genetic support
network of victoria

empowering * connecting * supporting

AUTUMN 2012

GSNV's Rare Disease Day Seminar

Rare Disease Day is an annual world-wide observance held on the last day of February to raise awareness for rare disease and to improve access to treatment and medical representation for individuals with rare diseases and their families. By Penny Dodds

A rare disease is a disease with a prevalence of 1 in 2000 people or less affecting less than 200,000 people in the population.

There are 8000 known rare diseases, collectively affecting up to 10% of the population or over 2 million people including about 400,000 Australian children. 80% of rare diseases have identified genetic origins whilst others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative. [1]

Anna Hickey is one such individual affected by a rare disease called Gorlin Syndrome.

Gorlin syndrome is an inherited "autosomal dominant" condition, which means there is a 50% chance that children can inherit the condition from a parent with the condition.

Gorlin syndrome, also known as nevoid basal cell carcinoma syndrome, is a condition that affects many areas of the body and increases the risk of developing various tumours.

Anna says the main issues for her has been "the tedium of continual medical appointments; (they are) draining physically, emotionally and financially."

Another issue for Anna is "that social engagements are planned around these appointments and there is the temptation to withdraw from society because of feeling conspicuous."

On a daily basis the team at the Genetic Support Network of Victoria (GSNV) assists people with the non-clinical or 'human' aspects of managing genetic conditions.

GSNV President Moira Rayner says, "The major focus of GSNV's work is to provide advocacy, education and peer support training for people affected by genetic conditions. We work in partnership with the Genetic Heath Services of Victoria and the Murdoch Children's Research Institute to better the lives for people affected by rare conditions."

Moira Rayner, Anna Hickey and Tina Costanzo (The Prader Willi Syndrome Association of Victoria) spoke on Friday March 2 as part of a Rare Disease Seminar hosted by the Genetic Support Network of Victoria (GSNV).

Anna and Tina presented their personal stories and Moira reported on the finding of GSNV's Survey to find the Top 5 Issues for People affected by Rare Genetic Conditions and Diseases.

[1] *Source: Improving outcomes for rare disease in Australia: A National Plan for Rare Diseases (Department of Health WA)*

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genetic support network of victoria

Committee of Management 2012

President	Moira Rayner
Vice President	Jan Hodgson
Treasurer &	
Public Officer	Geraldine Allen
Secretary	Amanda Springer
Secretary	Alice Weeks

General Committee Members
Christine Williams
Yvonne Waite
Maria Triantafillou
Amy Herlihy

Committee Meeting Dates 2012

Teleconferencing will be available at all meetings.

Thursday April 19
Thursday May 17
Thursday June 21
Thursday July 19
Thursday August 16
Thursday September 20
Thursday October 18 AGM Meeting
Thursday November 15
December (to be confirmed)

Message from the President

The political and economic climate in Victoria is one of uncertainty and it's a vulnerable time for any not for profit organisation especially one as small as ours. The only way to manage such uncertainty is to step forward briskly, doing what we have to, which is make sure your voice is heard.

We need to keep the conversation going between you, who are affected by genetic disorders, with others who are as lost and concerned, and the Murdoch Childrens Research Institute (MCR), who is servicing our little office. One way of doing this is to deliver the facts of your lives to researchers and administrators.

The last day in February is 'rare disease day' and on February 29 I chaired a Genetic Support Network of Victoria (GSNV) seminar aimed at students, researchers, clinicians, genetic counsellors and GSNV members on rare diseases.

We focused on our recent survey scoping our members' Top 5 Concerns regarding rare disease and how they may be addressed.

Two of our members talked about the lived experience of someone who has a rare disease, and how important it was to not only have the services you need but the money to be able to pay for them, and to feel and be supported through that adjustment process.

The other good news is that we have a much more user-friendly web site, which is now ready to go. It makes the conversation flow much easier.

Coming up to Easter – have a good, restful break.

Moira Rayner
President

National Disability Insurance Scheme

The Every Australian Counts campaign has a simple clear message for all of our politicians. In 2012 – Make the NDIS Real.

The campaign will be enlisting our more than 100,000 registered supporters asking them to create a short one minute video where they tell Australia why they are counting on the NDIS.

National Campaign Director, John Della Bosca said: "The NDIS is a vital social and economic reform. Currently people with a disability, their families and carers suffer under a disability support system that ranks at the bottom of all OECD tables. The NDIS will provide the basic aides and supports for people with a disability. It will mean all Australians count".

"A report from PWC last year found the cost of not acting to fix our disability system through introducing an NDIS is greater than the cost of the scheme itself. The NDIS is the only major social reform on the political agenda that all political parties and all state parliaments support. This is a fantastic achievement."

"The campaign has a clear message for our parliaments. Make the NDIS Real. We have bi-partisan support for the NDIS and now we want bi-partisan action to make the NDIS a reality. As a first step, the Every Australian Counts campaign is calling on the Federal Parliament to come together and legislate to create the NDIA, the administrative body that will run the NDIS".

"People with a disability, their carers and families are often isolated and their voices overlooked in our national political debate. That stops today. The Every Australian Counts campaign plans to assemble thousands of video stories - real people, telling real stories about why they are counting on the NDIS. I believe that these stories will make the case for the urgent need for an NDIS."

Visit Make It Real Video petition at: **http://video.everyaustraliancounts.com.au/**

Press Release – February 9, 2012

COVER STORY

GSNV's Rare Disease Day Seminar

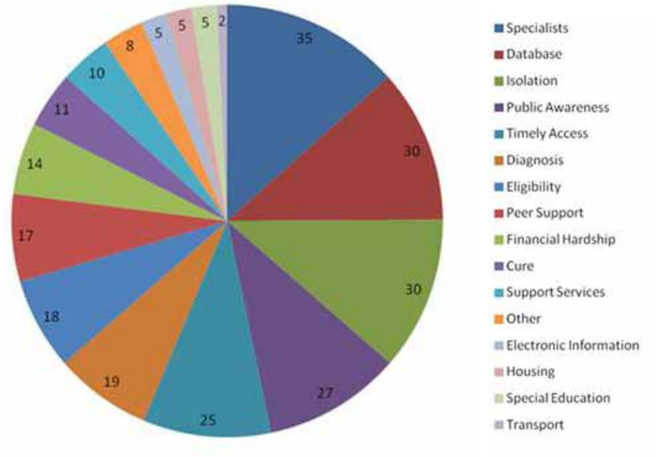
(cont. from page 1)

The GSNV conducted a survey of their members to find the Top 5 Issues for people affected by rare disease. The survey hit a chord with GSNV members with 56 respondents. People were asked to list their Top 5 issues from the following list of issues.

