genetic support network of victoria

empowering * connecting * supporting

SPRING 2012

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The role of support groups in facilitating family communication

There are many reasons why it is important to have open communication between parents and children about genetic conditions. Studies have shown that sharing genetic information with children is more beneficial than trying to protect them from the information.

By Emily Higgs

Explaining genetic conditions to children, in an age-appropriate way, is important for correcting misconceptions, demystifying conditions that may otherwise seem confusing or scary, and importantly so that children can make informed decisions about how to look after their health.

Genetic health professionals agree that relevant genetic conditions should be discussed with children and young people, however advice about how to do this is not always offered. This is where support groups can play an important role.

Support groups come in many forms. The Genetic Support Network of Victoria (GSNV) links together over one hundred different support groups. Some support groups are large and well-funded, while others may be made up of only a few people with very little resources. Some support groups are run by groups of parents who have a child with a specific condition, while others involve input from health professionals.

Support groups are valuable for emotional support and practical advice from those with similar experiences, and for reducing feelings of isolation by being surrounded by others 'in the same boat'. Research shows that support groups are a beneficial resource for their members.

A team of researchers from the UK have published a recent study about the role of support groups in facilitating communication within families about genetic conditions. These researchers from the University of Birmingham have been interested in how parents discuss genetic conditions with their at-risk children, and have published many recent articles around this topic. They interviewed affected and unaffected children, siblings and parents from families with a variety of genetic conditions.

The families in this study explained that their support groups didn't usually offer them direct advice about how to talk about genetic conditions in their families.

However, they did feel that attending support group meetings or activities, especially with their children, was a useful starting point to initiate conversations at home about the genetic condition.

Parents generally felt that their child would approach them if they wanted to talk about the condition, however the children interviewed often said they were hesitant to ask their parents questions for fear of upsetting them. This highlights the need for support groups to be used as a tool to open up conversation within families.

The GSNV is aware of the value of support groups in facilitating family communication about genetic conditions.

We promote a range of activities offered by our support groups through our website and newsletter. For example, the upcoming HeartKids Camp where children with Childhood Heart Disease can get together in a fun and safe environment, or Haemophilia

(Cont. page 4)

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Committee of Management 2012

All GSNV Committee positions will be declared vacant on October 18, 2012.

Positions available for your nomination include:

- **President**
- **Vice President**
- **Treasurer**
- **Public Officer**
- **Secretary**
- **General Committee Members (up to 16)**

Current office bearers are eligible for re-election. Nomination forms can be found on the GSNV website, along with the GSNV AGM notice, the agenda and proxy form.

Committee Meeting Dates 2012

Teleconferencing will be available at all meetings.

Thursday October 18 AGM Meeting
Thursday November 15
December (to be confirmed)

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Message from the President

Big picture

Louisa Di Pietro and I recently went to Sydney to meet with our NSW counterparts and also participated in a Rare Voices Australia consultation designed to explore the establishment of a Rare Disease National Peak body.

We discussed and workshopped many issues but it became overwhelmingly clear to me that there are limited funds and resources available and stiff competition for their application in the support sector. The last thing we need is to have more dilution and decreased cooperation among established services and new and emerging ones.

GSNV Inc. sees our role as supporting everyone with a genetic condition – (80% of all 'rare' diseases are genetic) – in having a voice, and making common cause for better participation in society and the best possible standard of not only medical but social support and care. We place value not only on the emphasis of services and better outcomes for support groups seeking to assist their members, but also in supporting those people whose conditions do not attract research or funding and who struggle to gain recognition.

We have to find ways for us and other established umbrella groups to work together wherever there is common ground with any new group that starts out on the same path. We are not competing for top billing – we want to lift as we climb. And we want everyone else to try to do the same. Worked for the women's movement – should work for us.

GSNV AGM (Oct 18 2012)

The Annual General Meeting of GSNV Inc. will be held on 18 October, Level 5 Murdoch Childrens Research Institute. All notices and the agenda have been circulated and we look forward to seeing our members. Guest speakers include a representative from the Department of Health Victoria and a Masters student who will present her research findings on a project directly related to the GSNV.

Please come and please put up your hand to join the committee. It's not hard work especially when it's shared among many.

The GSNV as a not for profit NGO needs the active input of people like you and me who are impacted and affected by genetic conditions and who know what the experience is like.

A committee made up a mix of consumers, professionals, researchers and passionate people makes for a well represented committee. Closer ties with support groups through representation on the GSNV committee assists us in knowing and meeting support group needs. Lots of small voices can join our committee meetings via teleconference, we pay for taxi and other travel to meetings.

We know how hard it can be but how critically important it still is, to speak up and have a voice about the things that really matter in daily life, and to lift our heads above our own navels. We need you. We invite nominations for the GSNV Executive which includes President, Vice President, Treasurer, Secretary and Public Officer as well as the general committee. Please join our committee and have a voice.

GSNV Small Grants

Thank you to every one of you who made submissions for a GSNV Small Grant. It was very exciting to read through the list of projects and award funding. A full list of successful applicants and projects is provided on page 12 of this issue.

We look forward to seeing you at the GSNV AGM!

Moira Rayner
President ■

Message from the team

Welcome to the Spring edition of the GSNV newsletter. It's been a very busy couple of months with a number of education, advocacy and support group activities demanding our attention.

Once again the GSNV attended the 2012 Human Genetics Society of Australasia (HGSA) Annual Scientific Meeting which is always an opportunity to catch up with colleagues and in between plenary and special interest group sessions, discuss and plan future projects.

This year's conference included scientific sessions but also provided some excellent case studies in genetic counselling practice. The GSNV was very pleased to see this year a noticeable contribution and voice from families affected by genetic conditions and what it has meant to them in terms of the demands of the condition and practice outcomes.

