genetic support network of victoria

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WINTER 2015

Fragile X: It's not just about the child

Fragile X – associated disorders can affect multiple people in the family in multiple different ways.

By Katrina Weir, Medical Community Communications Officer Fragile X Association of Australia

When a child is diagnosed with Fragile X syndrome, there are ramifications for the immediate and extended family members, all of which should be tested to determine if they carry the affected FMR1 gene (Fragile X Mental Retardation 1), or not.

Within the last 15 years, researchers have identified two conditions that can affect adult men and women who are ***carriers*** of the affected Fragile X gene. This means that there are three separate conditions that can result from a mutation to the Fragile X gene and collectively, these are known as Fragile X – associated disorders.

There are three separate conditions that can result from a mutation to the Fragile X gene, these are:

- Fragile X syndrome;
- Fragile X – associated Tremor Ataxia syndrome (FXTAS), and
- Fragile X – associated Primary Ovarian Insufficiency syndrome (FXPOI).

Cont. page 9

See page 8 for Hayley Reed's Personal Story

CONTENTS

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genetic support network of victoria

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Committee Meeting Dates

July 16
August 20
September 17
October 15
November 19
December 17

GSNV

Message from the Committee

Welcome to the first issue of the GSNV newsletter for 2015. I hope everyone had a good holiday break and start to the New Year. We have been busy in the first half and are looking forward to an exciting programme in the months ahead.

Our Annual General Meeting (AGM) was held on November 20 2014; we had low numbers in attendance but overall it was a good evening. The Committee were all re-nominated for the coming year, with a couple of vacancies still open, so if you would like to come along to a meeting and are interested in joining our committee please contact the office and they can let you know how to get involved and when and where our meetings are.

At the AGM we had two of our committee members present their research, in fulfilment of the University of Melbourne Master of Genetic Counselling course. Both were interesting – Rachel and Hannah, job well done! We congratulate both on their graduation and advise that Rachel has since obtained a position in New Zealand and Hannah has obtained a position in Adelaide. We are very proud of our graduate counsellors!

Further to our AGM the GSNV held a very productive Strategic Planning day in preparation for 2015. We have a varied committee at present with a number of different skills sets. All members on our committee are committed and active and have their work cut out for them in 2015.

We finished off 2014 with a meeting with the Department of Health Vic. The good news is the GSNV is assured of funding for the foreseeable future, but like most non-profits will need to be proactive is establishing effective cost recovery measures and budget efficiencies.

Moving forward, to making a difference, to help those that need help!
Best regards

Kay Timmins,
Chairperson 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: **www.gsnv.org.au/become-a-member.aspx**

Your Feedback

That looks fabulous! You do a beaut job with the layout. Thanks for making me a part of the GSNV Newsletter.
– Evelyn Bugel

GSNV

Message from the team

Dear members and the genetics community, In previous correspondence I have announced some exciting news regarding my role with the GSNV and my working arrangements in 2015. I am currently taking some time out and have relocated my life and work to Rimini which is a small city in the region of Emillia Romagna, Italy. This region is responsible for the famous dish spaghetti bolognese and piadina which many of you will know and love to eat.

As advised I will be in Italy until January 2016, or thereabouts. I have not disappeared however, I have my work with me and will continue to oversee many functions of the GSNV while abroad. I am however, attempting to stop and smell the pasta sauce and the challenge will be to maintain a good work/life balance while in Italy.

I currently have a number of projects on the way with support groups and will continue to progress these despite the challenges of Wi-Fi and internet access. Italy is not famous for its broadband network (it has none and relies on very slow ADSL) so I will conduct my work via email, Skype (online meeting forums) and telephone, when I get a line! Please bear this in mind if you do not receive a response to your email for some time. Also note that I am currently 8 hours behind you in Italian time. I am doing my best to keep on top of the time difference but do admit it's a challenge. The GSNV team will be available to the GSNV community and it will be business as usual as much as possible. Although I will be unable to physically meet with those requiring my assistance, I will be available remotely.

So what have I been doing since April 7? After a whirlwind departure from Melbourne and initial two weeks in Italy, it was a good month before I felt I had come back down to earth and settled into a sense of normality. As I have discovered, the life of an expatriate is somewhat challenging, particularly in terms of getting settled and re-establishing ones digital life in a new hemisphere. Between my laptop, iPhone, Mini iPad, Apple TV and hard drives I have had my share of frustration in trying to get all devices to sync and on a dependable network. It's been an interesting process indeed and still a work in progress.

After arriving in Italy April 7 I hit the ground running and was attending the 5th Alpha-1 Global Patient Congress and 2nd Biennial International Research Conference on Alpha-1 Antitrypsin in Barga, Lucca – Italy by April 9. The congress was superb and represented the very best of international cooperation, coordination and rare disease approach. I applaud the Alpha-1 Global Foundation for a very successful congress and for bringing together patient groups, the scientific/research community and clinicians in an open and cooperative environment. All stakeholders were given an opportunity to participate and contribute to an agenda that was interesting and engaging. It was apparent too from the very beginning of the congress that the Alpha-1 international community is strong, active, supportive and very well led.

I was very privileged to be part of the Alpha-1 Australian contingency and I thank Dr. Charlie Strange of the Medical University of South Carolina (MUSC) and the Alpha-1 Global Foundation for their support in getting me to beautiful Barga. Lucca. Steven Knowles of the Alpha-1 Association of Australia (AAA) was greatly missed but was absolutely with us in spirit. As a member of the Alpha-1 Global Steering Committee, Steven was integral to the organisation, and development of the congress. I also thank Steven for his vision in getting a few extra Australians to Italy (including me as a GSNV and Murdoch representative) and in facilitating an opportunity for key collaborators to meet and spend some time discussing future work.

The AAA was strongly represented by Jennifer Nankervis and John Arkinstall from Australia and it was wonderful to see the relationships they have developed with the international Alpha-1 community and the respect they receive from their peers. My experience at the congress was truly inspiring and in many ways, has set the scene for further opportunity, research and collaborations while I pursue my sabbatical (working) in Italy. I look forward to working with the Alpha-1 Australian community and progressing important projects with them.

Following the success of the Alpha-1 Global congress I had my feelers out and was keen to take up any immediate opportunities for learning and networking here in Italy. Upon invitation from some learned colleagues in the region of Emilia Romagna (Rimini) I was fortunate to attend the Regione Emilia-Romagna XXVIII Convegno – Anomolie Congenite Del Sistema Nervos Centrale. This one day workshop was facilitated by the Gruppo Di Studio Sulle Malformazioni Congenite (I.M.E.R) and included discussions on dysmorphology, syndrome diagnosis, and rare conditions of the central nervous system and case histories of disorders of the Corpus Callosum. With my very best Italian language understanding in progress, I was delighted to participate in the workshop and gain further understanding of the rare disease clinical and research environment here in Italy. With a long standing congenital malformations and anomalies of the central nervous system registry very active in this region, I was interested to read the I.M.E.R Annual Report and definitive data on such rare diseases in Italy. Two congresses in two weeks and a plethora of 'settling in issues to deal with' constituted a busy but wonderful start to my working life and very busy April.

This past month I have been determined to establish a good working routine and level of efficiency that is remotely like what I achieve at home… I give up, impossible! Italy is just not that sort of place where strict routine and 8 to 10 hours of continuous work is acceptable. It's much more balanced and I guess relaxed. Over here my friends and colleagues are shocked, horrified and appalled to learn we Australians have lunch at our desks at work and have no 'pause' in the middle of the day. This is unheard of in Italy. So in keeping up with the local culture I work half a day (well try very hard to with Wi-Fi and internet that doesn't cope at all with my email and internet demands) and then 'pause' for the obligatory lunch hour (its actually two) and downtime. Some days I do work through lunch and eat at my desk, but it does not go down well at all. There is a remarkable difference in the approach to

Cont. page 4

Message from the team Cont. from page 3

work and life in this part of the world and I'm learning that it's probably better and healthier overall. My fellow countrymen in Italy work to live… no confusion there. It does however reflect on efficiency but it is the norm to 'wait a while' for things to get done so I'm adjusting.

It is astounding that eight weeks has passed since I arrived and that May has now passed me. It's been yet another busy month with a workshop on international approaches to neonatal diagnosis and a number of meetings with academics from the University of Bologna under my belt. I am learning a great deal. A definitive highlight for the month was meeting up with Dr. Sue White of the Victorian Clinical Genetics Services in Bertiloro, Emilia Romagna. Very nice to meet an Australian and colleague for dinner.