SURVEY LIST OF ISSUES

1. The need for a National database to collect information about individuals affected by rare conditions. A National database would assist with diagnosis, research collaboration, and would help the Government to allocate funding equitably, and influence health incomes.
2. Access to specialist doctors, treatments and medication
3. Timely access to support services, like respite & personal care support, funding for ISP Packages
4. Eligibility for and sufficient funding for specialist equipment/ equity and access.
5. Isolation, stress for individuals and families, mental health and poor quality of life issues
6. Financial hardship
7. Public awareness/acceptance
8. Diagnosis and prevention, including lack of screening and diagnostic tools
9. A cure
10. Peer support/ support groups
11. Transport
12. Access to special education
13. Housing
14. Support to enter the workforce/ inability to maintain employment
15. Provision of electronic information available relating to rare diseases
16. Other (please nominate)

_____



The Top 5 Issues were as follows:

1. Access to specialist doctors, treatments and medication (35 respondents)
2. The need for a National database to collect information about individuals affected by rare conditions. A National database would assist with diagnosis, research collaboration, and would help the Government to allocate funding equitably, and influence health incomes. (30 respondents)
2. Isolation, stress for individuals and families, mental health and poor quality of life issues. (30 respondents)
3. Public awareness/acceptance (27 respondents)
4. Timely access to support services, like respite & personal care support, funding for ISP Packages. (25 respondents)
5. Diagnosis and prevention, including lack of screening and diagnostic tools. (19 respondents)

There were many comments highlighting the struggle and hardship that many people faced with managing their rare genetic condition/disease.

Many people also commented that all the issues were important and that it was difficult to nominate just five issues.

The chart below denotes the number of respondents to all of the issues listed.

For more information about Rare Disease Day please visit the website at **www.rarediseaseday.org/** or contact the GSNV on **info@gsnv.org.au** or phone (03) 8341 6315.

Many thanks to GSNV volunteer Rebecca Freedman for her work on this project, and to Anna and Tina for generously giving their time to share their personal stories.

GENETIC SUPPORT & ADVOCACY

GSNV's Peer Support Training

Professional Counsellors are helpful, but there is nothing more powerful than talking to someone who has been "in the same boat" as you.

When 10 December 2012

Where Walford Room, Murdoch Childrens Research Institute, The Royal Children's Hospital, Parkville

Peer support training is available for all individuals, including staff from support groups.

Our aim is to help you feel empowered to listen and talk to others affected by genetic conditions, and share your wisdom and experience.

Some areas that the peer support training course will cover include:

- Active listening
- Communication skills
- Empowerment
- Loss and grief
- Self-care
- Community resources
- And information about GSNV and what we can do to help the process.

You will receive a certificate upon completion as well as a Peer Support contract with the Genetic Support Network of Victoria. Costs will apply. Places are limited.

To register your interest please contact GSNV on 8341 6315.

2011 GSNV Members Survey

In 2011, the Genetic Support Network of Victoria (GSNV) conducted a members' survey both online and through email and post. By Lynley Donoghue

The main objectives of this survey were to:

1. Assess the efficiency of the level of support the GSNV currently offers to members
2. Conduct a "needs assessment" of members
3. Re-evaluate the goals of the GSNV to meet members' needs
4. Update the GSNV database

Thirty members responded to our survey overall, and provided us with some valuable feedback.

The majority of respondents were either individuals and families, or members of a support group. Most were either personally affected by a genetic condition, or close to somebody who is affected.

When asked about the use of our services, most respondents had used them at some point, and half of the respondents were either satisfied, or very satisfied with what we offer. People who hadn't used our services cited reasons such as not feeling it was relevant to their individual circumstances or support group.

We received some valuable feedback about how we could improve our services, such as offering support to individuals who have not been assigned a diagnosis, as these are the people who often miss out. This is something we hope to address in the future.

73% of the respondents would recommend the GSNV to their friends and families, providing a range of feedback such as it being a good avenue to gain access to information and support.

Thank you to all those who responded, your input is greatly appreciated.

Although our survey is now complete, we appeal to our members to assist us in our work by providing valuable responses to our surveys.

We also encourage our members to get in touch with us if there is any way in which we can assist yourself, your family or your support group. ■

CONFERENCES

Reflective Report – ICHG 2011

The ICHG in Montreal Canada last October was a wonderful opportunity for me to glean a deeper understanding of current advances in gene technology and genetic research on behalf of the Genetic Support Network of Victoria (GSNV). By Louisa Di Pietro

Advocate Program – International Congress on Human Genetics (ICHG), Montreal Canada 2011

More importantly, I was able to contextualize that with greater consumer (advocate) participation as there is the potential to build on and strengthen the quality of outcomes and translation of research into health care, with many stakeholders interested in the process.

The trip was made possible via an advocates grant provided by the US Genetic Alliance, which went a long way in meeting some of the costs in attendance.

The GSNV is grateful to have had the opportunity to be part of this important move in bringing research and those affected by genetic conditions together at such an important international meeting.

I was interested in participating in the Advocates Program to explore how advocates and researchers can collaborate and draw on each other's knowledge to build on and strengthen the quality of health and medical research. I believe this collaboration can be achieved through partnerships of consumers and researchers based on mutual trust and shared social responsibility, giving consideration to what each can reasonably expect from the other.

Consumer and community participation in health and medical research has mostly been in the areas of health services research, clinical research and multidisciplinary research. The potential contribution of consumer and community participation to scientific and genetic discovery has been less recognised, perhaps because this type of research is more often curiosity driven and based on the questions of researchers, rather than on immediate improvements in clinical practice and health outcomes.