Medical genetic testing in Australia is a hot topic at the moment! There is currently significant review on improving national standards for the funding and conducting of medical genetic testing.

There are a number of issues to consider: services and resources, education, training and research, workforce planning, funding, communications between clinical and laboratory services, the utility of investigations, the quality of investigations, consistency across all Australian states on the guidelines and standards of testing, slow progress in translating advances in genetic technology into clinical practice, and ethical and legal matters to name a few!

In a recent edition of GSNV's e-news bulletin Bits & Pieces, we featured a competition designed to get your feedback on access to genetic testing in Australia and issues that may be important for you. The GSNV supports the idea that overall health policy and health objectives should incorporate genetic testing and related clinical genetic services. This is a Federal priority! What do you think?

If you have something important you wish to express in relation to this issue, perhaps you can do so in a 'Letter to the Editor'.

We will be publishing these responses in the summer edition of the GSNV newsletter. This will be the last edition of the newsletter for 2012, so please forward your support group events and news items to us by late November. ■

Save the Date GSNV AGM

As we are at the business end of 2012, we remind you that the GSNV Annual General Meeting is coming up on Thursday 18 October. Please save this date as participation from GSNV members is very important to us.

We are going Green!

The GSNV encourages our members and community to help us become more environmentally friendly and receive our newsletter in electronic version.

Please email **info@gsnv.org.au** and request to receive future editions by email. We also encourage you to forward our newsletter to your own networks and if you receive a print copy of the newsletter, perhaps you could pass it on for others to read. ■

Your Feedback

"I just read your newsletter and wanted to congratulate you and the team on its excellent content and production. I found it very informative, well written and I really enjoyed reading it. Please pass on my congratulations to the team for a job excellently done. I just received the next issue so I look forward to another good read."

"Enjoyed the new format of Bits and Pieces. It is easy to read. Maybe a little more colour e.g. colour of each group when reading the piece. Otherwise it's all good, congrats on a great newsletter."

GSNV

Student Placement Reflection

As part of our training in becoming Genetic Counsellors (University of Melbourne, Master of Genetic Counselling) we are encouraged to take part in a several-week Community Placement where we are given the chance to meet with and help a variety of community groups and networks.

By Amy Schneider and Emily Higgs

This placement is designed to complement our academic studies in providing us with the opportunity to place our theoretical knowledge into a broader community perspective. We both chose a placement with the Genetic Support Network of Victoria (GSNV) because we believed we would be given the chance to help a diverse range of community groups and to learn more about this invaluable network.

As training Genetic Counsellors, we were interested in learning more about the various supports and resources that will be available to us in our future careers, and how we might be able to utilise this knowledge in helping our clients. As well as caring for our clients clinically, we hope to be able to use what we've learnt to provide better support emotionally and psychosocially.

The GSNV website lists support groups in Victoria who have an interest in helping those affected by, or caring for, an individual with a genetic condition. One of the tasks we were responsible for during our placement was updating this support group database. In doing this, we learnt that there are over 100 support groups; from groups for specific conditions to groups dedicated to the wider community. This number was much larger than either of us expected and we are grateful that we are now aware of these groups and will be able to access them when practising Genetic Counselling.

Every few weeks the GSNV e-bulletin, "Bits and Pieces", is released via email to members. By sourcing material for, and writing the August special edition, we realised how powerful a tool this newsletter is in advocating for, promoting awareness of and highlighting the needs of different support groups and their members. It was encouraging to see how enthusiastic the groups were in contacting us with information they wanted us to include in the e-bulletin and we realised how useful the GSNV is in promoting awareness for their support groups.

Amy Schneider and Emily Higgs.

We enjoyed helping many different support groups who are linked with the GSNV with their individual challenges, for example helping the Friends of Sammy Joe Foundation prepare for an upcoming Trivia Night and helping CleftPALS raise awareness of their services in the medical community. We felt inspired that just one morning of our time could help these support groups achieve more than they might have otherwise been able to accomplish, especially when we realised how little resources are available to the small 'kitchen table' support groups of our community.

In the last few weeks we've come to appreciate how many groups are run with limited funding, by time-lacking parents or busy volunteers. Despite these challenges, we have been very impressed by the organisation and local community support these groups manage to maintain.

We are grateful for the insights, advice and supervision we received from Louisa Di Pietro and Lynley Donoghue during our time at GSNV. This placement has provided us, as Genetic Counselling students, with the opportunity to realise the diverse supports and connections available to both ourselves and our future clients. We hope that students to come find the program as valuable as we have. ■

The role of support groups in facilitating family communication

(cont. from page 1)

Awareness Week where activities such as Red Cake Day or casual clothes day are encouraged in your school or workplace. These kinds of activities can be used as great starting points to get families talking about the condition. The GSNV continues to encourage support groups who wish to promote their activities in our newsletter to contact us.

The recent UK study also warns of the possibility of negative effects of being involved in a support group. This includes being reminded of one's own grief when seeing others with more advanced symptoms, or having an unwanted glimpse of the future. Also some members of the UK support groups reported feeling confronted by other members being overly negative about the condition.

The GSNV offers Peer Support Training to address these types of concerns. Our Peer Support Training is a half day workshop available for all individuals, including staff from support groups. Our aim is to help you feel empowered to listen and talk to others who are in a similar position, and share your wisdom and experience in an appropriate and sensitive way.

Many parents, children and health professionals agree that it is important to have open and honest family discussions about genetic conditions. Although recent research has found that support groups don't often offer direct advice about communication, attending support group activities may be a valuable way to initiate a discussion about the condition at home.

The GSNV continues to promote support group activities and involvement, and offers Peer Support Training to empower members to communicate in a useful and appropriate way. As the recent research has highlighted, an increased understanding of the role support groups can play is an important part of empowering, connecting and supporting families with genetic conditions.

