the close integration with MCRI allows us to be at the forefront of genomic technology and clinical test development. Our current flagship test, clinical exome sequencing, has recently been made accessible to local and interstate specialists in genetic services. Deploying clinical exome sequencing has led to a substantial increase in the diagnosis of previously unresolved cases. It may interest you, however, that the success of this approach is not only driven by technological advances. The bedrock of the clinical exome service are our laboratory scientists who generate and analyse the variant data, and the clinical geneticists and genetic counsellors at VCGS who work closely with the laboratory scientists to interpret the genomic data. This close integration between laboratory and clinic creates a highly skilled multidisciplinary team, ideally set up to investigate even the most complex syndromic cases. Despite such exciting new technology and the strength of our team, currently around half to two-thirds of cases remain unresolved. Even though we are looking at effectively every single gene of a patient, the whole exome approach can read only around one percent of the manual that is our genome. While this one percent arguably contains the most important information, many of the unresolved cases will have mistakes somewhere in the rest of the genome, which we currently cannot look at. This is why we continuously strive to improve existing tests, as well as to explore new technologies.

This includes expansions to the scope of variation we can assess, whole genome sequencing, which will allow us to read the whole manual, but also technologies that allow us to better understand the impact of changes in these less commonly assessed regions of the genome. These latter approaches, called functional validation studies, will allow us to better understand the variation we find and to decrease the number of variants of uncertain significance, all with the aim to make the technologies more accessible and to further increase the diagnostic rate.

GLOSSARY

EXOME: a small portion (about 1%) of your genome that contains your genes.
GENOME: your complete set of genetic information.
NATA ACCREDITED: NATA stands for National Association of Testing Authorities. It is a nationally recognised accreditation which sets out the requirements for the competence of testing and inspection laboratories, thereby allowing companies and consumers to identify reliable service providers.
SEQUENCING: reading the letters which make up your genetic code.
VARIANTS: changes in the genetic code.

SCOPE BETTER START EARLY DAYS WORKSHOP

Still finding your way in the disability system? Is your child eligible for $12,000 in funding to assist with maximising their potential through therapy?

The free Better Start Early Days Workshops will help you understand the funding and services available for young children with a disability.

Who is eligible to receive support under Better Start? Eligible children are aged under six years and have been diagnosed with Cerebral palsy, Down syndrome, Fragile X syndrome, moderate or greater hearing or vision impairments, including deafblindness, Prader Willi, Williams, Angelmen, Kabuki, Smith-Magenis, CHARGE, Cornelia de Lange, Cri du Chat syndromes, Microcephaly or Rett's Disorder.

See: **www.scopevic.org.au/news-event/better-start-workshops**

Disability Support for Women and Children with Disabilities Escaping Family Violence

Woman's Domestic Violence Crisis Service

This ground-breaking program for women and children with disabilities has passed its pilot stage and will receive ongoing funding.

This is a Statewide initiative that assists Victorian women and children with disabilities experiencing family violence who require immediate disability support.

Funding is available to meet immediate disability-related support needs for a period of up to 12 weeks to a maximum value of $9,000 per person. Requests for funds over $9,000 will be managed on a case by case basis.

To access the initiative the woman or her child must:

- Have been assessed through the Common Risk Assessment Framework (CRAF) as requiring 'immediate protection' and be supported by and referred by a Specialist Family Violence Service
- Have a disability as defined by the Disability Act 2006.
- Require specific disability-related support to either access a family violence crisis accommodation response or remain safely in her home or community.

The short-term crisis funding can be used for the following purposes:

- Attendant care support for disability related needs such as personal care, shopping assistance, meal preparation or support in providing care of children.
- Hire of equipment (where own equipment cannot be accessed) or linkage with the State-wide Equipment Program where appropriate
- Sign/Auslan interpreting in cases where the DHS Interpreter service is not available through the credit line
- Transport costs related to disability

The program is designed to complement existing specialist family violence services and supports.

For more information during business hours, contact Disability Family Violence Liaison Officer:
T: (03) 9843 6312
E: **disabilityfv@dhs.vic.gov.au**

For information after hours, contact Woman's Domestic Violence Crisis Service:
T: (03) 9322 3555 or 1800 015 188
(toll free for country callers)

BRAINWAVE

Brainwave aims to support the patient with paediatric brain illnesses and injuries, and their family in the transition from hospital to home.

Then there is the ongoing rehabilitation process – which can often be months or years. This support can be in a variety of forms; financial support, support equipment for home care, purchase of rehabilitation equipment, house modifications to suit wheelchairs and other rehabilitation needs, funding for on-going physio and speech therapy, whatever the special needs of a particular family may be in order to manage their child and his/her illness.