Advocates, consumers and community members have a legitimate interest in this area and would welcome greater public accountability and discussion. I would like to be directly involved in discussions around how we might do that in future congresses. Much of my time in Montreal was spent in discussion of this point.

My interests at ICHG Montreal

Each plenary session and those focusing specifically on genetic advances, translation, ethical issues and genetic education were of particular interest to me. The sessions were broken up in such a way that I was able to plan attendance around my interests and, opportunities to meet researchers and presenters.

Earlier in 2011, I attended the very first Australian Rare Diseases Symposium and was encouraged to see that there is indeed an acknowledgement in Australia that stakeholders includes: patients and carers, health, disability and social service providers, patient support organisations, patient advocates, researchers, biotechnology and pharmaceutical industry representatives and government. In an understanding of genetic health and future advancements the stakeholders are the very same people.

Patient support and advocacy groups are effectively driving policy development in many countries and I was humbled to meet advocates in Montreal and bring their 'how to' knowledge and insights back to Australia.

The panel discussion scheduled on October 11 on Whole Genome Sequencing (WGS) complimented my recent contribution to an Australian National Health and Medical Research Council (NHMRC) workshop on WGS and how the Australian health care setting might become equipped to handle the imminent entry of WGS and related technologies into clinical health care.

I was a little surprised at some of the messages and language around WGS and genomics but came home even more resolved that in any public health genomics, WGS or genetic health discussion in general, a clear advocacy message that translation into health care (including applications and utility), capacity and ethical, legal and social issues must be part of the international debate.

The ICHG did reaffirm to me that linking research with health consumer and community groups is increasingly important in the future. Some research teams are successful in engaging consumer groups, while others will need to begin by building these networks.

The general practice in Australia is that 'consumers' are identified in terms of who or what the research is relevant to.

The burning questions for advocates are around whether or not the research contributes to improved treatment and care for people with a particular health condition? If so, is there a consumer group organised around this condition? The national or state group (such as the Genetic Support Network of Victoria) may be able to provide contact details for local members but not always. Does the research have a geographical focus, such as a local community or health service facility? If so, are there any local community groups for the area or for the health service?

In conclusion

In recognition of the contribution that consumers can make to research and of their right to participate in research, I do see that future international meetings on human genetics should be based on a 'shared vision' with all stakeholders working in partnerships based on understanding, respect and shared commitment to research that will improve the health of humankind.

The 12th International Congress on Human Genetics was a professional and personal highlight for me and certainly wet my appetite for working towards greater participation in future meetings. ■

RESOURCES

Better Health Channel app

The Better Health Channel is promoting the free Better Health Channel iPhone and iPad app available to download at **www.betterhealth.vic.gov.au/app**

The App was recently launched by the Minister for Health, David Davis.

By way of a brief background, the mobile app provides Victorian health consumers with local health services information and access to Better Health Channel conditions and treatments fact sheets. It seeks to:

- Empower Victorians to manage their own health and wellbeing, and/or that of their family more effectively
- Improve health literacy and help Victorians make better informed health decisions, and
- Enable anywhere, anytime access to reliable health information and services.

Source: HFV Magazine, November 2011

ACSQHC Patient and Consumer Centred Care News

This newsletter targets key stakeholders with an interest in patient centred approaches and provides links to many resources and events of interest.

The newsletter can be accessed directly from the Commission's website: **www.safetyandquality.gov.au/**

For more information about the newsletter or to make a contribution, please contact Rhia Graney on 02 9126 3588.

Alternatively, you can also contact Ms Naomi Poole, Senior Project Officer, naomi.**poole@safetyandquality.gov.au** or by phone on 02 9126 3651.

Source: HEALTH ISSUES CENTRE eNews Bulletin - 21 February 2012

Consumers Health Forum of Australia

The Consumers Health Forum of Australia Inc. (CHF) is a peak organisation providing leadership in representing the interests of Australian healthcare consumers.

They work to achieve safe, good quality, timely healthcare for all Australians, supported by the best health information and systems the country can afford.

CHF Member organisations reach Australian health consumers across a wide range of health interests and health system experiences.

Telephone: 02 6273 5444
Email: **info@chf.org.au**
Web: **www.chf.org.au**

Consumers Health Forum of Australia

Medicines list iPhone App

Patients and carers with iPhones can now keep an up to date medicines list on their iPhone and email and print the list.

The new Medicines List iPhone App, developed by *NPS: Better Choices, Better Health*, also allows people to schedule reminders which prompt them to take their medicine. This is an exciting use of technology to help patients maintain an accurate list of their medicines and for the information to be available when they present to hospital or visit their community health care provider. For more information on the NPS Medicines List iPhone App visit **http://www.nps.org.au/iphonemedicineslist**

Source: The Australian Commission on Safety and Quality in Health Care, MEDICATION SAFETY ISSUE 7, FEBRUARY 2012

SERVICES

Newborn Screening

by Tarli Bogtstra

Newborn screening has been available in Victoria since the mid 1960s. The first condition screened for was phenylketonuria (or PKU) in 1966. In 1977, screening for congenital hypothyroidism began and in 1988, cystic fibrosis was added to the screening panel.

In 2001, new technology was introduced called tandem mass spectrometry which can look for a number of different chemicals at once using a tiny sample of blood.

This allows the laboratory to identify many more conditions, including medium chain acyl coenzyme A dehydrogenase deficiency (MCAD), homocystinuria and maple syrup urine disease.

In Victoria, 25 conditions are now screened for as part of the Newborn Screening program. A key criterion for adding conditions to the newborn screening panel is that early identification and treatment improves the health outcomes for affected babies.

A process of informed written consent for newborn screening was introduced throughout Victoria during 2011.

This process is being implemented to ensure health professionals and parents have all the information they need about newborn screening and to strengthen the informed consent process.

Research shows that many parents do not receive adequate information about the screening program and are not aware of the choices they have.