I am very excited, for this opportunity to spend some time abroad, continue my work and learn some new things from our European counterparts. I will be regularly reporting from my desk in Rimini and will provide updates via the GSNV communications.

I advise that you can continue to contact me via my work email address: **louisa.dipietro@vcgs.org.au** and I will respond at the first opportunity. Please also feel free to contact the GSNV team directly or via **info@gsnv.org.au.** In my absence Keri Pereira, Nancy Amin and Anna Jarmolowicz will continue business as usual in the GSNV office. The GSNV executive and committee will also continue business as usual in enacting our strategic plan and representing Victorian consumers. The next 10 to 12 months will not be without challenge but overall represents a wonderful personal and professional development opportunity for me as group leader of the GSNV, and as an advocate for genetic and rare conditions. I look forward to returning to my "regular life" in 2016 with renewed creativity and vigour.

Please see here on the following pages, lots of information and resources that may be of use to you in your work, and continue to provide feedback and contributions in order that the GSNV newsletter remains relevant to all.

Louisa Di Pietro
Group Leader GSNV

IN FOCUS

Changing Perceptions on Cystic Fibrosis

No longer simply a fatal childhood disease

Up until the 1960s a child born with cystic fibrosis (CF) was not expected to survive infancy. By the 1970s and even 1980s it was hoped that they would reach their teens. By Karin Knoester, Chief Executive Officer, Cystic Fibrosis Victoria

With developments in understanding and management of this condition people with CF currently have an average life expectancy in Australia of around 40 years. Tragically there are still many who don't reach this age but there are also a growing number approaching 50, and occasionally 60, or even 70 years of age.

Thus communications and education on CF have needed to evolve. Today we work to debunk the idea of CF as simply a fatal childhood disease. Cystic Fibrosis Victoria and other CF organisations around Australia and overseas try to ensure that when a couple receives the devastating diagnosis that their newborn has CF, they understand that while the road ahead may be tough, there is still hope for a relatively normal life for their child.

Finding a suitable descriptive for CF is not easy. It is often referred to as a chronic illness or as a degenerative condition or, in more severe cases, as a disability. Not only does each of these terms have social, economic and even political connotations, none of them can accurately be applied to the entire CF population, as experiences of CF can differ significantly.

At times it is still appropriate to communicate the major impact that CF can have on the lives of young children. We sincerely hope for the greatest chance at normalcy for all kids with CF, but these children often need support, tolerance and understanding from those around them, so explaining the impact of CF can be extremely useful.

It is also a reality that the concept of sick children can evoke a greater level of sympathy, than an unwell adult might. This is especially true when fundraising or dealing with the media.

Securing community support and understanding of CF is probably more important than ever. As the CF population ages, new challenges and health issues are emerging. Adults with CF need to juggle their health with employment. Many with CF want and are starting their own families, but reproduction is often complicated for people with CF, especially men who are usually infertile. CF also has a number of comorbidities or related illness that emerge with age. Most people with CF develop CF related diabetes. There is also an increased risk of osteoporosis and some cancers, not to mention their deteriorating lung function and susceptibility to infection.

Cystic Fibrosis Victoria seeks to assist those affected by CF to be well and live fuller lives. To do this we need to communicate the complexity of cystic fibrosis and create understanding that there is no "one size fits all" label for CF. To generate awareness and understanding of something so vast and variety requires targeted communication and flexibility.

Cystic Fibrosis screening update

By Alex Birnberg, Project Coordinator, Programs and Support Services, Cystic Fibrosis Victoria

It is a startling statistic that 95% of children born with cystic fibrosis (CF) the family had no prior history of CF. This is because there are no symptoms associated with being a carrier of CF. With the generosity of the Rotary Club of Balwyn, Cystic Fibrosis Victoria has embarked on an awareness campaign to make sure people know their options when it comes to carrier screening. CF carrier screening is a way for individuals to find out if they carry a CF causing gene change, and if they are at risk of having a child affected by the condition.

If two carriers of a CF causing gene change have a child, there is a 25% chance that the child will be affected by CF and a 50% chance that the child will be a carrier of the condition. It is estimated that 1 in 25 Australians of Caucasian ancestry are carriers of the gene change that causes Cystic Fibrosis.

Carrier screening requires a pathology request from your doctor and a blood or saliva sample, depending on the testing provider. Carrier screening for cystic fibrosis is not currently funded by Medicare.

Since the Summer 2014 issue, the campaign has raised awareness through Facebook advertising, newspaper advertising and through Avant cards. 16,000 Avant cards were distributed through-out Victoria's many cafes and theatres. This was an important first step and the hope is to now build on this momentum.

For further information on carrier screening online please see **www.cysticfibrosis.org.au/vic/carrier-screening** or contact Cystic Fibrosis Victoria on **(03) 9686 1811.**

IN FOCUS

Three parent IVF legalised in Britain

In the GSNV Newsletter Summer 2014 edition we introduced the topic of three parent IVF. At that time its implementation was being considered by regulatory authorities in the US and UK.

Mitochondria are tiny organelles with a small amount of their own DNA (representing 0.1% of total cellular DNA and are inherited solely from the mother) and serve as the 'powerhouse' of cells. Mitochondrial diseases vary in severity, but in many cases individuals who have a mitochondrial disease live with debilitating illness caused by multiple organ dysfunction and have a reduced life span. One Australian child born every week will develop a severe form of the disease, for which there are currently very few effective treatments.

Three parent IVF or mitochondrial replacement is a technology that has the potential to help women who are affected with severe mitochondrial diseases to conceive healthy children. This technology involves the collection of eggs from both an unaffected egg donor and from a mother who has a genetic fault in her mitochondrial DNA. The mitochondria from the mother's egg are then 'swapped-out' with the unaffected mitochondria from the donor egg. The resulting egg will have healthy donor mitochondria and the mother's nuclear DNA. This egg could then be implanted and fertilised using IVF techniques. These techniques therefore prevent any disease caused by faults in mitochondrial DNA from being passed on to the next generation.

In Britain 2,500 women are at risk of having a child with a potentially devastating mitochondrial disease. In February of this year British Members of Parliament voted to legalise this technology and it was approved by the House of Lords a few weeks later. This means that the first such baby could be born next year, with suggestions that approximately 150 couples in Britain, could use this technology each year.

This British decision has been applauded by the Chief executive of the Australian Mitochondrial Disease Foundation, Sean Murray. He urges the Australian Government to reconsider its ban on the creation of embryos using mitochondrial replacement and estimates that about 60 women a year in Australia may be interested in the technique if it was made available to them.

Critics however, have pointed out that there have not been clinical trials that have conclusively shown the long term safety of these treatments in humans. For this reason, the Cellular, Tissue and Gene Therapies Advisory Committee of the U.S. Food and Drug Administration (FDA) have concluded that they will withhold their decision to legalise mitochondrial transfer techniques until further research has been conducted in humans.

Current Australian law seemingly leaves the option open for three parent IVF. The law does prohibit the production of embryos this way, unless authorised by a licence. See the 'Prohibition of Human Cloning for Reproduction 2002' Act under Part 2, division 2: "Practices that are prohibited unless authorised by a licence" 23 Offence — creating or developing a human embryo containing genetic material provided by more than 2 persons. A person commits an offence if:

(a) the person intentionally creates or develops a human embryo by a process other than the fertilisation of a human egg by a human sperm; and
(b) the human embryo contains genetic material provided by more than 2 persons; and
(c) the creation or development of the human embryo by the person is not authorised by a licence

Source: http://www.comlaw.gov.au/Details/C2008C00694

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Experts support a moratorium on CRISPR gene editing technology in babies

Scientists around the world are currently conducting experiments in animal and human cell models using a new gene editing technology called CRISPR to help reverse the gene changes that are inherited in various genetic conditions.

However, a consortium of scientists who have spearheaded and are currently using this technology in their research aimed at helping cure inherited diseases, have issued a warning. In an article recently published in the journal Science ("A prudent path forward for genomic engineering and germline modification"), the scientists advocate for postponing the use of this technology in human embryos or the germline (a woman's egg, or a man's sperm) pending public consultation. They warn that such changes can be inherited in future generations, the effects of which are currently unknown.

What is CRISPR?

CRISPR is a genome engineering method known as Clustered Regularly Interspaced Short Palindromic Repeats. CRISPR are naturally occurring short repeating DNA sequences in the genetic makeup of some types of bacteria that protects them from bacterial viruses. These repeat sequences match the invading viral sequences and work as a type of immune system. This system allows the bacteria to recognise foreign DNA and inactivate it by dicing it up into small pieces.

