Child health inequalities exist in all western countries including Australia and constitute a significant and potentially preventable public health problem.

Although the data are limited, in Australia we know the predictors of future health inequities – differential health outcomes that are unjust, unnecessary and unacceptable, and potentially preventable – begin in utero.

The Australian Early Development Index (AEDI) is a cross-sectional population census of early childhood development in communities across Australia. It provides the first national data on developmental outcomes for a population of five year olds. There are subgroups of Australian children who are much more likely to experience developmental vulnerability. They include:

- children with additional health and developmental needs
- children from language backgrounds other than English
- Aboriginal and Torres Strait Islander children
- children from the most disadvantaged SES communities
- those living in remote areas of Australia.

Alternately, children from the most advantaged SES communities, and children living in major cities of Australia were less likely to experience developmental vulnerability.

Children with additional health and developmental needs

Data from the AEDI suggests that around one fifth of five-year old children experience additional health and developmental needs, including 4 per cent with well-established and formally diagnosed conditions, and 18 per cent with emerging developmental problems.

This includes children with a wide variety of conditions impacting primarily on their physical health (for example, diabetes, epilepsy) and psychosocial wellbeing (such as Attention Deficit Hyperactivity Disorder and autism spectrum disorders).

The percentage of children with and without additional health and developmental needs on each of the AEDI domains (AEDI 2009)

Cont. page 8
Message from the President

Kay Timmins was born and raised in Ballarat, Victoria. She has worked in the fields of nursing, aged care and rehabilitation, as well as setting up multiple support groups for lymphoedema across Victoria. Kay currently continues to study and sit on various committees involving disability, advocacy, governance and policy and procedures regarding funding grants. The GSNV thanks Moira Rayner for her great contribution as she steps down as President and welcomes Kay Timmins to the role.

As the new President of the GSNV Committee, I would like to see us in 2014 working as a team to move forward in our policies and achieve our strategic goals.

We have a unique relationship with the Murdoch Childrens Research Institute which has encouraged us to develop more expertise among its members and a professional approach to strategic planning and committee business.

We have some new members on board and look forward to settling into a new rhythm, hearing some fresh ideas, and exploiting the skills and enthusiasm of us all.

I would like to thank Moira and the previous committee for their ongoing hard work and commitment, especially through our growth spurt in 2013, and I hope that we are able to continue their good work in the coming year. It is great to see a turnover of positions on the committee and some new members, without losing any previous members.

I hope that the new committee brings new and exciting ideas to the floor, so that we can achieve our goals for the coming year of 2014. I believe in team work and for us all to help each other as much as possible.

Kay Timmins
President of GSNV Committee

In memory of Maria Kastoras

Christmas Day 2013 will forever be etched in the memories of the haemoglobinopathy community in Australia and indeed around the globe. On this day we sadly said goodbye to our dear friend, advocate and ‘mother of all Thals’, Mrs Maria Kastoras. Thalassaemia Australia has lost a great fighter and advocate as Maria was the consummate ambassador, passion, vigour and drive defined her approach and her dedication to all those affected by genetic blood disorders. Maria was an educator, peer supporter, international representative and above all unwavering friend. Her smile, effervescence and contagious laugh are her eternal epitaph. Maria lived life to the fullest and although personally and professionally dedicated to the cause, was not defined by Thalassaemia.

Many in the research and support communities knew Maria and have been touched by her passing. In her memory we will strive to continue the work she loved and make a difference for the ‘littlies’, the children in the haemoglobinopathy community around the world that she so loved. We will remember her work as a dedicated Red Cross Blood Services ambassador and her dedication to thanking and acknowledging blood donors and recruiting new donors to give ‘the precious drop’.

I am deeply saddened by the loss of my dear friend, colleague and peer and will forever cherish the work we did together, the connections we created and the inroads we made.

Louisa Di Pietro,
Group Leader
Genetic Support Network of Victoria

Committee Meeting Dates 2014

Teleconferencing will be available at all meetings.

Thursday 20 February
Thursday 20 March
Thursday 17 April
Thursday 15 May
Thursday 19 June
Thursday 17 July
Thursday 21 August
Thursday 18 September

The information in this newsletter is provided by the Genetic Support Network of Victoria for educational/informational purposes only. It is not a substitute for professional medical care and medical advice. The contents express the opinions of the authors who alone are responsible for their views expressed. GSNV does not accept any legal responsibility for their contents.
Welcome to the first edition of the GSNV Newsletter for 2014. The year is well underway and our team are busy preparing for important calendar and awareness dates, education and special seminars and our responses to Victorians seeking assistance and support.