Source: Plumridge G, Metcalfe A, Coad J, Gill P. The role of support groups in facilitating families in coping with a genetic condition and in discussion of genetic risk information. Health Expectations, 2011, vol.15, pp.255-266 ■

SERVICES

Creating an ER protocol

When you have a child with a serious medical condition, it's important to have different plans in place to address a situation as it arises.

By Alyson Krokosky

One of those plans should be for your visits to the emergency room. Creating an ER protocol for your child is an important step in streamlining your visit so that all the doctors and nurses in charge of treatment are aware of your child's condition and what is necessary to treat immediately.

This protocol should include:

- Child's name
- Birth date
- Primary or coordinating physician's name and contact information
- Specialist physician's name and contact information (if appropriate)
- Primary or most critical diagnoses
- Any allergies
- General medical considerations
- Physician's signature

Following this information should be instructions for the ER staff regarding testing and care for your child, including the symptoms, vital signs that need to be monitored, necessary tests, medications to give or to avoid, and anything else to avoid in general.

This will help prevent a crisis situation for your child, and help everyone involved provide the best care for your child.

Source: Genetic Alliance Weekly Tip series. Weekly tip #110. ■

GSNV RESOURCES

The GSNV produces a broad range of resources used in a number of different contexts including support groups, community awareness and professional education.

In the GSNV Winter Newsletter, we featured an article on Microarray written by Lynley Donoghue, our Genetic Support Coordinator. This article has been converted into a factsheet and is being used as a resource by genetic clinicians as a guide for discussions with families who undergo microarray testing.

We also hope to provide this resource to other clinicians including paediatricians in the near future to enable families to be provided with accessible and relevant information.

This resource can be obtained by visiting **http://www.gsnv.org.au/media/208612/microarray_factsheet_gsnv.pdf**

The GSNV can also provide and produce a range of different resources that may be helpful for you and your support group.

For further information, or to discuss the need for development of a resource that may not currently exist, please don't hesitate to contact us. ■

Disclaimer The presence of a link to an online resource does not represent an endorsement by GSNV of that site or its content. GSNV assumes no responsibility for the type, amount or quality of assistance, support or information service produced by other agencies or organisations.

Cancer Council Hotline

The Cancer Council Hotline is a valuable resource for not only patients and families but also healthcare professionals and general public.

They are up-to-date with cancer information and use reliable, evidence-based information to support callers. The Hotline is the gateway for many other services including:

- Financial assistance and financial counselling
- Cancer Connect (peer support)
- Victorian Cancer Clinicians Communication Program (an education program for health professionals run by a psychologist)
- Living With Cancer Education Program
- Telephone and Internet Support Groups (teleconference/private chat rooms, eg. for people who live regionally)

Cancer Council Hotline – 13 11 20 or go to **http://www.cancervic.org.au/how-we-can-help/cancer_council_helpline**. ■

Cancer Council Free Wigs

In August this year, the Cancer Information and Support Service (CISS) team was pleased to announce the launch of their new Wig Service, located at the Cancer Council Victoria offices in Carlton, Victoria.

This service offers people undergoing cancer treatment, including children, the opportunity to come in and be fitted with a free wig regardless of where they are being treated across Victoria.

Although, in the past, this service has existed within CISS on a very small scale, the dedication of program manager Alma Hughes, and more recently, a generous donation from Simply Energy, has allowed for expansion of this free service.

For more information, please encourage patients to call Cancer Council Helpline on 13 11 20 or they can visit the Cancer Council offices at 1 Rathdowne Street, Carlton between the hours of 9am-4pm.

Source: CISS News for Health Professionals, 24 August, 2012 ■

Graduating from the Royal Children's Hospital:

The Adolescent Transition Team

Young people who have a chronic health condition have often been treated at a children's hospital and by paediatric staff for much of their lives. Eventually, the time will come when it is necessary to move into the adult health care system and learn to take a more independent role in their own health care and health planning. This process is called adolescent transition.

By Amy Schneider and Emily Higgs – Master of Genetic Counselling 2012

In the past, the transition process has been difficult for many Royal Children's Hospital (RCH) patients and adolescents. In response to patient feedback, and in an attempt to improve the transition experience, the RCH has created a new transition program.

Studies have shown that negative transition experiences can have adverse outcomes on a patient's long-term medical, emotional and social health.

The new RCH transition process strives to help adolescents experience their transition positively, by helping them to develop better skills in communication, decision-making, and self-advocacy.

We interviewed Tim Demos, an Adolescent Transition Youth Mentor at RCH, about his own transition experience and his new role in the adolescent transition team. Tim describes his past transition experience as fortunate. Tim's medical history has been comparatively straight forward and not as complicated as some of the other patients he now sees. Tim's confident and outgoing personality has also allowed him to cope more easily with the change.

However, he was well aware that this process comes with many challenges for others. Some common emotions that young people can experience as they move into adult care are fear of the unknown, worry over not yet knowing how to interact with their new doctors, and apprehension about being alone in the room with a doctor for the first time. Practical issues that they have not had to previously deal with can seem overwhelming; remembering important medical details such as the

names of prescriptions, and learning how to fill out their own medical documents and application forms, such as applying for a Medicare card.

The new RCH transition initiative aims to respond to all of these issues.
RCH patients can be referred to the Adolescent Transition staff who then work collaboratively with the patient's specialty team/s at any age from 12 years onwards, based on the individual needs and preference of the patient.

"Transition is best begun early, in a planned and collaborative manner, that is age and developmentally appropriate, accounting for school and social considerations", says the RCH Transitions webpage.

Often, first contact with the transition team (and normally their specialty nurse or care manager) will be an informal meeting around the age of 15, where the transition process is introduced.