Researchers have harnessed this system to edit genes in organisms such as fruit flies, mice as well as in human cell culture, for research involving models of human diseases. Scientist can easily design molecules that recognise the DNA sequence that they wish to edit using the CRISPR system. The molecular machinery is then targeted to the gene of interest and can silence it or change its sequence. This type of gene editing is akin to editing a sentence with a word processor to delete extra nonsensical words or to correct spelling mistakes.

Video introduction to genome editing using CRISPR: **vimeo.com/106957770**

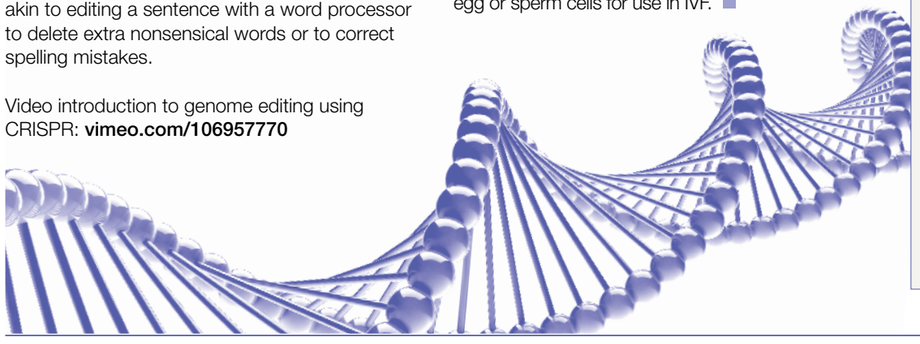
CRISPR for adults

One application of this technology is to treat human genetic diseases in adults. The faulty or mutant form of the gene can be replaced with the correct sequence. Researchers recently used this technology to 'cure' a mouse model of the human diseases hereditary tyrosinemia (a rare liver disease) and Duchenne muscular dystrophy as well as β–thalassemia in human cells.

It is envisioned that this kind of genome editing could one day help treat a variety of human diseases in adults. Somatic cell (cells in the body) modification hasn't raised deep ethical concerns as these changes cannot be passed on to future generations. The real controversy begins when there is the potential for genetic changes to be passed on to the next generation, an ethical line that many believe should not be crossed. There are suspicions that scientists in the US, UK, and China have already started conducting experiments involving editing human germline cells.

Editing the germline

A variety of ways has been proposed to edit germline cells. The CRISPR molecular machinery could be injected into an early stage embryo using in vitro fertilisation (IVF). In this scenario, CRISPR's inherent repair inefficiency may lead to incomplete expression of the gene change in all of the embryo's cells. Alternatively, individual egg cells could be injected prior to IVF. Others have suggested that stem cells could be edited and then turn them into egg or sperm cells for use in IVF. ■



Current Australian law prohibits germline modification, under the 'Prohibition of Human Cloning for Reproduction 2002' Act under Part 2: 15 Offence—heritable alterations to genome

(1) A person commits an offence if:

(a) the person alters the genome of a human cell in such a way that the alteration is heritable by descendants of the human whose cell was altered; and

(b) in altering the genome, the person intended the alteration to be heritable by descendants of the human whose cell was altered. Maximum penalty: Imprisonment for 15 years.

(2) In this section: ***human cell*** includes a human embryonal cell, a human fetal cell, human sperm or a human egg.

Source: http://www.comlaw.gov.au/Details/C2008C00694

ARGUMENTS AGAINST GERMLINE MODIFICATION	ARGUMENTS FOR GERMLINE MODIFICATION
This technology could lead mankind back into another era of eugenics. Instead of limiting modifications to cure disease, people could be tempted to edit in enhanced versions of genes to improve traits such as intelligence and appearance (i.e. designer babies).	Just because a technology is available won't necessarily mean it will be misused. Continued ethical debate and regulation can avoid misuse.
We already have a technique called pre-implantation genetic diagnosis (PGD) that can help avoid passing on genetic disorders. This involves the creation of a number of embryos by IVF and subsequent genetic diagnosis. The healthiest embryos that do not carry the mutation are then selected for implantation.	Has the potential to help couples that are unable to conceive through IVF due to the inability to find a suitable embryo (e.g. couples that carry multiple gene faults) or due to inherited causes of infertility.
CRISPR repair efficiency is not currently very high. Therefore many embryos still need to be injected and screened to determine which of them are repaired. This means that unrepaired embryos would still be discarded. Stem cell conversion into viable germline cells is not currently efficient enough to be feasible (and brings its own ethical concerns).	Embryos that carry the faulty gene are discarded after PGD. Gene editing stem cells or germline cells, before an embryo is created, would avoid the need to overproduce and discard some embryos.
The unborn child is unable to choose for themself whether or not to have the treatment.	You are giving your child the best possible start by correcting known genetic errors.
The long term effects to the child are not known. At the moment this technology is prone to off-target effects. This means that other unrelated genes maybe unintentionally modified, potentially causing new diseases. On-target effects are also a concern. Changing a gene in isolation may have unintended effects.	There is continual advancement in strategies to monitor and reduce off-target effects.
This would affect future generations. The edited gene can be passed on when the child reaches adulthood and has their own family.	

RESEARCH UPDATE

Rumours that gene editing has been trialled in human embryos were confirmed on the 18 of April with the publication of a study that describes the use of CRISPR to repair the β–thalassaemia gene change.

In this study researchers used human IVF embryos obtained from fertility clinics that would normally be discarded, in order to alleviate ethical concerns about experimentation with embryos. The embryos used, had an extra set of every chromosome and therefore could not lead to a live birth.

The results of this study highlighted known problems with CRISPR inefficiency and off-target effects. The scientists injected 86 embryos and of those, 71 survived. Of the embryos tested, the successful gene repair rate was only 7%. In addition, all of the gene edited embryos were mosaic i.e. not all of the cells in each of the embryos had been correctly repaired. This means that PGD cannot be used to select for 'healthy' embryos because PGD involves sampling and testing only a small number of cells from the embryo. Significant off-target effects (unintended mutations that could be harmful) were also observed in all of the embryos. This study highlights the infancy of this technology and the need for further research before its clinical use can be considered.

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IN FOCUS

Hayley Reed with children Tylah (13 years), Levi (10) and Zharia (4)

FRAGILE X: *Personal story*

Hayley Reed is a Victorian mother of three children, all of whom are impacted by Fragile X syndrome. She hasn't let this diagnosis alter her positive outlook.

"Don't let Fragile X rule your life," advises Hayley to other families living with the condition."Enjoying your kids is more important than anything else and sometimes the stress and anxieties of life mean we lose sight of the joy that they can bring." Tylah Reed is now 13 years old. He was diagnosed with full mutation Fragile X syndrome when he was three years old after day care staff noticed issues with his verbal communication. He also had sensory defensiveness, developmental delays, disturbed sleep and recurrent ear infections. He was referred to a paediatrician who completed a battery of testing; the DNA test for Fragile X syndrome was positive. He has 200 CGG repeats.

Hayley was advised that due to Tylah's diagnosis, she had a 50% chance of any subsequent children also having Fragile X syndrome. She was also told that her twin brother, father, aunts and uncles could be carriers and that genetic testing was advised. Hayley was already pregnant with her second son Levi, when Tylah was diagnosed with Fragile X syndrome. Given the family history of Fragile X, Levi was tested at 18 months and results showed that he is a carrier of the Fragile X gene with 130 CGG repeats. Testing of a third child, Zharia, showed that she is also fully affected by Fragile X with mosaic CGG repeats between 325 – 725. She too was 18 months when diagnosed.

Following Tylah's diagnosis, Hayley and several members of her extended family were tested and the results were mixed. Hayley's twin brother's results were negative but both Hayley (63 CGG repeats) and her father (100 CGG repeats) are carriers of the Fragile X gene. Hayley's father has since developed FXTAS. Hayley's father was formally diagnosed with FXTAS at age 55 although he was diagnosed with 'chemical imbalances' at age 45. She says he has always been impulsive and angers very easily. He is now 65 and in the past decade, his symptoms have become more pronounced and difficult for him to manage. He has great difficulty controlling his impulses, is inconsolable during an angry outburst and his concentration has significantly deteriorated.