Parents will now be asked to provide written consent for the screening test before sample collection. They will also be free to make a choice about use of the screening card in research.

Sally Morrissy is the Newborn Screening Nurse at Genetic Health Services Victoria. With all the recent changes to the Newborn Screening Program we have taken the opportunity to ask Sally some questions:

We have no family history of a genetic condition, why should my baby be screened?

"The conditions screened for as part of the newborn screening program do not show any signs or symptoms at birth and usually, there is no family history. By the time symptoms of a condition do show, development may already be impaired. Through screening, affected babies can be identified early and in most cases, treated to prevent or minimise the health impact of the condition."

When and how is the screening done?

"The screening test is performed when the baby is between 48-72hrs old. Your midwife will collect a few drops of blood into a screening card by pricking your baby's heel. If you discharge from hospital early, the sample will be collected during a home visit."

How much does the testing cost?

"This is a public health program which is funded by the Victorian Department of Health and is operated by Victorian Clinical Genetic Services."

How will I receive the results of screening?

"Parents are not contacted when screening results are normal, but this process can take approximately 6 weeks for a small percentage of babies that need further testing for cystic fibrosis. The hospital where the baby was born receives a copy of all results."

My baby needs to be re-tested, what does this mean?

"A repeat collection will be requested by the laboratory for prematurity, collection taken too early, inadequate/contaminated samples or sample giving borderline abnormal results. This is concerning to parents but in approximately 99% of the time the repeat sample will return a normal result."

What does a positive screening result mean?

"A positive screening result is not a diagnosis, and does not necessarily mean your baby has a particular condition. Newborn screening identifies babies at increased risk of a condition. Parents of babies with a positive result will be contacted by clinical staff from Victorian Clinical Genetics Services and arrangements made for a referral to a specialist for further testing to confirm results."

There is a history of a genetic condition in our family; can this condition be tested for too?

"Unfortunately only the stated conditions will be tested for, another test would need to be performed for alternative conditions. There is a place for detailing family history on the newborn screening card, if that condition is being tested for, we would investigate further at that time."

Will the results of this testing effect our insurance?

"Yes it would as a pre-existing condition but I think it's too complicated to put an answer here."

Should you have any further questions regarding Newborn Screening please contact Sally Morrissy on (03) 8341 6460.

RESEARCH

Patients with serious genetic conditions given fresh hope of diagnosis

Patients with serious and potentially fatal genetic conditions have been given new hope of receiving a diagnosis after researchers successfully trialled next generation sequencing technology.

by David Thorburn

Patients with serious and potentially fatal genetic conditions have been given new hope of receiving a diagnosis after researchers successfully trialled next generation sequencing technology.

Researchers at Murdoch Childrens Research Institute and the Broad Institute of Harvard & MIT trialled gene sequencing technology, which can sequence thousands of genes at the same time, with the aim of determining its ability to be used in a clinical setting as a diagnostic tool to identify mitochondrial disease and other genetic conditions.

Mitochondrial disease is a serious genetic condition that affects the way the body coverts food into energy. There are over 100 types of mitochondrial disease, and about one child born every week in Australia will develop a severe mitochondrial disorder in their lifetime; about half of them will die in childhood.

Mitochondrial disorders are notoriously difficult to diagnose due to the multitude of genes and the highly variable symptoms across various organs. The current diagnostic process is an extremely invasive and expensive process for patients, and often doesn't result in a definitive diagnosis.

While the next generation sequencing technology has proven successful in uncovering novel disease genes in a research setting, researchers wanted to see whether it could also play a functional role in diagnostics in a clinical setting.

Researchers looked at 42 patients with mitochondrial disorders and sequenced the DNA of the mitochondrial genome, the 100 genes previously linked to mitochondrial disease, and over 1000 additional genes that are known to play a role in mitochondrial biology.

The study, which is published in *Science Translational Medicine*, found that using the technology 25 per cent of cases were immediately diagnosed; and a further 25 per cent of cases will be able to be diagnosed in the next few years as more genes are formally linked to disease.

Lead researcher, Professor David Thorburn, from Murdoch Childrens Research Institute, said the rate of diagnosis was likely to significantly increase in the future and the technology will greatly improve their ability to diagnose some of the most complicated genetic disorders.

"New 'next generation' DNA sequencing technologies are transforming the way we do research on inherited diseases. However, it remains a real challenge to transfer these approaches from a research tool into methods that can be used efficiently by doctors trying to sort out if a patient's disease is due to a genetic condition," he said.

"Although not successful in every child, this new technology is a big advance that will allow us to speed up diagnosis for families to end their diagnostic odyssey. It should mean that within the next few years we can diagnose most children with suspected mitochondrial disease within about a month using just a blood sample, instead of needing a muscle biopsy and taking months or years.

"Our approach is also suitable for other complicated genetic conditions like epilepsy, deafness and some forms of heart disease."

Professor John Christodoulou, Director of the Western Sydney Genetics Program at The Children's Hospital, Westmead, said the study shows how this new technology will be useful for a clinical setting and could eventually make targeted treatments a reality for mitochondrial disease patients.

"The use of this technology will mean families can finally get off the diagnostic treadmill, which for some families can take years, with all that heartache and uncertainty for the families, and the discomfort for the patient who is subjected to multiple tests," Prof Christodoulou said.

"Hopefully our discoveries will lead other doctors to test their patients with suspected mitochondrial disease. Then, as more individuals with these genetic mutations are identified, we will hopefully get a better correlation between specific mistakes in the genes and the severity of the disease for patients, enabling doctors to be more accurate when discussing prognosis with families.

"It also gives more certainty about the potential genetic risks to other family members and allows them to consider their reproductive options more definitively."

Australian Mitochondrial Disease Foundation (AMDF) president Dr Doug Lingard said the findings represent a huge step forward in transforming the diagnosis of mitochondrial disease, of which there are more than 100 known types.

"Scientists only discovered in 1988 that mutations in mitochondrial DNA caused disease, so it's been a steep learning curve to find and develop effective diagnostic methods," Dr Lingard said.