Tim emphasises the importance of introducing young people to the idea of transition as early as possible, to give them the time to adjust and prepare for the future change.

Over the course of several years, adolescents will meet with their assigned Youth Mentor to discuss what to expect and how to cope with the emotional and practical aspects of taking over the role of manager of their own health care.

Tim describes his experience as sudden, as he didn't have this long-term process, and felt "caught off guard" at the prospect of transferring in the next couple of months.

The program is still developing and as such the experiences of the team members are valuable in directing the approach taken in providing this service. Tim enjoys passing on knowledge he learned through his own transition experience, and believes the adolescents he sees benefit from a personal insight on what to expect.

Adolescent Transition Youth Mentors like Tim are also happy to speak with adolescents in the program about other aspects of being a young person with a chronic condition outside of the transition process. Adolescents are not the only ones experiencing a change in their health care role. The team acknowledge the important role that parents play in the transition process and the challenges that may arise for them.

Tim and the other members of the adolescent transition team often talk with parents about how their role will change, rather than disappear altogether; they will always be a parent, carer and support person, however their role as 'medical manager' of their son or daughter will change.

The RCH Transition website, **www.rch.org.au/transition**, contains valuable information for all those involved in an adolescence transition, including the adolescent, their parents, and health care professionals.

For example, there is a Health Care Skills Checklist, Readiness to Transfer Checklist, Goal Setting Worksheet, Young Person Self-Referral Letter Template, and a range of useful fact sheets.

A successful transition into adult care can have lifelong advantages for parents, health care providers and adolescents themselves.

The RCH Adolescent Transition Team has acknowledged this and is aware of the challenges and issues that can become apparent in an individual's transition.

They are well equipped to help adolescents as they 'graduate' from paediatric care and take on the medical world as confident young adults.

We would like to extend a warm thank-you to Tim for his time and insights.

Source: **www.rch.org.au/transition** ■

Tim Demos, Adolescent Transition Youth Mentor at RCH

Should you have your whole genome sequenced?

Have you ever contemplated having a genetic test? Maybe someone in your family has a genetic condition and you've been curious about whether you too might develop it in the future.

Or maybe you've had the opportunity to be part of a research project that involves genetic testing, offered through your workplace, school or university.

It seems that yet another option might soon be available to you; whole genome sequencing.

The race to develop fast and affordable methods for sequencing an entire genome has been on for some time now, and the $1000 genome sequence might soon be within reach for all of us, meaning it would be as affordable as your average computer. The question is; would you want to have your whole genome sequenced?

Direct-to-consumer genetic tests have received much attention over recent years, with companies such as 23andMe and Navigenics offering online genetic tests for anyone who has a few hundred dollars to spare.

Sending off a DNA sample to one of these companies will provide you with a detailed report about your personal risk of developing more than 200 conditions and traits, including breast cancer, lupus, diabetes, alcohol dependence, obesity, schizophrenia, Alzheimer disease and even earwax type.

Companies offering online direct-to-consumer genetic tests currently use targeted genetic testing to provide information about genetic risk, instead of whole genome sequencing.

There's a big difference between these two types of analysis, and it helps to understand the difference by imagining your whole genome is a telephone book with about 3 billion individual letters.

Targeted genetic testing involves turning to specific pages within the telephone book and examining those bits only. Whole genome sequencing, on the other hand, is akin to printing out the entire telephone book. If you're contemplating paying for a whole genome sequence, there are some important facts worth noting.

First, unless your whole genome sequence is accompanied by a detailed report interpreting what it contains (and possibly an experienced geneticist to answer your resulting questions) it will be much like opening up a telephone book in a foreign language.

Second, even with a full report interpreting your genome sequence, current knowledge extends only to a tiny part of the human genome and so, even with an accompanying report to interpret your genome, the vast majority of your genome sequence will remain meaningless and thus be of no use to you, or your doctor.

For example only about 3,500 of the 23,000 genes in the genome have been connected to a particular disease or diseases. The genetic basis for common diseases such as cancer, heart disease and diabetes remains largely unknown.

Third, our knowledge of genetics is evolving so rapidly that the report you receive about your full genome will alter over time as new information becomes known. The report you receive now will be very different from the report you would receive if you waited five years, as much more will be known then, both altering and adding to the information you would receive now.

Fourth, and perhaps most significantly, for a small proportion of people, serious genetic mutations will be found. For some genetic conditions, such as the risk of breast and ovarian cancer due to mutations in the BRCA1 or BRCA2 genes, preventative and treatment options are available to decrease the risk of developing the condition later in life.

However, for other conditions, such as Huntington disease, there is little that can be done to prevent them. Such testing can also identify that a person's parents were closely related, including that the person was the result of incest.

Research indicates that individuals experience a variety of reactions following receipt of a genetic test result. For some people, having information about their future level of genetic risk is comforting, relieving a sense of uncertainty and allowing them to plan for their future and make reproductive decisions with increased knowledge.

For others, the information can be distressing and highly challenging. Most research that has measured the impact of genetic knowledge has studied people who received results in a supportive environment with associated genetic counselling. We therefore know little about the implications of receiving information about multiple genetic conditions at once, with minimal support, advice or counselling.

Finally, most of the genetic diseases and conditions being researched currently occur due to a combination of multiple genetic and environmental influences, meaning that a range of known and unknown genes contribute to their onset, in addition to a range of known and unknown lifestyle factors.

This means that the information provided to individuals is likely to be nothing more than a range of probabilities. For example, you might find out that you have a 20% higher chance of heart attack than the general population.