Hayley says that early intervention has not been that effective for her two boys and they now both attend a special school. Her daughter attends day care/kindergarten and has made some progress since being involved in an early intervention program. The family has recently been appointed a new case worker from the Victorian Department of Human Services who is providing valuable practical assistance such as liaising with schools, identifying respite services and investigating more affordable housing. According to Hayley, he has been a 'godsend'. Hayley's energy and commitment to helping her children live with Fragile X is matched only by her interest in helping others who are in a similar situation. She has started a Facebook group called FragileXplained to increase interaction and support among families and now has members from around Australia and the world.

Hayley also recommends a poem by Emily Pearl Kingsley titled 'Welcome to Holland' **(www.our-kids.org/archives/Holland.html)** which she says has really helped her understand and manage her situation. ■

About FRAGILE X Association of Australia

The FXAA is a national non-profit member organisation that provides services and support for children and adults affected by Fragile X – associated disorders, and their families and carers.

FXAA provide free specialist counselling, educational workshops, access to assessment clinics, and current information resources. FXAA also promote community awareness of Fragile X. For further information, visit **www.fragilex.org.au** or call **1300 394 636.**

The Victorian Clinical Genetics Services (VCGS) **(vcgs.org.au)** offers Fragile X syndrome carrier screening as part of the Reproductive Genetic Carrier Screen which also tests for spinal muscular atrophy and cystic fibrosis carrier status.

This carrier screening panel costs $385 and is also offered through private practice obstetricians or general practitioners.

For more info see: **http://goo.gl/6AIfnn** ■

IN FOCUS

Fragile X: It's not just about the child

Cont. from page 1

Fragile X syndrome

Fragile X syndrome is the most common known inherited cause of intellectual disability and the most common known single gene cause of autism spectrum disorder. It affects 1 in 3600 males and between 1 in 4000-6000 females, however, **the prevalence of carriers is considerably higher with 1 in 800 males and 1 in 170 females being carriers of the Fragile X gene (FMR1).** This means they are in the pre-mutation range and have the potential to produce a child with Fragile X syndrome.

Fragile X syndrome occurs in both males and females but males are diagnosed more frequently and generally appear more severely affected than females. Fragile X syndrome is a lifelong condition that causes intellectual disability, speech and language delay, and a number of behavioural and emotional problems such as anxiety and aggression. The 'normal' form of the FMR1 gene has between 6 and 44 copies of a repeated sequence at the beginning of it.

Fragile X pre-mutation carriers: People with the Fragile X pre-mutation carry an altered version of the FMR1 gene, where the repeated sequence has expanded to between 55 to 200 repeats. People with a pre-mutation may develop a Fragile X associated condition.

Fragile X full mutation: People with the Fragile X full mutation have more than 200 repeats in the FMR1 gene and may develop Fragile X syndrome.

Fragile X Tremor Ataxia Syndrome (FXTAS)

Discovered in 2001, FXTAS is a progressive neurological condition with a presentation similar to Parkinson's disease.

It predominantly affects male pre-mutation carriers. The risk of FXTAS increases with age and in males, is **approximately 50% by age 79.**

There is no cure for FXTAS although some medications can help control the symptoms which may include tremors, balance problems, numbness in the extremities, irritability and other changes in personality and gradual intellectual decline (see Hayley Reed's story page 8). **People presenting with symptoms of FXTAS should be referred to a neurologist, regardless of family history of Fragile X syndrome.**

Fragile X Premature Ovarian Insufficiency Syndrome (FXPOI)

FXPOI affects **approximately 20% of female pre-mutation carriers** and causes premature ovarian failure leading to irregular periods, early menopause and usually, infertility.

All women, regardless of family history of Fragile X syndrome, who experience early menopause before the age of 30, should have a blood test for Fragile X and chromosomal assessment. ■

RESOURCES

Do you suffer from a mitochondrial disease (mito)? Please join the Mito Registry!

The Mito Registry, the first of its kind in Australia, will collect limited information about people suffering from mito to track a natural history of mito outcomes. It will ensure patients have the earliest possible opportunity to enter any relevant study or clinical trial and will aid diagnosis.

The Australian Mitochondrial Disease Foundation hope to build a registry of all mito sufferers across the country.
www.amdf.org.au/mitoregistry

Global registry for Multiple Sclerosis

NARCOMS (North American Research Committee on Multiple Sclerosis), is looking to capture the real-life experiences of people living with MS.

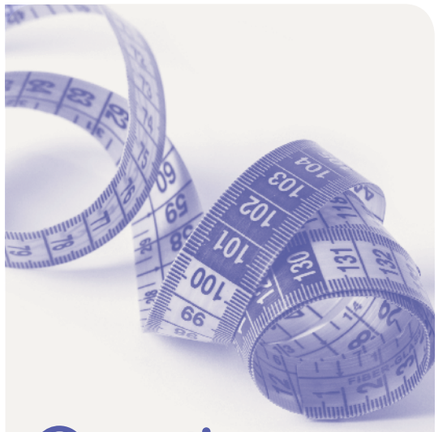
Find out more: **www.nationalmssociety.org/About-the-Society/News/NARCOMS-MS-Research-Effort-Seeks-Participants-MS-R**

Neurofibromatosis Type 1 (NF1) Patient Registry

The NF1 Patient Registry Initiative (NPRI) team are studying the risk factors for childhood brain tumors in individuals with NF1. The NPRI is currently recruiting both adults and children with NF1 who either (1) HAVE previously been diagnosed with a brain tumor younger than 18 years or (2) HAVE NEVER had a brain tumor.
nf1registry.wustl.edu

The Alpha -1 International Registry (AIR)

AIR is an invaluable source of information for medical researchers and others interested in working towards a cure for Alpha-1 Antitrypsin Deficiency. This register requires the involvement of your respiratory physician.
www.aatregistry.org ■



Genetics of anorexia nervosa study

Past and present sufferers of anorexia nervosa are encouraged to take part in a worldwide study aimed at finding a cure.

The project wants blood sample results from 13,000 women and men who are suffering, or have suffered, from the disorder in order to understand the genetic factors involved, which can then inform treatment and prevention.

For more info: **www.abc.net.au/news/2015-03-31/anorexia-27survivor27-urges-past-and-present-sufferers-to-tak/6361554**
To take part: **angi.qimr.edu.au**

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.

RESEARCH

Parental Support Needs Survey Needs Fathers!

Lemuel Pelentsov is undertaking a PhD investigating the supportive care needs of parents caring for a child with a rare disease. He is also a parent of a child diagnosed with a rare disease.

If you are a parent, mother or father, of a child with a rare disease that is 18 years or under, you are invited to complete a short survey aimed at identifying your supportive care needs. The purpose of this research is to better understand the support needs of parents caring for a child diagnosed with a rare disease (irrespective of what disease your child has), and to develop a tool for use by health professionals to assist them in identifying parental support needs. The information obtained should eventually lead to more appropriate individualised supportive care for parents.

Parents of children with a rare disease play a vital role in the daily lives of their children with a rare disease. They carry significant daily responsibilities of care. Yet, very little is actually known about their supportive care needs.

My research aims to give parents of children with rare diseases an opportunity to have their voices heard and their needs recognized with the ultimate goal to improve the way health providers identify needs, tailor support and plan and implement services within the rare disease community.

So far, over 150 completed surveys have been received. Of these, majority (96%) have been mothers. So, fathers are strongly encouraged to also complete the survey, so that this research can represent the support needs of both mothers and fathers.

Your time in completing this short survey is very much appreciated. The following link will take you directly to the survey: **www.surveymonkey.com/s/3NYKPH6**. It will take about 20 minutes of your time to complete.

ALS Quest

ALS Quest is a research study being conducted by Dr. Roger Pamphlett of the University of Sydney to look into possible causes of Amyotrophic Lateral Sclerosis/Motor Neuron Disease (ALS/MND).

The format of the research study is an anonymous online questionnaire, which has been set up to gather a wide range of information that can be compared between people who have, and those who do not have, ALS/MND.

We hope this information will prove useful in searches for preventative measures and treatments of ALS/MND. Both people with and without ALS/MND are eligible to complete the questionnaire. To begin the questionnaire follow this link: **www.alsquest.org**

RESEARCH

Survey for Australians born with congenital variations in sex characteristics

University of New England researchers are conducting a survey of Australians born with congenital variations in sex characteristics (Intersex/DSD/hormonal, chromosomal or other biological variations/conditions).

Some terms with which you might have heard used to describe these variations are: Intersex, DSD/Disorder of Sex Development/Diverse Sex Development, atypical reproductive or sex development condition or variation whether hormonal, chromosomal, or otherwise biological (e.g. CAIS, Klinefelters, Turners, PCOS and many others).