Message from the team

Welcome to the first edition of the GSNV Newsletter for 2014. The year is well underway and our team are busy preparing for important calendar and awareness dates, education and special seminars and our responses to Victorians seeking assistance and support.

Rare Disease Day is only 5 weeks away and we invite you to think about this international day of recognition and how you might honour it. The GSNV is planning a special seminar with details posted on our website. We hope to see our members and community there. All our welcome!

I take this opportunity to formally welcome our new Committee members who have been inducted subsequent to our October 2013 Annual General Meeting. We have 15 Committee members this term and representation from a cross section of interests, experience and skills.

I look forward to working with our committee to successfully deliver our strategic plan and business objectives, but overall improve the quality and services delivered by the GSNV to all Victorians affected by genetic conditions.

We sadly say goodbye to a valued and respected member of our team, Ms Marleen Susman. As an experienced and senior cytogeneticist, having completed a PhD and genetic counselling training and with a wealth of experience, Marleen added greatly to the capacity, skills set and output of the GSNV. We duly acknowledge her contribution to our work and wish her all the very best for the future.

The GSNV will be recruiting to fill the position of ‘Genetic Support and Education Coordinator’ in the coming weeks. We welcome expressions of interest via email info@gsnv.org.au. This position will be advertised via the Human Genetics Society of Australasia (HGSa), Australasian Society of Genetic Counsellors (ASGC) and the Murdoch Childrens Research Institute (MCRI).

The current GSNV team comprises myself, Nancy Amin, Emily Higgs and Catherine Beard (sometimes) plus our valued and appreciated volunteers. We look forward to bringing you news, information and education in the coming months.

Louisa di Pietro, Group Leader

GSNV membership

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

Membership entitles you to:
- Newsletter and eNews
- Genetic support and peer connection
- Information and education
- Free Peer Support Training
- Advocacy on behalf of members


Rare Disease Day®

28 February 2014

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

In Australia, Rare Disease Day has taken on the momentum it deserves and people from all states and territories gather to celebrate and raise awareness.

The GSNV will be hosting a Rare Disease Day event that will be designed to bring together the Royal Children’s Hospital (RCH) and Murdoch Childrens Research Institute (MCRI) communities, but also focus on the important work of support groups in Victoria. Stay tuned for more details on how you can join the GSNV on Rare Disease Day in 2014 and play a part in celebrating this important day.

For information on what’s happening around the world on Rare Disease Day go to www.rarediseaseday.org/

RESEARCH STUDY

Are you a parent or caregiver to a child with a neurogenetic condition such as 22q11.2 deletion syndrome, Prader-Willi syndrome, Down syndrome, an as yet undiagnosed syndrome or other neurogenetic syndromes?

Following on from an earlier study, the University of Newcastle would like to further understand how your child has impacted your life and how you have coped. You can do this study even if you participated in the Disclosure study in 2012/2013.

To read more, or complete the survey click the link below: www.wix.com/c3094005/geneticdisorders

Your Feedback

“I enthusiastically read each addition of the GSNV newsletter, I just wanted to sincerely thank everyone that contributes to this important publication. It’s such a vital service that is being provided and I’m truly grateful, I love learning about the latest research being conducted and services and support available. It makes a huge difference to families like mine and provides us with hope and a feeling of being more in control and able to handle our daughter’s condition.”

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Louisa di Pietro, Group Leader
‘Doing it with us, not for us’

The CHF’s Hip Pocket Pain campaign – creating an equitable health system

By Hanna Leslie, BSc (Genetics), LLB, MGenCouns (Yr 2 student)

Hanna is a Master of Genetic Counselling student at the University of Melbourne, and a volunteer in the GSNV office. She is also a general GSNV Committee member. She enjoys drawing on her experiences in care and support work, professional development program management, genetic counselling studies, and legal advocacy, and channelling her energies towards enabling informed discussion and choices, particularly around the ethical, legal and social implications of genetic technologies and information.

What is the Hip Pocket Pain Campaign?

In early 2013 the Consumers Health Forum (CHF) launched the ‘Hip Pocket Pain’ campaign which calls for action to address Australia’s internationally high and arguably inequitable ‘out of pocket’ healthcare costs. With headlines such as ‘Hip pocket pain is hitting Australians hard’ and ‘Healthcare is universal for those who can pay’ this campaign highlights an emerging two-tiered health system and lack of public awareness of the ‘plight’ of those living with a serious illness and struggling to pay their significant medical costs.