Website: **brainwave.org.au**
Email: **enquiries@brainwave.org.au**

TOP-UP FUND FOR CHILDREN

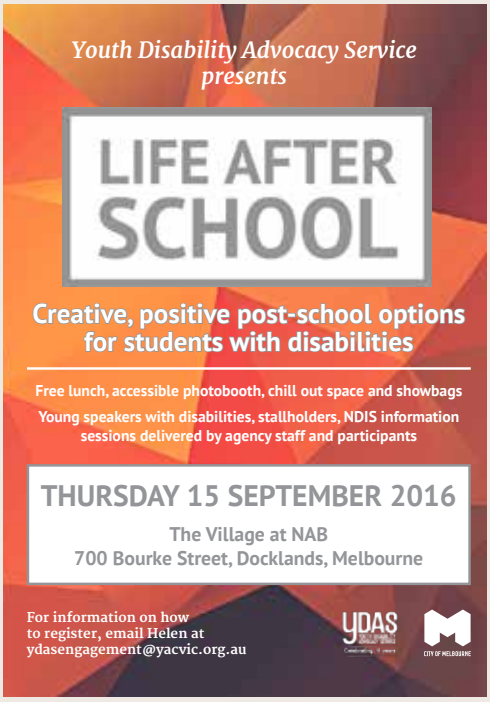
The Top-up Fund for Children provides families and children with funding to meet the difference between existing subsidies and the full cost of eligible mobility equipment, including manual & powered wheelchairs, pressure cushions and walking aids.

Website: **swep.bhs.org.au/top-up-fund-for-children**
Email: **swep@bhs.org.au**

FAMILY INFORMATION PORTAL

An online portal developed by consumers in partnership with the RCH and the Vic Paediatric Palliative Care Program, to help families find equipment, funding, support and assistance.

These links are relevant to most families caring for children with life altering illnesses and chronic conditions. Support groups are encouraged to submit links to further resources they know of for inclusion via the Feedback button at the bottom of the landing page. This way we'll be able to build up a really robust resource.
Website: **goo.gl/7yRpAG**



IN BRIEF

SEEKING CONTACT

The GSNV strives to connect individuals and families with others who have shared similar experiences.

If you would like to make contact and share your experiences, please contact the GSNV office by phoning (03) 8341 6315 or by emailing **info@gsnv.org.au**

Disclaimer: *The GSNV works to support contact between individuals and families to share experiences. However, in individual cases there may be differences in approach and opinion. Although the GSNV strives to make thoughtful and appropriate connections, those placed in contact are alone responsible for the views and opinions shared.*

ACKNOWLEDGEMENTS

TThe GSNV is proudly supported by a grant made possible by the Department of Heath, Victoria. The GSNV thanks the Murdoch Children's Research Institute and the Victorian Clinical Genetics Services for ongoing collaboration and support of our work. The GSNV thanks its financial members and acknowledges the kind donations.

EVENTS

JULY – FRAGILE X AWARENESS MONTH	
24 July	Australian X & Y Spectrum Support Seminar
28 July – 3 August	Hepatitis Awareness Week
28 July	World Hepatitis Day
30 July	Cystic Fibrosis Victoria Community Conference

AUGUST	
22 – 28 August	VCFS 22q11.2 Awareness Week
26 August	Daffodil Day
27 August	NF1 Paediatric Seminar

SEPTEMBER	
1– 2 September	Strengthening Disability Advocacy Conference
7 – 8 September	National Paediatric Bioethics Conference
5– 9 September	Women's Health Week
15 September	World Lymphoma Awareness Day
19 – 25 September	Global Mitochondrial Disease Awareness Week
21 September	World Alzheimer's Day
23 – 24 September	First Asia-Pacific Bone Disorders Symposium
25 September	International Ataxia Awareness Day
28 September – 2 October	MDDA Family Retreat and Teens Camp
30 September – 2 October	Australasian CHARGE Syndrome Conference

OCTOBER – RETT SYNDROME AWARENESS MONTH AND DWARFISM AWARENESS MONTH	
5 October	World Cerebral Palsy Day
7 – 8 October	Biennial Angelman Syndrome Association Australia Conference
9 – 15 October	Haemophilia Awareness Week
23 October	CAHSGA Family Conference Day

NOVEMBER – ALPHA-1 AWARENESS MONTH AND MUSCULAR DYSTROPHY AWARENESS MONTH