This project has been developed under the advice of a Reference Group including Morgan Carpenter of Organisation Intersex International (OII) Australia; Bonnie Hart of The Androgen Insensitivity Syndrome (AIS) Support Group Australia, and Dr. Gávi Ansara of the National LGBTI Health Alliance.

If you are interested in sharing your experiences of health, education and social supports, please see the survey's first page which goes into detail about the survey and what it involves, and how this data will be used.

See: **www.surveymonkey.com/s/ausvariations**

Carers – Your feedback is needed

Carers are invited to take part in a survey to further understanding of which services you have found most useful in supporting you in your caring role.

This information will be summarised and placed on a website for GPs called "HealthPathways Melbourne".

To participate: **www.surveymonkey.com/r/RJ8XVVC**

SMA clinical trial

A world-wide clinical trial for infants who have been genetically diagnosed with SMA but are currently pre-symptomatic.

For more information: **www.smatrust.org/sma-clinical-trial-biogen-idec-and-isis-pharmaceuticals-announce-the-launch-of-nurture/**

Call for adult participants – Genetic basis of stuttering project

The Murdoch Childrens Research Institute and The Royal Children's Hospital are conducting a project to help understand the relationship between genes and stuttering.

Who can take part?

We are looking for adults aged 18 years and above who have never stuttered. We are interested in people who use English as their primary language and who have not been diagnosed with a neurological disorder.

What is involved?

In order to participate, we will ask you to complete the following:

- Have a five minute conversation with one of our researchers.
- Provide a sample of your saliva so that we can study your DNA to see whether there are common genes that influence the risk of stuttering.
- Complete a short question about your birth, developmental and medical history and history of communication problems in your family. Complete a short questionnaire that measures how anxious and/or stressed you might be feeling.

You can contact us over the phone to arrange for us to give you a participation pack as well as a reply paid envelope for you to return the questionnaires and your saliva sample. Our research team will organise a time to meet with you in person or over the phone to record your speech sample.

If you would like further information about the project, please contact:

Peta Newell
Murdoch Childrens Research Institute
Phone: (03) 9345 4752
Email: peta.newell@mcri.edu.au

Spinocerebellar Ataxia Australia Group

By Marie Dunn. Secretary, Spinocerebellar Ataxia Australia Inc.

The Spinocerebellar Ataxia Australia Group recently held a meeting for members and guests on Sunday 15 March at the Neurological Conference Room, at The Alfred, Melbourne.

Guest speakers were Professor Elsdon Storey on 'Genetics' and Dr. David Szmulewicz from the Royal Victorian Eye and Ear Hospital Melbourne and Tamar Black from the Temporal Bone Donor Society on 'Temporal Bone Bank Donations.'

SCAA Newsletter Summer edition is available on the website **scars.org.au.** Our thanks to the editor Alice Poulton and the Royal Children's Hospital for assistance with this production.

The SCAA group's Fundraising and Awareness Committee have been very busy holding sausage sizzles at Officeworks Werribee and Highpoint Shopping Centre during the Australia Day long weekend and the following Saturday in January. All proceeds go to the SCAA research fund.

The SCAA group said goodbye to a much loved and long time member Melissa Gordon who attended the first meeting of the group at the old Royal Children's Hospital in December 2007 and was interviewed by Jenny Brockie on the Insight program "In your Genes' SBS One 13 October 2009. Sadly Melissa left us on 2 January 2015 at the age of 30 years.

Fond regards and thanks ■

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 2015, dates to be advised.

In 2014 the following groups received a small grant from the GSNV:

- **Alpha-1 Association of Victoria (AAA) –** $200, A shopping cart for their website to enable AAA to advertise and sell awareness raising materials.
- **Australian Disorders of the Corpus Callosum –** $200. AusDoCC meet up 2015.
- **Australian Leukodystrophy Support Group Inc. –** $200. Victorian Family Day 2015.
- **Dialysis and Transplant Association of Victoria Inc –** $100. Membership subsidies.
- **HAE Australasia –** $250. Online membership registration and multiuser database.
- **Metabolic Dietary Disorders Association –** $350. Peer cooking demonstration venue hire, commercial kitchen, ingredients and take away container costs.
- **Down Syndrome Victoria (DSV) –** $200. Purchase of book on how to breast feed a child with down syndrome for inclusion in a parent pack.
- **Mucopolysaccharide and Related Diseases Society Aust. Ltd –** $150. Logo T-shirts and caps for their team of Conference Volunteer Carers.
- **Neuromuscular Support Group for Young Adults –** $300. To fly over a guest speaker.
- **People with Multiple Sclerosis Victoria –** $200. To help support the PwMS Conference.
- **Syndromes Without A Name Australia –** $250. Undiagnosed Children's Awareness Day.
- **The Aussie Hands Foundation Incorporated –** $100. To create an online section to the Aussie Hands website specifically for parents who have received the news that their unborn child has a hand difference. ■

Neuromuscular Support Group for Young Adults

My name is Hannah Pennington and earlier in 2015 I applied for a Small Grant from the Genetic Support Network of Victoria and was granted $300 which as I outlined in my application will be utilised for a small support group founded in 2014 called Neuromuscular Support Group for Young Adults (NMSGYA). The grant will be able to assist the group in moving forward, growing and hopefully increasing awareness for young adults or adults in general living with neuromuscular disorders.

This kind sum of money will go towards hosting our first event and inviting Karni Liddell who is an Australian Paralympian who suffers from Spinal Muscular Atrophy. Karni is an excellent role model and motivating voice for not only sufferers of neuromuscular conditions but also in a more general sense too, actively representing what it means to live the best possible life that you have been handed.

With some further organisation required the event will take place in mid-late 2015 and I will update the GSNV accordingly. Thank-you so much for assisting our small (but mighty) support group.

Hannah Pennington ■

Rare Voices Australia Summit. March 27 – 28, 2015

Rare Voices Australia (RVA) held its first national summit for rare diseases over March 27 – 28.

This was a chance for people living with rare disease, clinicians, government, industry, researchers and policy makers to come together to discuss a national approach to address the needs and concerns of people affected by a rare disease. Megan Fookes, co-founder and Executive Director of RVA opened the proceedings, introducing Senator Richard Di Natale who passionately spoke of the significance of rare disease in our society and that real change could only occur if driven individuals got organised and collectively worked together.

The first day of the summit was an introduction to the ground work that has already been laid in progressing a National Rare Diseases Plan for Australia. Megan Fookes detailed the findings of the National Roadshow convened by RVA in 2014.

Discussions took place with key stakeholders in many states on the needs of the rare disease community as well as current barriers to change. From these discussions the principles and objectives to progress a national plan were collated including that rare diseases should be recognised as a national health priority. Key themes that emerged included the need for good data collection models, patient care models and equitable access to diagnostic and therapeutic services. The results of the Australian Rare Disease Survey were also disseminated, revealing that families and individuals affected by rare disease need better coordination of health services, access to timely and accurate diagnosis, access to peer support and support groups, psychosocial support and access to better financial support.

The remainder of the day was invited speaker presentations on individual themes emerging from the National Roadshow. Talks included patient, clinician, government, industry perspectives as well as international viewpoint from areas which have had success in implementing rare disease initiatives. Sessions were informative and engaging, with a panel discussion at the end of each theme to begin to provoke thought and discussion in delegates in preparation for the workshops of day two. In these workshops, facilitated by RVA, delegates discussed the theme of interest and devised key principles for inclusion into the national plan.

The focus was on promotion of what is working now in the current system and how this could be expanded to a national approach. At the end of day there was a wealth of information available to be included in a National Rare Diseases Plan for Australia.

The GSNV is proud to support RVA in its progress moving forward. ■

Ballarat Lymphoedema Education and Exercise Support Group

(BLEES Group) By Kay Timmins Group Leader

Kay is at present Chairperson of Lymphoedema Association of Victoria and is also the President/ Chair of Genetic Support Network of Victoria committee, at The Royal Children's Hospital.

What is Lymphoedema you may ask?

It is a chronic swelling of a body part (usually) arms or legs, trunk or face caused by an accumulation of fluid and protein in the tissue. It can be hereditary, which is known as primary lymphoedema, where people can be born with this condition. Years ago it was also known as "Drop see" or "Elephantitis" because of the thickness of the skin. There is now treatment and care available that can help anyone that has this condition to be able to manage this. It is a combination of Manual Lymphatic Drainage (massage) (this is different to Lymphatic Drainage) management of skin care, compression garments, exercise, elevation, education.