The campaign has broad application to all Australian healthcare consumers, with a special focus on the elderly and those with chronic illnesses. It is extremely important for the GSNV to support this and other similar campaigns, such as the CHF’s Medicines Savings campaign, in order to provide a consumer voice for individuals and families who face the costs associated with a genetic condition, and in particular those with a rare condition. The emotional, financial and social challenges faced by families affected by a rare condition can be burdensome and often overwhelming.

Hip pocket pain for those with rare genetic conditions

Our cover story for our Autumn 2012 newsletter, “GSNV’s Rare Disease Day Seminar”4, discussed the GSNV’s work to support people living with a rare condition, including the non-clinical or ‘psychosocial’ impact and challenges. Our members survey placed ‘access to specialist doctors, treatments and medication’ as the first of the Top 5 Issues for people affected by a rare genetic condition. Anna Hickey, who spoke at the Rare Disease Day Seminar on 2 March 2013 about her experiences of having Gorlin Syndrome, says the main issue for her has been “the toedium of continual medical appointments; (they are) draining physically, emotionally and financially”.

For those where medicines and therapeutics exist, the out of pocket costs can be significant – orphan drugs are not usually available through the PBS or Medicare, and if one cannot access a drug through the Life Saving Drugs Program (LSDP), clinical trials, or drug companies’ compassionate access programs, they can find themselves in the desperate situation of being precluded from accessing a known treatment or being denied hope, unless they can pay from their own pocket.

People in this position are vulnerable healthcare consumers – rather than being supported by the system, they are borrowing from family and friends, remortgaging houses, fundraising, or using superannuation or inheritance money. Nearly 1 in 10 Australians don’t fill their prescriptions because they are too expensive, but for those with a rare genetic condition who cannot pay, this may be a question of forgoing a life saving medication, a predicament the general community may not truly understand or appreciate.

More than hip pocket pain: The pain of inequity

Many people can understand the emotional impact of living with financial hardship, but the Hip Pocket Pain campaign encourages us to imagine and appreciate the greater challenges faced by those who experience the cumulative effect of:

• significantly higher financial costs
• coping with other unique and often marginalising and stigmatising aspects of living with a rare condition
• the emotional and social impact of directly experiencing health care inequity, through being unable to access necessary treatments

Often the heart of this debate focuses on challenging moral and ethical questions about which conditions or medications (or more crudely, whose lives) should be prioritised within a system with limited funds? Having the value of one’s life questioned can be demeaning, dehumanising, and an affront to individual dignity and autonomy, irrespective of health economics rationales. This can compound and augment feelings of isolation, marginalisation and stigmatisation.

Hence we are encouraged to reframe the discussion as “How can the system change to ensure the limited resources are allocated to all who need them?” – requiring a meaningful public conversation about our definition of ‘health equity’.

The Chronic Illness Alliance takes the view that in order to have a more productive, equitable and healthy nation, health
reform must be ‘person-centred’: it must go beyond being only concerned with health costs and delivering health services to valuing those with chronic illness as community members, and focusing on investing in people’s lives.

The Hip Pocket Pain campaign may convey a focus on the pain of having less loose change in your hip pocket. However, addressing the real underlying systemic issues of Australia’s increasingly inequitable healthcare system will alleviate more than hip pocket pain: It will go a significant way towards lifting the additional emotional and social burden borne by those, who through no fault of their own, are currently subjected to marginalisation and inequity in more ways than one.

CASE STUDY

Myozyme®: enzyme replacement therapy for Pompe disease

Mr Raymond Saich, President of the Australian Pompe’s Association, uses the example of a 12 year old child who is classified as having adult onset Pompe Disease. The cost of his medication – $400,000 out of pocket per year – is prohibitive for his family.

“Every day he goes without treatment his young muscles are further, irreparably damaged. I am sure his parents watch the postman and start at every telephone call hoping that today will be the day he gets treatment approved.”

“Pompe Disease is one rare disease that can be treated and we ask that people talk to their friends and neighbours, and when they see their local member of parliament, remind them that there are 30 Australians with Pompe disease who need some help.”

Examples of this are the Department of Health’s ‘Doing it with us, not for us’ consumer participation policy and the Commonwealth Government’s support for the ‘Our Health, Our Community’ project, which includes an online consumers’ forum specifically established to enable patients, carers and families to share their views and ideas on how to improve healthcare in Australia.