"This Australian breakthrough shows that combining world-leading research with the latest technology can make a real difference to patients and their families.

"It's particularly exciting because the AMDF has recently funded priority access at Royal Perth Hospital to one of Australia's first Next-Generation DNA Sequencing facilities, which will enable much faster, cheaper and more accurate diagnoses of mitochondrial disease for Australian patients."

Dr Lingard and his wife Margie lost their seven-year-old son Alex to mitochondrial disease 28 years ago, when knowledge of the disease was scarce. Their daughter Rose also suddenly developed debilitating symptoms several years ago at the age of 20.

"We waited over two decades for Alex's diagnosis, and had an agonising wait of many months for Rose to be diagnosed before doctors could manage her symptoms properly and she could come home from hospital," Dr Lingard said.

"Having a quick and simple test to diagnose mitochondrial disease would have saved much distress and despair. However, while diagnosis may provide considerable relief and resolution for patients and their families, there is still no targeted treatment and no cure, so continued research is vital." ■

RESEARCH

The Accelerated Gene Identification Project (AGIP)

The Accelerated Gene Identification Project (AGIP) was set up to understand the genetic basis for rare conditions that run in families.

by Kate Pope

Many diseases and syndromes arise from genetic mutation caused by a chromosome or gene change. To date, the genetic causes of many inherited diseases and syndromes are still unknown.

The diagnosis of any illness or syndrome is a heartbreaking experience for any families, especially when the cause of the disease is unknown and there is no cure or treatment. Families such as these could enrol into the AGIP project, where the Bruce Lefroy Centre (BLC) lab and Walter and Eliza Hall Institute (WEHI) bioinformatics teams sought to determine the genetic cause of their disease.

Besides facilitating the development of disease treatment, knowing the genetic mutation of the disease can give vital information as to recurrence risks for the parents, the child, siblings and extended family members, especially in family planning.

The Genetics of Brain Development program (GBD) focuses on children who suffered abnormal brain development. The GDB clinic is held every fortnight with Dr Rick Leventer, a neurologist, and Dr George McGillivray, a geneticist, to help parents to understand the MRI brain scan and the condition of their child's brain and how it would affect their child. Most families are enrolled in the GBD research project so that we can document their information for study either by the BLC lab, or in collaboration with another unit overseas to investigate the genetic cause of their conditions.

Both of these projects have many families that they are working on at various stages in the often long process of discovery. It never ceases to amaze me how willing families are to participate and give that extra little bit.

Improving Schooling for Students with Disability

A new group which brings together parents, teachers, and young people with disability and peak bodies to advise the Gillard Government on issues affecting students with disability has met for the first time today.

Media Release, The Hon Brendan O'Connor MP Minister Assisting for School Education, 14 February 2012

Minister Assisting for School Education Brendan O'Connor opened the inaugural meeting of the Schools Disability Advisory Council, part of the Government's continued commitment to improving education for all students.

"Every child or young person, irrespective of their background, deserves opportunities and support to participate fully in education and their community," Mr O'Connor said.

"Today, the Council will discuss some of the big issues in education including the Disability Standards for Education, funding and specific measures to help students with disability, as well as getting an update on progress towards the collection of nationally consistent data on school students with disability."

The Council, which was announced by Minister for School Education Peter Garrett last year, comprises members from peak organisations representing people with disability, educators and parent groups, as well as academic experts and young people living with disability.

"We will rely on council members' advice on developing policy and implementing school education programs and initiatives to improve the educational outcomes, social inclusion, resilience and wellbeing of students with disability," Mr O'Connor said.

The Gillard Government has a number of important initiatives to improve the quality and consistency of support schools can provide to students with disability, including:

- The $200 million More Support for Students with Disability initiative, with funds to be available to schools this year;
- The collection of nationally consistent data on students with disability to allow more informed and effective government policy supporting students with disability; and
- The Review of School Funding, chaired by David Gonski, which examined the funding support provided to schools working with students with disability.

The full list of members is available at:

http://www.deewr.gov.au/Schooling/Programs/Pages/SchoolsDisabilityAdvisoryCouncil.aspx

To read the full media release online. visit: **http://www.ministers.deewr.gov.au/node/3832** ■

DonateLife Victoria

My name is Twanny Farrugia and I am a volunteer speaker with DonateLife Victoria (organ and tissue donation).

From one person's death, others can live through transplantations of various organs and tissues.

I am passionate in telling others of organ and tissue donation, as I was one of the fortunate ones to receive this Gift of Life 40 years ago when I received a kidney transplant.

I was the 18th patient transplanted at St Vincent Hospital in Melbourne. If it were not for the generosity of the donor family, I would not be here today to offer your organisation a free speaker to inform your members about the joy of giving life to others.

My family is aware of this joy of giving life to others as twenty-six years ago our family completed the circle of the "Gift of Life," as on the death of my father, we became a donor family. Accordingly the circle was complete. I received an organ transplant and my father was an organ donor.

Accordingly if you would like to engage my services as a free speaker I can provide your organisation an overview of the current situation, facts and figures regarding organ and tissue donation in Australia.

Should you require my services, please contact me to arrange a date and time that will suit your club.

For more information please contact:

Mr Twanny Farrugia,
Volunteer Speaker, DonateLife Victoria,
Email: **twannypac@gmail.com** or
Mobile: 0412 107 734

International DNA Day

April 25 marks International DNA Day, which commemorates the discovery of DNA structure and the completion of the Human Genome Project.

By Tarli Bogtstra

Celebrations began each year from 2003, the year the Human Genome Project was completed and 50 years after the discovery of the 'double-helix' structure of DNA.

Cells are the basic building blocks of all life on Earth. Human bodies are made up of approximately 6 billion cells, and each cell in our bodies contains all of our genetic material.

Our genetic material is made up of deoxyribonucleic acid (DNA), an amazing molecule that is the key to life on Earth. DNA is arranged into chromosomes, and each human cell contains 23 pairs of chromosomes. Females have two X chromosomes and males have an X and a Y. Thousands of genes are situated on each of these chromosomes.