This all comes under the education in what, when, why, how?

We support everyone that comes to our group. Our support group meets on the 1st Tuesday of the month, at Eastwood Leisure Complex in Eastwood Street, Ballarat. Our support group is run by a practitioner and that has set up and has run other support groups. Our group meets as a social group to help people that need help and offer support even just to go out and have a cuppa. We have some guest speakers through the year as well. Not everyone is into support groups but for some it is there only contact with others that suffer this condition, as they are not alone.

Coming up in our Education dates:

BLEES Group/Ballarat Lymphoedema Support Group
Sat 15 August 1.30pm at Eastwood Leisure Complex, A Get together and Education Session to help answer questions people may have. Gold coin donation. With Devonshire tea supplied. Booking essential. Ph **0417 155 511**

The Lymphoedema Association of Victoria also held a Public Awareness Seminar day on 16 of May 2015 – **Lymphoedema Doesn't Discriminate.** ■

SUPPORT GROUPS

CONNECTIONS 2015

The Inaugural Australian Conference for disorders of the corpus callosum

March 21 – 22

By Pieta Harris

Left: Pieta Harris and daughter Matilda

Connections 2015 had over 100 attendees and was the inaugural ausDoCC Australian conference for disorders of the corpus callosum (DCC). It included speakers from America, Brazil, France and Australia and established ausDoCC as a support group for families and individuals with DCC as well as beginning a strong rapport with professionals from all perspectives of health and education. Three years ago the GSNV awarded ausDoCC a small grant for an initial face to face meeting and helped to establish ausDoCC's mission and vision statements. ausDoCC have grown from that point and thank GSNV for the wonderful board from which to spring.

Being in a room with people to whom there is no need to explain yourself, people that understand the grief, the confusion and the sheer joy that you have experienced, people that also continue to feel forever proud of their child however always a little worried for the future, people that actually know what the corpus callosum is; this was the highlight for me in attending the first conference put together by the ausDoCC committee.

Although I will also admit that I was maybe just a little excited to be in the room with the leading specialists in the world of DCCs. I was a little star struck, to me these people are royalty, are celebrity. I actually couldn't bring myself to chat to them on day 1, I was like a tongue tied adolescent meeting Katy Perry! The two day conference certainly took me on an emotional rollercoaster and I'm sure I'm not alone in saying this. The high of meeting people face to face who I had spoken to for years online and the low of the reminder that no matter how well Matilda is going, she does indeed have a congenital condition that may bring some hurdles along the way. Even though this reminder was hard to hear it was timely as I had been somewhat getting a little comfortable with how she was developing.

I had recently been considering giving up our early intervention place thinking we were using a resource that others need more. I now feel that while yes, perhaps others do have greater needs, Matilda is still worthy of the support and assistance. She's not the "fraud" her paediatrician called her; she does indeed have a DCC so it's time to regain momentum in assisting her. I feel I have somewhat more direction hearing from Dr. Lynn Paul, that I should be working to build compensatory strategies for Matilda from a young age; implement a diary, to do lists, encourage her to ask questions, ensure she knows about personal space and more.

I feel like finally someone could give us some strategies beyond the 'wait and see'. While the conference didn't lead to some "ah ha" moment of clarification answering all my questions, I feel more comfortable in sitting with these questions, I feel like I can accept having questions and that I'm not alone in my journey. Perhaps that is the best thing I could take from any conference. ■

Adults with ACC. Chavon, Taylor, Emily and Abbie

Registrations Day 1

Professor Linda Richards (Queensland Brain Institute), Abbie and Maree Kinniburgh (ausDoCC and GSNV) Professory Lynn Paul (California Institute of Technology)

SUPPORT GROUPS

Klinefelter Syndrome Seminar – 29 March

The Genetic Support Network of Victoria in collaboration with Genetic Alliance Australia (GAA) hosted an XXY, (Klinefelter Syndrome (KS)), Seminar. The seminar was held in Sydney at the new home of the GAA, the Garvan Institute.

The day had a number of great speakers including endocrinologist, Dr. Kishani Kannangara and paediatric endocrinologist Dr. Shubha Srinivasan, both of whom gave insights into the medical side of KS. The two endocrinologists were very well received and brought the audience up to speed with the current state of KS. It was clear that the audience appreciated these informative talks as they posed many questions to the two specialists regarding medical care at different ages.

Rex Toomey, whose adult son has KS, kindly shared his family's story, speaking about the ups and downs of living with KS. Monica Bray who is spearheading the reinvigoration of the Australian XXY support group (now called Australian X and Y Spectrum Support), also told her story of her son's experience with KS. She also introduced us to the new support group and what they endeavour to achieve. Monica and her team definitely need to be commended on all the work that they have put into making the group and the seminar a success! The GSNV conducted a brief needs assessment survey with the attendees, which was designed to get a general sense of what the KS community feels are the information and service gaps. The data gained from the survey will be used to develop a wider national survey to further understand the service and information gaps for people impacted by KS.

Some of the early findings include:
Thirty individuals participated in the survey; 56% of which had a child with KS, 33% had KS, 6% had a partner with KS and 3% had a family member with KS. Overall the greatest concern was education and access to integration aids, especially among parents with 35% of parents stating this was their main concern. The biggest concern amongst individuals with KS is the lack of a national resource. Services accessed by people impacted by KS include paediatricians, endocrinologists, Centrelink, speech therapists, occupational therapist, psychologists and psychiatrists. About two-thirds of individuals are being seen by an endocrinologist and 30% of individuals stated that they have access to Centrelink funds. Just over half of individuals are connected to more than one service. The internet was a popular tool used to search for service information (26%) however, 40% of individuals were referred to the services they accessed, by a medical or health professional.

The biggest issue in accessing services seems to be that the services that people feel they require do not exist in Australia (39%) such as health professionals with appropriate knowledge of KS and school support and funding for children with KS. The financial costs of the services are also a barrier for many people (26%). This was a very interesting look at the gaps in services for people impacted by KS. In the future it would be most beneficial to combine efforts and establish a national KS survey, capturing feedback from the KS community in all states. Such a survey would represent a true collaboration but would not be successful without the participation of consumers. A national KS survey would also provide important evidence to funding agencies in terms of the information and service gaps for a very important group of people – the KS community.

The GSNV would like to thank the GAA, especially Dianne Petrie and Doriane Ranaivoharison for all their hard work leading up to the seminar and congratulate them on the success of the day.
For more information on Klinefelter support in Victoria and Australia please visit: **gsnv.org.au/klinefelter-support** ■

My experience of life on the Disability Support Pension (Blind) By Twanny Farrugia

Twanny is a self-funded retiree who cares for his elderly mother. He is legally blind and is assisted by a guide dog called Val. He has also had a renal transplant, open heart surgery and has artificial hips and knees. Twanny writes below about his experiences living with the assistance of the Disability Support Pension (Blind).

I am on multiple medications, most of which are on the Pharmaceutical Benefits Scheme (PBS) but I also take several others that aren't on the PBS and therefore have to pay full price. Additionally, my guide dog has also got health issues (allergies) and therefore I have additional expenses for her. Due to my low vision, my electricity bills are also higher, as when able sighted people can often use natural light from windows, I am unable to do so and need the lights (high Wattage) on. Furthermore, I am a high user of electronic devices to assist my independence. I can have up to five devices being recharged over night, which I would need to utilise during the day for my day to day activities.

On top of this I have taxi fares, clothes, insurance, leisure (not much of) etc. Now days I am very conscious prior to buying anything, I need to clarify to myself if it is a 'want' or a 'need.' I often go without many 'wants' (i.e. going to concerts, opera, out with friends, holidays etc.) to ensure my 'needs' (i.e. medicines, Meals On Wheels, insurance etc.) are met. I often have to juggle items of needs to fit within my low income and very tight budget.

Finally, I am fortunate to be on a Disability Support Pension (Blind), as it is not means tested like the normal Disability Support Pension. This allows me to earn other income (I am a self funded retiree) without my pension being reduced. However, I am not rich and therefore still have to live on a tight budget.

Above: Twanny and Val

Given my circumstances, I hate to think how much more difficult it is for people living on the normal pension which is means tested. Furthermore, I feel further fortunate because I am living at home and I don't have to pay private rental, which must really make it so much more difficult for pensioners. ■

Rare Disease Day 2015 and Genetic Support Awareness Week

Above: Sophie

In the lead up to Rare Disease Day 2015, the GSNV hosted a week long awareness event entitled "Genetic Support Awareness Week (GSAW)". From the 23 – 27 of February, GSAW aimed at raising awareness about living with a genetic and rare condition and to support people impacted by genetic and rare condition.