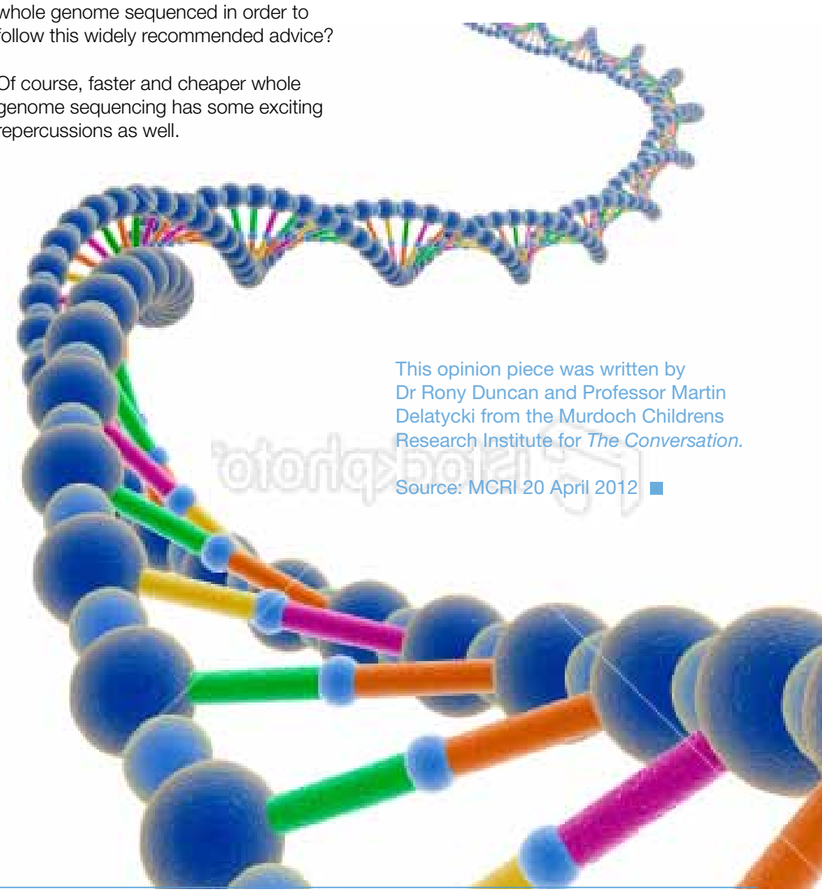
But what will you do with this information? Exercise more? Eat less saturated fat? Stop smoking? Do you really need your whole genome sequenced in order to follow this widely recommended advice?

Of course, faster and cheaper whole genome sequencing has some exciting repercussions as well.

The implications for biomedical research are considerable, and are likely to greatly expand our understanding of the genetic basis of common diseases.

Being able to sequence and then analyse and compare whole genomes in such a fast and inexpensive way is also likely to bring about important advances in our study of pharmacogenomics, where drugs are prescribed specifically to best match an individual's genome, in order to obtain the highest possible benefit.

However, if you are not a biomedical researcher and you are not a clinician wanting to prescribe the most effective drugs possible, whole genome sequencing may well be of limited value ... for now.



This opinion piece was written by Dr Rony Duncan and Professor Martin Delatycki from the Murdoch Childrens Research Institute for *The Conversation.*

Source: MCRI 20 April 2012 ■

Breathe Easy: Cystic fibrosis

Cystic fibrosis (CF) is the most common inherited, life-shortening condition affecting Australian children. It is an autosomal recessive disorder caused by mutations in the cystic fibrosis trans-membrane conductance regulator (CFTR), a transport protein found in epithelial linings.

By Fiona Wylie

Defective CFTR function leads to the build-up of thick sticky mucus in a number of organs, the most problematic of which being the lungs. Bacteria is trapped in the mucus there causing progressive infective disease that eventually damages the lungs so much that normal gas exchange becomes more and more difficult until eventually a lung transplant is the only choice to prolong life.

The last 20 years has seen many new treatments introduced for CF that have improved survival, including Pharmaxis' Bronchitol, but there is still no cure. In order to stay well, people with CF live a very 'medicalised' life, requiring ongoing treatment by a committed and multidisciplinary team comprising the patient, their medical team and their family.

According to Associate Professor John Massie, an invited speaker at this year's Human Genetics Society of Australasia (HGSA) meeting, one of the most positive things to happen in CF care in the last 20-30 years is newborn screening. With first hand-experience of CF disease and its lifelong consequences, Massie has always been a passionate advocate for CF screening, which Australia introduced quite early in 1981, with genetic testing added in the 1990s.

With the CFTR gene identified in 1989, much has been learnt about the mutational genetics of CF, and testing for CFTR gene mutations is now reliable and relatively inexpensive. Australia was the first country to have a nationwide genetic test available for CF incorporated into newborn screening programs for the early identification of affected individuals.

Newborn screening ensures the early diagnosis of most affected individuals, and enables parents of affected individuals to be identified as carriers.

Options with regard to further children can then be discussed, including pre-implantation genetic diagnosis and identification of carriers by genetic testing.

"Our initial research in this area told us that of families who have a baby with CF and who know all about the testing available, about 80 per cent elect to have more children. And of those 80 per cent, the majority elects to use prenatal testing on the next pregnancy. So people who are informed about CF choose to do something about it and use that information."

Carrier screening

How and when carrier screening should be offered and/or recommended by doctors has been a subject of study by Massie and others including the HGSA in Australia for many years.

"In the US, carrier screening for CF was recommended by the regulatory and medical bodies in 2001, with a subsequent 50 per cent reduction in the incidence of affected infants," says Massie.

Similar programs have been since introduced in other countries (including Scotland, Denmark and England) and uptake rates are generally up around the 80 per cent mark. Such results provided more fuel for Massie and his colleagues in advocating a national policy and funding scheme for CF carrier screening in Australia.