DNA is made up of four chemical base pairs, called nucleotide bases, and they comprise the genetic 'alphabet'. The bases are all arranged on the twisted, ladder shaped DNA molecule, called a double helix. The order of the bases determine the meaning of the instructions encoded in the DNA molecule, a little like the order of letters determining the meaning of a word. As a result, genes determine the way in which we all look and function.

James Watson and Francis Crick published an article in the journal "Nature" on April 25 1953 about the double helix structure of DNA. To this date it has proved to be one of the most significant discoveries about DNA. Other scientists also contributed to this achievement, including Rosalind Franklin and Maurice Wilkins who produced X-ray crystallography data of DNA which contributed to the development of the DNA model.

Identifying the DNA structure was a major turning point in the field of genetics. The DNA model helps us understand the way in which genetic information is stored, copied and passed on to future generations. By understanding the chemical nature of DNA it became possible to understand genetic disorders.

The Human Genome Project was an International study focussed on mapping the entire human genome. It was coordinated by the National Institute of Health in the US, however scientists from across the world contributed to the project which was completed on April 25 2003, 13 years after the project began.

The sequencing of the Human Genome is one of the most historic culminations in scientific projects to date. All human genes together are known as the 'genome'. The international projects goal was the complete mapping and understanding of all the genes of human beings.

The Human Genome Project has revealed that there are approximately 23,000 human genes. The completed Genome has been able to map the locations of human genes for major sections of all human chromosomes.

As a result, we now have a resource of detailed information about the structure, organisation and function of the complete set of human genes. It is not a genetic sequence of one person; every individual has a different genome. Rather, the Human Genome is a representative or generic sequence.

Although both of these events, the discovery of the Double Helix and the Human Genome Project, are remembered and celebrated, International DNA Day also promotes learning about other areas of achievement in the genetics and genomics fields. ■

MDA launches National Duchenne MD Registry

The Hon David Davis, MLC, Shadow Minister for Health officially launched the all-new National Duchenne Muscular Dystrophy Registry at the MDA Headquarters in North Melbourne.

The launch was in front of the Australian neurological medical community, including Director of Neurology of the Royal Children's Hospital, Associate Professor Andrew J Kornberg, Scientists and PhD Students from the National Muscular Dystrophy Research Centre (NMDRC), Muscular Dystrophy Association staff and MD family and friends.

Assoc. Prof Andrew J Kornberg, Dr Monique Ryan, Hon David Davis MLC, Boris M Struk, Brandon, Brodie & Kaya

Dr Monique Ryan, the principal driver behind the registry project, provided a deep insight to the long term objectives of the registry and what it means to Australian males who are affected with Duchenne muscular dystrophy.

The National Registry will allow children and young people with DMD have increased access to lifesaving treatments through clinical trials, in not only Australia, but around the world.

The Registry is set to provide valuable centralised information and an interface between patients, doctors and researchers, and at the same time, will be a platform to monitor and benchmark data to improve healthcare performance across institutions and providers.

The DMD registry initiative was driven by patient support and advocacy groups in response to recent developments in genetic technology, and in particular, clinical trials currently being planned, that offer the hope of treatments for this devastating disease.

The Registry, collates the patient's clinical and genetic mutation data to improve the care of DMD patients, and to accelerate the recruitment process for Australian DMD patients into international multicentre clinical trials. The Registry links into the TREAT-NMD global network or registries, opening up opportunities for Australian DMD patients to participate in clinical trials being undertaken anywhere in the world.

The TREAT-NMD global network of national registries has proven effective in improving the health and management of boys with DMD. The next step is to launch registries that will enable rapid access for trials of other neuromuscular and rare diseases such as spinal muscular atrophy (SMA).

According to Muscular Dystrophy Executive Director, Boris M Struk, the new National DMD Registry is a significant step forward in unifying all those affected with MD and provides scientists and clinicians the best tools to match potential treatment with suitable individuals.

"The registry will improve opportunities for international collaboration by facilitating and accelerating recruitment process of Australian DMD patients into new clinical trials and for participating in studies for the benefit of the world DMD community and the advancement of medical science," he said.

"It is hoped that with this bank of information, we will be able to improve the level of care for DMD patients through the co-ordination of diagnosis and therapy and by ensuring new intervention strategies are available on an equitable and consistent manner across Australia."

The National Registry is a significant step forward for the MD community to gain access to the research and techniques used here in Australia and overseas that could offer potential benefits to the Australian MD community.

http://www.mda.org.au/events/mdregister/index.asp

SUPPORT GROUP NEWS

Australasian Tuberous Sclerosis Society (ATSS)

Tuberous Sclerosis Complex (TSC) affects 1 in 6000 people. TSC causes benign tumours to grow in various organs in the body and commonly affects the brain, skin, heart, lungs and kidneys.

Depending on where the tumours grow, TSC can cause epilepsy, developmental delay, autism and a variety of other symptoms. Worldwide research has lead to the discovery of a class of medicines, mTOR inhibitors that are in clinical trials to treat the various symptoms of TSC.

The Therapeutic Goods Administration recently approved the first ever medicine specifically for Tuberous Sclerosis. Afinitor, also known as Everolimus, was recently approved for treatment of subependymal giant cell astrocytomas or SEGAs. SEGAs, a benign brain tumour, affect 15-20 % of people with TSC and typically become symptomatic in children and adolescents. If left untreated, SEGAs can lead to a build up of fluid in the brain and even death. Afinitor has been approved for use in patients that are not candidates for surgical resection.

To learn more about TSC and hear the latest on mTOR inhibitors and other research, people affected by TSC and their families are invited to attend a one day conference. The conference is also open to any interested professionals from the medical, allied health or education areas.

Conference details

Saturday 2 June at The Royal Children's Hospital. For more details, visit **www.atss.org.au** or call 1300 733 435 (Australia only).

The ATSS supports families affected by TSC by providing access to current information, helping families build support networks and contributing to research. Services include a twice yearly journal, phone and email support and regular conference events.

Foundation for Angelman Syndrome Therapeutics Australia

The Foundation for Angelman Syndrome Therapeutics Australia (or FAST) is an organisation of families and professionals dedicated to finding a cure for Angelman Syndrome and related disorders through funding research, education and advocacy.