During the countdown to GSAW, the GSNV highlighted achievements in the field of genetics and shared stories from the community. In the next few issues of the newsletter we will showcase some of this wonderful narrative and share the stories of the GSNV genetics community, in their own words.

The finale of GSAW was the GSNV Rare Diseases Day celebration on the 27 February 2015. We hosted a presentation morning, which included talks from:

- Kathryn North who gave the key note address
- Dr. Sue White (Melbourne Genomics Health Alliance) who spoke about the experience of doing exome sequencing in the Melbourne Genomics Childhood syndromes flagship
- Heather Renton (Syndromes Without A Name Support Group) who gave a family perspective of living with a rare disease
- Panel discussion and questions from the audience, facilitated by Louisa Di Pietro – Panel: Dr. David Amor, Dr. James Pitt, Ivan Macciocca, Dr. Sue White

The event was followed by a morning tea and opportunity to mingle, with guests - raising their hands in support of Rare Disease Day.

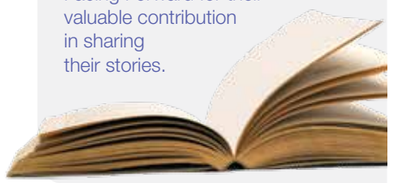
A very successful part of our GSAW program was the GSNV “Facing Forward” campaign, which aimed at giving the genetics and general community a visual insight into the lives of people impacted by genetic and rare conditions and to raise the awareness of the importance of support services in genetic health. The GSNV invited its members and community to send in their visual interpretations of living with a genetic and rare condition with a short message. We were very pleased to showcase these on the GSNV website and social media. The contributions were stunning and the campaign a great success.

"Being born with a significant intellectual disability does not mean that you can't learn. Sophie spent a year at University in 2013 and loved learning!" ■

SANOFI

The Genetic Support Network of Victoria would like to thank Genzyme, a Sanofi company, for their generous grant to create a photo book commemorating our successful “Facing Forward” campaign held in the lead up to Genetic Support Awareness Week (23 – 27 February) and Rare Disease Day 2015.

The photo book will feature a collection of entries received during the campaign and hopes to raise awareness of our genetic and rare disease community in Victoria. We thank everyone who participated in Facing Forward for their valuable contribution in sharing their stories.



As part of raising awareness for Rare Disease Day, the RAISE AND JOIN HANDS campaign is a way to express solidarity with rare disease patients around the world!

Rare Disease Day events from all over the world are recorded on the international website using this symbolic gesture of RAISING AND JOINING HANDS.
www.rarediseaseday.org/join-your-hands/

GENETIC SUPPORT & ADVOCACY

Rare Disease Day 2015

Michelle's story:

Having a child with a rare disease is a journey into the unknown. Michelle Karam was lost when the doctors told her they suspected something was wrong with her little girl AJ. After bringing her daughter in for a formal blood test, she was told they thought it might be Maple Syrup Urine Disease, a recessive metabolic disorder leading up to a build-up of amino acids and their toxic by-products in the blood and urine.

"At this point I was completely lost! I had never ever heard of MSUD let alone how my little lady (that we tried many, many years to have) ended up with it. Finally we were taken in for the blood test. My little AJ was two weeks old with nurses jabbing needles in her tiny little arm, and I was trying to understand what was going on (my father had passed away 8 weeks earlier) and the weight of it all got too much to hold. Looking around at all these strangers running around frightened me and I realised that we were in a very serious position.

After the nurses got the blood from AJ, I was sent to wait in a cubical for her results. I started praying to my Dad to protect AJ and really hoping that my world was not going to be turned upside down. A few hours later a clan of doctors and dieticians walked in, my heart dropped!

The clan took me into a private room and explained what AJ has and how she got this disease. I was still in a spin as to what this disease was and how it would affect my family. I wanted to know how long she would live for. To my relief they felt my pain and so thoughtfully answered "we're here to help her live a long normal life." As time has passed I understand a hell of a lot more about this very rare disease and how to control her levels thanks to the most amazing team God has put on this earth. Without the support and efficiency of the metabolic department I would have lost my baby girl, but instead I have a very healthy and happy 11 month old." ■

Reflections on How ChIPS Breaks Down Barriers

Vassie is involved with Glen Eira Council Disability in Schools Program that is aimed at raising awareness in schools to help students understand the experience of living with a disability. Her first presentation was held in May at Caulfield South Primary School. For further information on Council’s Disability Awareness in School’s Program, contact Council’s metro access officer on 9524 3333 or visit www.gleneira.vic.gov.au Vassie is also currently studying a Bachelor of Youth Work at Australian Catholic University, which she will complete in June.

ChIPS – (Chronic Illness Peer Support) is a peer support program for young people aged 12 – 25.

Beginning with an eight week introductory group meeting, the challenges of living with a chronic illness as a young person were addressed weekly, run by a health professional with a peer leader assisting. Once people have finished the initial group there are opportunities for socials, camps and to complete leadership training and to take on leadership responsibilities, including helping with organising events.

Recently when I was at Lakes Entrance and rocking a ChIPS T-shirt as I still often am, an older man asked mum where ChIPS camp was, what it is and if I was a participant or group leader. This also highlights that no matter where we are as people, when we wear the annual camp T-shirt in public we represent the group and therefore indirectly raise awareness.

Something that is understated about ChIPS is that it breaks down barriers between young and old and between medical staff and young people. In practical terms this means at camp, the medical staff are dispersed among the young people with one staff member to a team of young people. This means that the staff blend in with the young people and move between their professional roles and engaging young people. The impact of this, after many years of camps, has been that when I have been faced with medical experiences as an adult (despite many scary experiences as a child) I was able to manage.

The camps allowed me to see the medical staff simply as other people as opposed to people that only did scary things to me. This was a huge shift and gave me the ability to self-manage my medical care. ChIPS have also increased my social circle beyond school and increased my independence. I also had the privilege of hearing how youth older than me manage as well, which also proved to be invaluable to learn from, about life ahead.

These conversations also influenced where I studied after school as I became aware, through older members, of which education institutions were well resourced. ■

"What's that lab?"

Our new 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 Greta Gillies, Research Assistant in the Bruce Lefroy centre's Neurogenetic Research Group, tells us about the lab's role.

The Neurogenetics research group is part of the Bruce Lefroy Centre and undertakes laboratory-based molecular neurogenetic research. Our primary focus is gene discovery and functional genomics. We utilise new genomic and bioinformatic technologies like high density SNP arrays, massively parallel sequencing, association studies, together with traditional techniques like sanger sequencing for novel gene discovery. Subsequent studies using cell and animal models allows us to elucidate the function and pathogenesis of novel disease genes.
We have successfully used these techniques to identify genes causing chromosome breakage syndrome, Parkinson's disease, brain malformations and other disorders.

We collaborate with clinical researchers within VCGS, MCRI and RCH, with flagship projects including The Accelerated Gene Identification Program (AGIP), Genetics of Brain Development (GBD) and The Collaborative AuTism Study (CATS).The experimental design of our projects involves a co-ordinated clinical/research pipeline. We work very closely with clinical and public health professionals, bioinformaticians, national and international collaborators. We maintain excellent communications with affected individuals and families to maximise our opportunities for genetic diagnosis, result feedback, counselling and possibly treatment as part of a holistic service.

For further information about the Bruce Lefroy Centre, please visit our webpage
www.mcri.edu.au/research/themes/gd/bruce-lefroy-centre/

Some recent publications:

Lessel, D. et al. (2014) Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nat Genet Nov 28;46(11):1239-44. Epub 2014 Sep 28.

Wilson, G.R. et al. (2014) Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with-Synuclein Pathology. Am J Hum Genet Dec 26;95(6):729-35. Epub 2014 Nov 26.

SERVICES

Children's Tumour Foundation (CTF)
Victorian Neurofibromatosis Family Information Day

By Sally Maspero and Gemma Brett

The Children's Tumour Foundation (CTF) provides support to children and adults impacted by Neurofibromatosis, three genetic conditions known as NF (NF1, NF2 and Schwannomatosis).

We provide assistance and information to families and friends, educators and health professionals, and assist those impacted to find help and friendship within the NF community. We also fund some clinics and raise funds for research and support programs.