In the absence of a federally supported program and to gather better data on such efforts in Australia, the Victorian Clinical Genetics Service initiated a pilot carrier-screening program for CF in 2006. It was offered to couples planning a pregnancy or in early pregnancy on a fee-for-service basis. Attitudes towards the program from those offered the test were generally positive, particularly if accompanied by their doctor's recommendation and the understanding of CF as a severe condition.

"We also described getting those disconnected elements together and starting a program that then represented genetics, CF and the CF organisation and a sympathetic ear from the obstetricians. But we couldn't convince the state government to fund it, and we didn't have health economic data at the time, hence the fee.

"We then put it out there to obstetricians and then GPs. We have had various seminars, mail outs and talk fests and things and the literature to try and spread the word. So far, the program has had fairly stable numbers of 1500 women per year. Now we want to really step it up and make it broader."

Massie also wants to standardise how the information is distributed, which now differs widely amongst those offering it. "GPs would be an ideal source, but if we want to target the pre-pregnancy stage, not many people in Australia really turn up for pre-pregnancy advice.

"Thus, we need better access to information from a range of sources, but I think actually the bottom line is that a lot of people take direction from their doctor. And I think that governments coming on board would sort of give it that extra stamp of approval in peoples' minds."

Massie has also noted that the HGSA has put out a position paper on the CF carrier screening program, available to view on the HGSA website. "It's very carefully worded to focus on the provision of appropriate knowledge and information. Basically, we want to get everyone informed, and not advocate that everyone get screened. Then what people do with the information is their business."

"In my opinion, we as clinicians all have an obligation to inform our patients about the available options, especially when it comes to pregnancy. It doesn't take very long. If you provide 100 per cent information and get 70 per cent uptake, you might find 50 per cent reduction in incidence. This is not about eugenics – it's not trying to save the health care system – this is a personal choice by individuals. So, that's what we are trying to do: simply offer people choice."

Associate Professor John Massie is the Head of Education and Training in Respiratory Medicine at the Royal Children's Hospital in Melbourne.

He is also a clinical faculty member at the University of Melbourne and a Research Fellow at the Murdoch Children's Research Institute. Massie has published extensively in the area of CF diagnosis and population screening, and helped to establish the population-based carrier-screening program for CF in Victoria.

Massie was also on the subcommittee involved in developing the HGSA policy on CF carrier screening.

For the full version of this article please refer to: **http://www.lifescientist.com.au/article/435293/feature_breathing_easy/**

This was reprinted with permission from the editor Tim Dean. Original article was published on 03 September, 2012 in the July/August 2012 edition of Australian Life Scientist Magazine. **www.lifescientist.com.au** ■

Research Study

RMIT University, School of Health Sciences, Honours Research – Meaning Making and Family Quality of Life

This project focuses on understanding families' experiences of quality of life and how they give meaning to their lives when experiencing stressful parenting.

To participate in this study, all you need to do is complete an 'Expression of Interest for Parent Participants' and a very quick questionnaire.

For full details, please contact Amanda Yee at **s3359448@student.rmit.edu.au**.


RMIT UNIVERSITY

NEW RESEARCH PROJECT

A WALK IN YOUR FAMILY'S SHOES

THANK YOU FOR YOUR INTEREST!

Where GSNV has been

July

- GSNV New Team Commences
- RCH Grand Rounds Seminar: Treating Childhood Obesity
- Seminar: Molecular Medicine
- Human Genetics Society of Australasia (HGSA) Conference Canberra
- Master of Genetic Counselling GSNV Tutorial

August

- Whitehorse Community Health Service: GSNV Presentation on Rare Diseases
- DOHA: Genetics Working Party Consultation
- Melbourne University Public Health Genomics Course – GSNV Presentation

September

- Syndromes Without a Name Support Group Meeting
- GSNV Friday Seminar: 'Supporting Victorians with Cancer'
- Rare Voices Australia Consultation, Sydney
- Friends of Sammy Joe Trivia Night
- Australian Catholic University (ACU) Presentation: Human Genetics
- PSA Education Session

GSNV SMALL GRANTS

The annual GSNV Small Grants Scheme

The GSNV recently called for applications for the annual GSNV Small Grants Scheme. This year, we opened applications to all support groups who are listed on the GSNV support group directory, regardless of financial membership status. The Committee of Management recently reviewed all applications against the criteria, and we are proud to announce that this year we are funding 11 projects.

Charcot Marie Tooth Association of Australia (CMTAA)

CMTAA inaugural youth weekend and Camp – Assistance for two Victorian attendees

iCare Special Needs Group

General costs and assistance with organising and providing a networking luncheon for carers

Introfish Inc

General costs and assistance with fishing excursions

Syndromes Without a Name (SWAN)

Assistance with start up costs - web domain, printing of brochures, governance and set up

Ballarat Lymphoedema Education & Exercise Support Group

Ongoing costs - paper and ink for education material that is provided at support group meetings

Australian Disorders of the Corpus Callosum

Financial assistance for a facilitated meeting in Victoria with all 11 AusDocc committee members from different states in attendance

Muscular Dystrophy Association

Assistance with a project which aims to translate selected MDA publications from English to Vietnamese and Sudanese

Metabolic Dietary Disorders Association (MDDA)

Cost of organising a National Youth Wellness Retreat. Retreat is designed to educate young people, with Inborn Errors of Metabolism about their diet management and exercise requirements in order for them to maintain fitness and health for their particular metabolic disorders.

Cleft Palate & Lip Society of Victoria

Funding for Cleftstars, specifically the costs of transfers and flights for two eligible Tasmanian Cleftstars members

Wiliam's Syndrome Family Support Group (VIC)

Seeking funding for camp activities - sea kayaking and boat tours

Klinefelters Support Group of WA

Travel assistance to attend a KS seminar in Vic hosted by the GSNV ■

SUPPORT GROUP NEWS

Fabry's patient id care

The Fabry Support Group of Australia has developed an excellent resource for Fabry patients.