The Foundation is committed to assisting individuals living with Angelman Syndrome to realise their full potential and quality of life.

Angelman Syndrome (or AS) is a neurodevelopmental disorder affecting approximately 1 in 15,000 live births. Although the cause of AS is known, there are currently no treatments available for this disorder.

FAST is committed to funding the research that will lead to treatments and eventually a cure.

Learn about Angelman Syndrome and why we believe a cure is within reach by visiting **www.cureangelman.org.au/content/2533/WhyThereIsHopeForaCure/**

New look website

Our new website is aimed at empowering families, professionals and researchers to provide the best possible solutions to support those affected by Angelman Syndrome. Our site will give you access to Australian and international information.

To keep up to date with news, announcements and tailored content, see **www.cureangelman.org.au**

Minocycline Clinical Trial

The *Minocycline in the Treatment of Angelman Syndrome* study will examine if the off label administration of minocycline will alter the severity of symptoms associated with Angelman Syndrome.

Minocycline, an FDA approved antibiotic, is traditionally used to treat bacterial infections in several organ systems.

Minocycline is an approved drug in Australia. FAST Australia is working closely with Dr. Weeber and FAST-US to ensure that this trial & any future trials have the ability to be replicated in Australia if required.

You can read more about the trials on our US website **http://cureangelman.org/news/ClinicalTrial/ClinicalTrial.html**

If you have any questions about what this means to you in Australia please do not hesitate to contact us at **info@cureangelman.org.au**

Australian research announcement

FAST Australia together with the Mater Hospital & Mater Medical Research Institute are undertaking our first research project; the Angelman Syndrome Register.

The register will be a "first of its kind" collection of AS patients, related medical professionals and researchers who can be contacted in the event of the development of a relevant research study, participation in a trial, a therapeutic or a cure.

Read more or join here **www.cureangelman.org.au/register**

Management of Charcot Marie Tooth (CMT) in the Classroom

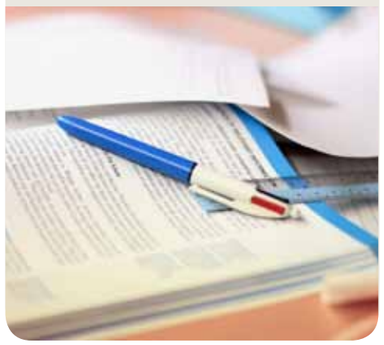
CMT initially presents symptoms in children aged five through fifteen and therefore education may be impacted for sufferers of CMT.

By David Critchely (CMT Sufferer and High School Teacher)

Charcot Marie Tooth Syndrome

Charcot Marie Tooth Syndrome is a physical disability. Learning ability is not affected by having the disease, yet learning ability and social integration may be impacted by the physical and social characteristics of CMT; physical and emotional pain from symptoms such as muscle weakness, fatigue, loss of balance and coordination, tremors, the loss of fine motor skills, a lack of stamina and bullying.

In recent years, physiotherapists, occupational therapist and educators have contributed much to the understanding of CMT and lead CMT people to enjoying more normal lives through tolerance and making the classroom a better place in which to learn while minimising the problems associated with CMT.



CMT initially presents symptoms in children aged five through fifteen and therefore education may be impacted for sufferers of CMT.

CMT does not affect learning ability, as it is a physical disability, or orthopaedic impairment. However secondary problems related to coordination, fatigue, anxiety and emotional factors might impact learning ability. Pain is not a common feature of CMT, but may be due to the secondary effect on the muscles and limbs.

CMT students might be bright but may struggle to complete work in a timely fashion. CMT students may appear weak and lack stamina. Everyday activities such as walking, running, walking up or down stairs, walking on sand, uneven ground or just standing still or standing for long periods of time become difficult.

Even the simplest things like writing, opening books, fastening buttons, turning on a tap or opening a door handle can pose problems.

Bullying, the stigma of 'being different' and the inability to fully participate in many sports and adventurous activities are all part of life for schoolchildren with CMT.

All schools in New South Wales have access to local School Therapy Team supported by the NSW Department of Health who provide consultative services at the student's school, including assessment and therapy. Therapy programming is integrated into the school program making it ideal for students where highlighting and spotlighting need to be avoided.

Writing assessments for class and homework can cause fatigue and pain and there should be consideration for additional time for written work due to slow writing so as not to penalise a student for slow or poor writing skills.

Adaptive equipment such as wrist and forearm supports, foam that fits over pens and pencils that assist in writing support may assist in improving writing but not writing speed.

Similarly access to a Laptop or PC will make reading a students work easier for the teacher, but using a keyboard still presents difficulties to a student without fine motor skills and coordination and there may not be any marked improvement in typing speed over writing.