On Saturday 7 February 2015 CTF Australia held a family information day at the Murdoch Childrens Research Institute in Melbourne, combined with the official launch of the new paediatric NF Clinic based at the Royal Children's Hospital (RCH) in Parkville. The day was a great opportunity for individuals and their families living with NF to meet each other to form friendship and support networks, and to meet the CTF and RCH NF Clinic teams.

While the thirty five children in attendance enjoyed balloon animals and games with the wonderful child carers arranged by the Victorian CTF State Committee, over 75 adults participated in an interactive information session. The highlight was an interesting and informative presentation by Professor Rosalie Ferner who was joining us all the way from London, where she is the Consultant Neurologist and Lead Clinician for Neurofibromatosis at Guy's and St. Thomas' NHS Foundation Trust. Professor Ferner guided the audience through a discussion on the diagnosis of NF1 and some of the issues that can arise in tackling this disease, all the while demonstrating her sense of humour and encouraging audience participation. One young man even stood up to talk with the Professor in a discussion on pseudoarthrosis, to show how he is not letting it impact on his life. Others in the audience emphasised the need for greater advocacy due to the lack of school and services support funding and a number of adults described the impact arising from cosmetic issues due to neurofibromas. While the vast majority of attendees were there to hear about NF1, Professor Ferner also spent some time at the end of the day speaking to the one person with NF2 who attended.

Response to the day was overwhelming and demonstrates the need to continue providing information and family events for Victorians living with NF. An evaluation is currently underway, the results of which will help us in planning future activities.
For more information, and to keep up to date with future CTF events in your area, please visit our Facebook page: www.facebook.com/CTFAus (note that you can view the public posts on this page without needing a Facebook account!). Alternatively you can join the closed Facebook group called Children's Tumour Foundation (NF Australia).

We are excited to welcome Natalie McLean,

NF Support Co-ordinator for Victoria and Tasmania to the MCRI and the GSNV. Natalie is now working out of the GSNV office until Louisa returns in 2016. Natalie is hard at work liaising with families and individuals with NF, providing them with information and assisting them with meeting others in the community with NF. Natalie will also provide assistance for self-advocacy, and advocacy within the community for people with NF. She will also help organise group activities, information sessions and potentially a family camp.

SERVICES FOR YOUNG PEOPLE

Youngcare

By Matt Adams Corporate Communications and Media Manager, Youngcare

Youngcare understands that currently more than 7,000 young Australians (under the age of 65) with full-time care needs are living in aged care simply because there are few alternatives. There are also 700,000 more young Australians being cared for at home by family and friends.

These young people are often living in inappropriate conditions with limited support, simply due to a lack of alternative age-appropriate housing options. Youngcare believes that every young person deserves to live a young life, regardless of their care needs. Being young is about having a lifetime ahead of you, yet aged care is designed for someone who is at the end of their life.

Youngcare's mission is to help young people avoid new admission to aged care right now. Instead, the aim is to develop viable and replicable models to solve the problem, giving young people with high care needs the choice in care and accommodation they deserve.

Youngcare At Home Care Grants program was introduced in 2009 to assist young people with high care needs who are living at home and are at risk of entering aged care facilities. Through the generous support of corporations including foundation partners Suncorp and GIO grants between $2,000 - $10,000 can fund equipment, home modifications, and emergency respite care. In 2013/2014 $772,000 in funding was distributed to 139 successful applicants across Queensland, NSW and Victoria. The Youngcare Connect phone service had over 2,250 incoming and outgoing calls made during the year and expanded with the appointment of a Youngcare Connect Liaison Officer in the Sydney office. Youngcare welcomes the NDIS as an historic initiative that will change the lives of young Aussies with high care needs. The NDIS will not however solve the accommodation issue for young people with high care needs, which reinforces the need for Youngcare to continue its work to deliver best practice models of accommodation options. As the Australia-wide NDIS trials progress, Youngcare will continue to engage with the National Disability Insurance Agency and with state and federal governments to highlight key issues that will continue to be faced by young Australians with high and complex needs and the lack of funding for capitol works for age appropriate accommodation solutions.

Youngcare is celebrating its 10th anniversary this year and will continue to raise much-needed funds and awareness for young Aussies with high care needs, and their right to live life with choice, independence and dignity.

Rick (pictured above) was granted funding for respite through Youngcare's At Home Care Grants program. This respite will take the pressure off his partner, and allow them to spend quality time together as a couple.

Haemophilia Foundation Australia youth website

This site has been created to talk about life, being young and having a bleeding disorder.
www.factoredin.org.au

Youth Affairs Council of Victoria

YACVic is the peak body and leading policy advocate on young people's issues in Victoria.
www.yacvic.org.au

Youth Disability Rights Hub

A powerful self-advocacy resource for young people with disabilities. It acts as a starting point for young people seeking information, resources, help and ideas to understand and stand up for their rights.
youthdisabilityrights.org.au

Youth Disability Advocacy Service

YDAS offers a free individual advocacy service to young people aged 12-25 who have a disability.
www.ydas.org.au

Foundation for Young Australians

FYA delivers a range of initiatives (co) designed with young people to deliver change across Australia. Collaborating with schools, government and business FYA have imagined programs that connect, influence, shape, equip, innovate and transform.
www.fya.org.au

Youthlaw, Young People's Legal Rights Centre

Youthlaw is an accessible legal service to young people under the age of 25, focusing on areas of unmet legal need.
youthlaw.asn.au

Royal Children's Hospital

factsheets for young people
www.rch.org.au/transition/factsheets_and_tools/brochures

CALENDAR OF EVENTS

JUNE	
27 June	HeartKids 25th Anniversary Gala
28 June	Stay in Bed Day 2015
29 June	World Scleroderma Day 2015

JULY	
July	Fragile X Awareness Month
1 July	Haemophilia Treatment Centre Boys Day Out
15 – 18 July	The Angelman Syndrome Foundation 2015 Biennial Conference
15 July	Carers Vic - Support group facilitator training – session 3
22 July	Fragile X Awareness Day

AUGUST	
2 August	HFV Grandparents and friends lunch
8 August	SMA Fundraising Diamond Gala Ball
13 August	Carers Vic - Support group facilitator training – session 4
13 – 14 August	1st Australasian Undiagnosed Disease Program (UDP) Workshop
15 August	Ballarat Lymphoedema Education and Exercise Support Group meeting
24 – 30 August	VCFS 22q11 Awareness Week
28 – 30 August	CMT youth weekend 2015

SEPTEMBER	
1– 7 September	Spina Bifida Awareness Week 2015
16 – 19 September	11th Asia Pacific Conference on Human Genetics 2015
27 – 30 September	Fragile X/Cognitive Disorders Workshop

OCTOBER	
11 – 14 October	15th Annual conference of the Australasian Genomic Technologies Association (AGTA)

NOVEMBER	
November	Alpha-1 Awareness Month and Muscular Dystrophy Awareness Month

EVENTS

CMTAA
Charcot-Marie-Tooth Association Australia Inc.

CMT YOUTH WEEKEND 2015

Are you a young school kid with Charcot Marie Tooth Disease and feel like a fun, challenging time away from home? Experience heaps of fun activities like archery, giant swing, cable glide, canoeing, rock climbing and more.

To be eligible to attend you only have to be a student at school aged from about 10 years; be a member of the CMTAA (if you're not already a member, contact us) and have some form of CMT, oh... and want to have fun with other kids just like you!

Date: Friday, 28 August – Sunday, 30 August 2015
Venue: NSW Sport and Rec Centre at Berry on the NSW South Coast

Email **Jillian at cmtaussiekids@gmail.com** or call mobile **0428 221 264** for more details or to register your interest and we'll send you the details.

SUPPORT CMT AUSSIE KIDS

If you don't have kids who could come to this great weekend, why not consider sponsoring the weekend by making a one off donation to the CMTAA youth weekend fund?

Your donation will help kids with cmt learn to deal with and manage their day to day problems and look to a brighter future.

Check out the video from previous camps at **www.cmt.org.au** or on youtube
To donate to cmtaa youth weekend or to find out more, contact CMTAA on **(02) 9767 5105**

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 either 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.

FA GUIDELINES

Researchers from the Murdoch Childrens Research Institute have led the development of clinical guidelines for the diagnosis, treatment and management of Friedreich Ataxia (FA). This is a great step forward in the international standardisation of clinical assessment of FA; helping to educate physicians and thereby improving treatment and medical outcomes for individuals living with FA. **curefa.org/_pdf/ClinicalManagementGuidelinesForFA.pdf**

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