The Fabry Disease alert card is wallet sized, contains essential details on Fabry disease, symptoms and organ association. Much like a diabetes alert card or medical alert bracelet, this simple and effective resource can help in emergency situations and when medical information needs to be communicated.

For rare and complex conditions this basic information is a practical and useful tool for patients and health professionals alike. ■

The Friends of Sammy-Joe Foundation Market Day

The Friends of Sammy-Joe Foundation is holding a Market Day.

Sunday 28 October, 2012, 9.30am – 4.30pm
Craigieburn Leisure Centre
127 Craigieburn Road, Craigieburn, VIC

As part of the Market day, there will be a raffle and auctioning of items to raise funds to the Foundation. Set up a stall representing your organisation and donate a gift or voucher towards the raffle. For more information visit **www.friendsofsammyjoe.org**. ■

GSNV Seminar: Klinefelter syndrome

The GSNV held a Friday Morning Seminar in conjunction with VCGS, entitled 'Klinefelter syndrome: clinical and personal perspectives'.

The seminar consisted of a clinical perspective, the research findings of a study investigating the experiences of parents whose child is diagnosed with KS, and three individuals with KS who generously shared their stories.

This seminar was a great success and we welcome input from support groups and individuals who would like to share their stories at GSNV hosted seminars in 2013. ■

Marfan Forum – 'From a Parent Perspective'

The Marfan Association of Victoria has announced their AGM and family fun day.

Sunday 28 October, 2012, 11am to 3pm
Lilydale Lakes, 435 Swansea Road, Lilydale, VIC

This forum is designed to give parents an opportunity to share their experiences on caring for children with Marfan and maybe even help others living with the challenges of Marfan Syndrome. For more information, please visit **www.marfanvic.org.au**.

Many support groups use the AGM process and date to organise a social gathering, family friendly activities and peer support discussions. If you support group requires some information and or support in organising an AGM or group activity, please contact the GSNV. ■

GSNV Peer Support Training

GSNV is excited to announce this year's Peer Support Training session is to be held on Tuesday 4 December, 2012.

This workshop is available for all individuals, including staff from support groups. It is designed to empower you to help those that are undergoing difficult situations. Some areas that the peer support training course will cover include:

- active listening
- communication skills
- empowerment
- loss and grief
- self-care
- community resources
- information about GSNV and what we can do to help the process

Please note a nominal fee to cover GSNV charges will be imposed on the training. For more information, please contact the GSNV office on (03) 8341 6315. Places are limited and we advise you to make a booking. Further details will be announced closer to the date. ■

Help launch a National Disability Insurance Scheme

People with disability, and people who work with and support them, are being encouraged to express their interest in working for the NDIS Launch Transition Agency.

The Australian Government is investing $1 billion in the first stage of a National Disability Insurance Scheme, which will start from July next year.

Around 20,000 people with disability, their families and carers living in launch sites in the ACT, South Australia, Tasmania, the Barwon region of Victoria and the Hunter in NSW will benefit from the first stage of the scheme.

The NDIS Transition Agency is now seeking people to fill a range of roles, including in management, leadership and finance as well as the vital roles of local area coordinators and customer service officers.

Expressions of interest are also being sought from people who have allied health qualifications who can help people with disability plan for their current and future needs.

The Government is particularly encouraging applications from people with disability or people who have worked with or supported people with disability to submit an expression of interest. This lived experience of disability will be an important part of delivering the scheme on the ground.

Agency positions will be available across all five launch locations and are expected to commence from early to mid-2013, with a small number starting in the coming weeks or months.

The Agency will match people with the required skills and experience from the Expression of Interest list with vacant positions as they become available.

For more information, go to **www.ndis.gov.au**

Source: Joint media release, 19 September 2012, Hon Jenny Macklin MP, Minister for Families, Community Services and Indigenous Affairs and Minister for Disability Reform, and Senator Jan McLucas, Senator for Queensland. ■

Join us for a DisabiliTea

To support the NDIS

Friday 26 October 2012

The Every Australian Counts campaign for the National Disability Insurance Scheme (NDIS) will hold its second annual 'DisabiliTEA', a nationwide tea party.

The GSNV continues to support both the NDIS and Every Australian Counts Campaign and will be hosting our own DisabiliTEA at the Murdoch Institute.

The GSNV encourages all support groups and the general community to have a cuppa and also support the NDIS as a national priority.

Registrations are now closed but late registrations will be accepted by emailing **info@everyaustraliancounts.com.au**.

Or, if you would like to attend an event, please go to **www.everyaustraliancounts.com.au** to participate in a DisabiliTEA in your area.

The more people who participate in this event, the more likely our politicians will hear our call for the NDIS. ■

MEDIA RELEASE: Consumers welcome better access to medicines

"Health consumers across Australia can now rest easier knowing their access to recommended medications will not be deferred by Federal Cabinet unless the cost of new medications is above $10 million per annum," said Carol Bennett, CEO of the Consumers Health Forum (CHF).

Following extended discussion between CHF, the Minister and other key industry stakeholders, the Government has announced an extension of the moratorium on deferrals of medicines with a cost of under $10 million per year until the expiry of the current Memorandum of Understanding with Medicines Australia, due in June 2014.

CHF led a campaign to overturn a previous Federal Cabinet decision that saw thousands of Australians unable to afford important medication after listing of recommended medications on the subsidised Pharmaceutical Benefits Scheme (PBS) was delayed.

In welcoming the announcement Ms Bennett said, "This announcement is a victory for common sense and a win for health consumers. I congratulate the Federal Government, particularly Prime Minister Julia Gillard and Health Minister Tanya Plibersek, for coming to the table and negotiating a sensible outcome."