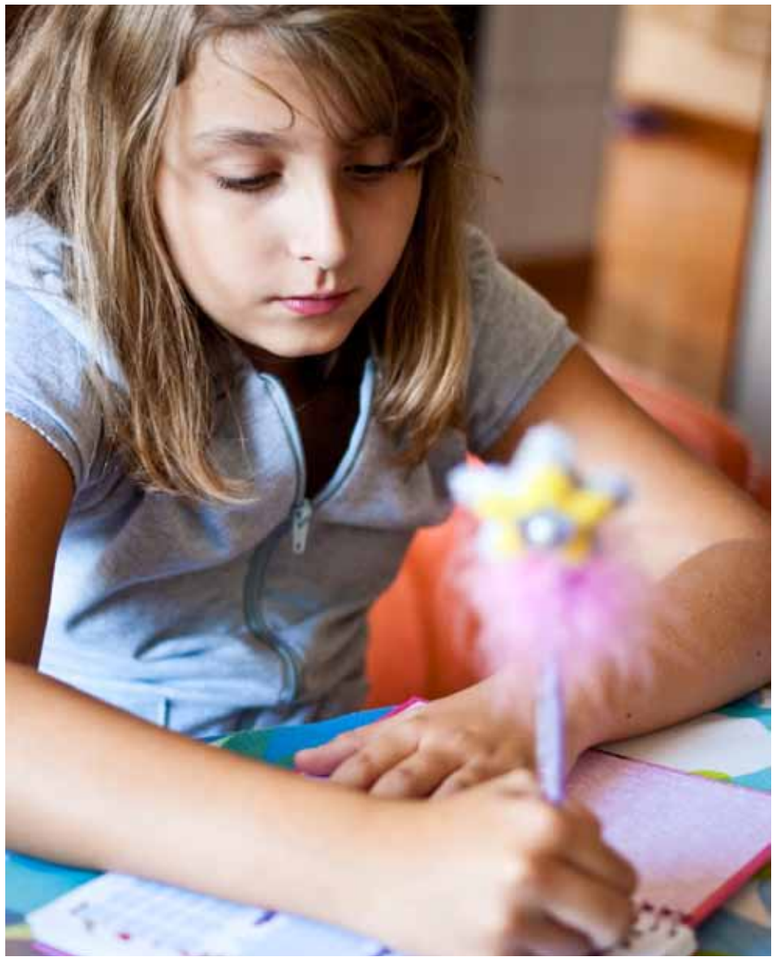
Around the school, foot drop, walking manner and apparent clumsiness will make moving from classroom to classroom tough and climbing or using stairs problematic.

Handrails need to be fitted to all stairways to aid balance and climbing (many CMT sufferers use their hands and arms to pull themselves upstairs to compensate for weakened legs) and additional time allowed if there are long distances between classrooms. In extreme cases, schools may need to provide wheelchair access for CMT students, however as CMT is progressive, it is uncommon to have CMT students who are wheelchair dependent, except when they are recovering from a limb break or surgery.

Carrying heavy textbooks will further slow down a CMT student and the extra weight will increase the risk of trips and falls. Therefore lockers and storage for books and equipment in suitable, accessible locations should be considered to minimise carrying heavy loads. Additionally giving the students two sets of textbooks, one for school use and one for home use may also be beneficial to CMT students.

Physical Education (PE) and playing sport and games is perhaps the most stressful time for CMT sufferers as all the physical characteristics of CMT are fully on display and all inadequacies are highlighted.

It starts in the change-room where changing clothes is difficult due to the loss of fine motor skills. CMT students may need extra time to change clothes and personal assistance or adaptive equipment to do up buttons, tie shoelaces and change trousers. Devices that aid buttoning and zippers are available that will assist CMT students completing tasks. As a minimum a chair must be provided for the student to sit o n while changing.



On the sports field a CMT student will look clumsy, slow and unskilled. Running, jumping, skipping and ball games will highlight the muscle weakness, the lack of coordination and control. They will always be chosen last in a peer picked team as they will be perceived to be of least benefit to a team in the field and this is humiliating and depressing for CMT students. Perhaps they could keep score, umpire (cricket and baseball) or assist off the field rather than being 'on show' in the field.

As an alternative sport, swimming or other low impact sports should be encouraged. Swimming and aqua-aerobics are especially good as the water supports and, to some extent, hides the body.

For Victorian school children with disabilities seeking additional help in the classroom please read the Association for Children with a Disability 2010 FAQ information sheet.

This FAQ information sheet is part of the Inclusive Classroom School Resource published by the Association for Children with a Disability with support from the Department of Education and Early Childhood Development.

For more information go to **www.acd.org.au/inclusive_classroom**

Association for Children with a Disability, Suite 2, 98 Morang Road, Hawthorn VIC 3122

Phone 03 9818 2000 or 1800 654 013 (rural callers)
Email **mail@acd.org.au**
Web **www.acd.org.au** ■

IN BRIEF

NATIONAL CARER RECOGNITION ACT (2010)

In October 2010, The National Carer Recognition Bill passed through both houses of Parliament. The Act formally acknowledges the valuable contribution that carers make to Australian society.

The Act defines what is a carer and sets up reporting and consultation arrangements for certain public service agencies. The 'Statement of Australia's Carers' sets out ten principles to guide how public service agencies and their funding providers should treat and consider carers.

The Act is part of a broader National Carer Recognition Framework that committed the Government to the establishment of national goals relating to carers, a National Carer Strategy, and three year action plans to improve policy and service delivery to better support caring families.

Community Services Minister Mary Wooldridge said the Bill would bring Victoria into line with legislation in other states and territories and the Commonwealth.

ACKNOWLEDGEMENT

The GSNV is proudly supported by the Department of Health Victoria. The GSNV thanks the Murdoch Childrens Research Institute for their ongoing support of our work and the Lord Mayor's Charitable Foundation for the generous grant which has made much of our work possible.

CALENDAR OF EVENTS

The Royal Children's Hospital Good Friday Appeal

Friday April 6
For more details go to the website at **www.goodfridayappeal.com.au**

Charcot Marie Tooth International DNA Day

25 April 2012

Chronic Illness Alliance and Peer Support Network

Inaugural Peer Support Conference:

Friday May 25, 2012
9am to 4.30pm
Multicultural Hub,
506 Elizabeth Street, Melbourne,
(Opposite Queen Vic Market)

Key speaker: Doctor Craig Hassed, Senior Lecturer, Monash University, Dept of General Practice

Social Connectedness and the benefits of psychosocial supports

Contact: Marion Wilde or Denise Sheard on 9882 4654

Conference to be officially opened by:
Andrea Coote, MLC; Member for Southern Metropolitan, Minister's Parliamentary Secretary for Families and Community Services, March, 2012

Circus Skills for All Abilities

Crossing the tightrope Together is a Circus program for boys and girls aged 8–16 years with special needs.

Professional trainers from Westside Circus will teach you and your parent or carer a variety of skills to thrill, including juggling, hoops, tightrope walking and more!

Where: Westside Circus,
Warehouse 29
Cameron Street, Brunswick

To register please contact the Community Access Program Coordinator at Westside Circus on 9383 2299 or email **accessprogram@westsidecircus.org.au**.

Places are limited!

On behalf of GSNV we wish you and your family a safe and happy Easter.