For the full media release, please see **https://www.chf.org.au/pdfs/med/med-280912---Consumers-welcome-better-access-to-medicines.pdf**. Reprinted with permission from Consumer Health Forum. Media Release, 28 September, 2012. ■

NATIONAL FOCUS

Volunteers worth more to Australia than mining

A University of Adelaide study has found that volunteering in Australia is now worth more than the mining industry, declaring the true extent of its monetary value to be more than $200 billion a year.

Its economic contribution to Australian society outstrips revenue sources from mining, agriculture and the retail sector, according to Dr Lisel O'Dwyer, a Senior Research Associate in the University's School of Social Sciences.

"More than 6.4 million people volunteer their time in Australia, which is double the number in 1995. And with the looming retirement of the first wave of baby boomers, these figures are likely to increase at an even more rapid rate," Dr O'Dwyer says.

"There are many ways to measure the value of volunteering and the benefits flow both ways. Volunteers get a lot of satisfaction from helping others, enhancing the quality of their life and their health. The benefits to the recipients are obvious and there are also positive spin-offs for governments and workplaces."

"The value of volunteering is difficult to measure. Volunteers gain a broad range of new skills that are transferable to their workplace, for example. They are healthier, fitter, more mentally alert and more socially connected than people who do not volunteer. These benefits may even act as a pathway to employment," Dr O'Dwyer says.

She says current estimates relating to the economic value of volunteering are likely to be "gross under-representations" but warns that focusing on the monetary value may even be damaging if it reinforces the notion that volunteering is all about saving money.

"The research conservatively calculated the hourly rate for volunteers at around $7 per hour or 25 per cent of the equivalent paid job".

Dr O'Dwyer said a previous study in 2003 by Dr **Peter Mayer** from the University of Adelaide revealed one of the less tangible, potential effects of volunteering is a reduced crime rate.

Dr Mayer's study suggested that even a one per cent increase in social capital (including volunteering) was likely to lead to falls in homicides, sexual assaults, burglaries and vehicle thefts.

"If a volunteer fire fighter saves the life of a child, what is that worth? If environmental degradation is slowed because of millions of trees planted by volunteer conservationists, what is that worth? And if an elderly person receives a hot meal five days a week, what is that worth?

According to **Volunteering Australia**, people aged between 40-54 comprise the highest bracket of volunteers, with slightly more women (40 per cent) than men (37 per cent) giving their time to voluntary work.

Dr O'Dwyer's research findings form a chapter in a forthcoming book, Positive Ageing: Think Volunteering, which will be published by Volunteering SA & NT later this year.

For full version, please visit **http://www.probonoaustralia.com.au/news/2012/08/volunteers-worth-more-australia-mining**. Reproduced with permission. ■

GSNV Volunteering Program

The GSNV, in conjunction with the Master of Genetic Counselling program (University of Melbourne) is working on introducing a new Student Volunteer Program.

This program is an opportunity for support groups to source suitable volunteers to help them with small tasks. This program will be rolled out between late 2012 and early 2013 and the GSNV looks forward to informing members on how we may provide some volunteer time.

Please stay tuned for further information as this program is still in development stages.

IN BRIEF

ABOUT THE GSNV

The Genetic Support Network Victoria (GSNV) is a vibrant and active organisation committed to promoting the interests and well-being of people affected by genetic conditions.

We assist individuals and families with the 'human', non clinical side of genetic diagnosis, living with a genetic condition, and gaining access to supports and services.

The GSNV assists in the set up of new support groups and for those already established, helps to broaden awareness and assist in their activities. The GSNV is committed to improving the sense of isolation the community may feel in dealing with genetic conditions.

The GSNV is closely associated with a wide range of support groups, clinical genetic services and peak professional bodies.

ACKNOWLEDGEMENT

The GSNV is proudly supported by the Department of Heath Victoria.

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

GENETIC SUPPORT & ADVOCACY

Peer Support Requests

The GSNV works hard to connect individuals and families interested in sharing their experiences and insights with others. People interested in contacting others "in the same boat" can advertise their details through the GSNV and we will assist in making connections.

SEEKING CONTACT:

Individuals with the below conditions are seeking contact with others:

- Osteogenesis Imperfecta
- Ring Chromosome 8
- Langer-Giedion syndrome
- Hereditary Sensory Autonomic Neuropathy
- Incontinentia Pigmenti
- Palmar Plantar Keratoderma
- Undiagnosed Genetic Condition
- Carriers of balanced translocations
- Stickler syndrome

Contact the GSNV office by phoning (03) 8341 6315 or by emailing **lynley.donoghue@vcgs.org.au** if you are interested in making a peer connection. ■

CALENDAR OF EVENTS

Huntington's disease Charity Gala Ball
October 12

Thalassaemia Australia Inc AGM
October 16
For more information visit
www.thalassaemia.org.au

GSNV Annual General Meeting 2012
October 18

VCFS 22q11 Forum
October 20
Monash Medical Centre
For more information visit
www.vcfsfa.org.au

DisabiliTEA
October 26

FSHD "I Believe in Miracles" Gala
October 27

The Friends of Sammy-Joe Foundation Market Day
October 28
Craigieburn Leisure Centre
For more information visit
www.friendsofsammyjoe.org

Marfan Ass. Vic. Family Fun Day and AGM
October 28
Lilydale Lakes
For more information visit
www.marfanvic.org.au

National Cleft Awareness Week
November 8-14

CleftPALS Victoria Family Picnic Day
November 11
Diamond Valley Railway
For more information visit
www.cleftpalsvic.com

Fabry Support Group Australia AGM
November 10
For more information visit
www.fabry.com.au

GSNV Peer Support Training
December 4