What can the GSNV and you do?

• The GNSV supports equitable access to medications for all individuals regardless of income and other factors, and therefore supports campaigns such as this Hip Pocket Pain and Medicines Savings campaigns.

It does so by providing position statements, networking with other consumer advocate groups, and supporting individuals and genetics support groups in their own advocacy endeavours.

• Feedback to us at the GSNV your experiences and views which we can use to inform and support our advocacy for you.

• Spread awareness by talking to your friends, family, neighbours and local member of parliament.

• Have your say on the CHF’s ‘Our Health, Our Community’ forum at http://ourhealth.org.au/

Successes, but more can be done

In addition to important programs such as the Orphan Drugs Program and Life Saving Drugs Program, the Commonwealth government has listened to consumers and recently announced large price cuts and new additions to the PBS.

The CHF hailed this move but also highlighted that Australian consumers still pay up to 16 times more for medicines than in NZ, the UK, and Ontario, and the potential for an even more substantial drop in medicines costs. The overall message is that the Australian health system needs to be overhauled, including benchmarking medicines prices against other countries.

Another huge success is the increasing recognition that consumer input is critical in health system reform. The Commonwealth and Victorian governments are moving beyond allowing token sideline input, to involving consumers as major stakeholders in policy and wider social discussions and decision making about what consumers value in their health system.

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Get inspired by the Empower: Access to Medicine campaign, a UK endeavour driven by personal experience of a rare condition, at www.accessstomedicine.co.uk.

Sources:
2. CHF media release 29 April 2013
3. Brisbane Times 29 April 2013
4. GSNV Autumn 2013 newsletter article ‘GSNV’s Rare Disease Day Seminar’
7. Grattan Institute report ‘Poor Pricing Progress: Price disclosure isn’t the answer to high drug prices’ 1 December 2013
9. Chronic Illness Alliance policy statement August 2013
10. Pharmadispatch website article 29 April 2013 www.pharmadispatch.com/families-bewildered-by-myozyme-rejection/?pgnc=1
12. Personal communication, Mr Raymond Saich, President, Australian Pompe’s Association 15 January 2014
13. CHF media release 1 December 2013
14. CHF Health Voices Issue 9 October 2011 p16
Three-parent IVF: Why the controversy?

A new reproductive technique, ‘three-parent IVF’, has recently appeared in the media amidst a great flurry of controversy. This technology has the potential to help women who are affected with severe mitochondrial diseases to conceive healthy children. The implementation of this is currently being considered by regulatory authorities in the US and UK.

By Venessa Miller BA, BSc (honours) and Nancy Amin, B.A./B.Sc. (Hons), Ph.D

What are mitochondria?
Mitochondria are the ‘powerhouses’ in every cell of our body that provide us with energy. Although most of our DNA is contained in the nucleus, our mitochondria have some (about 0.1% of total cellular DNA) of their own DNA ("mtDNA").

Normally, a child inherits their nuclear DNA from both their biological parents; half from the mother and half from the father. In contrast, all of a child’s mtDNA is inherited from their mother. Each cell in our body contains around 1000 copies of mtDNA. If there is a genetic fault in the mtDNA, it may not affect all of the copies. It takes a certain proportion (or threshold) of faulty mtDNA for disease to occur in an individual.

There are hundreds of types of mitochondrial disease, which vary in severity. In many cases, individuals who have a mitochondrial disease live with debilitating illness, with symptoms such as seizures, dementia, migraines, heart failure, diabetes, liver disease, muscular dystrophy and reduced life expectancy.

How does three-parent IVF help parents to avoid passing on mitochondrial diseases?
Current options to assist women with mitochondrial diseases have an unaffected child include:

• Adoption
• Egg or embryo donation with IVF
• Conceive naturally and have prenatal diagnosis
• Preimplantation genetic diagnosis (PGD) with IVF and the selective transfer of embryos that have the lowest chance of being affected

These options have limitations. Due to the nature of mitochondrial inheritance, prenatal diagnosis and PGD for mitochondrial diseases can never give a definitive negative result. Moreover, for some people prenatal diagnosis or PGD are not realistic options because it means making choices about termination of pregnancies or discarding embryos. The financial cost of IVF may also be prohibitive for many families.
Mitochondrial replacement-assisted IVF involves the collection of eggs from both an unaffected egg donor and from a mother who has a genetic fault in her mtDNA.

The mitochondria from the mother’s egg are then ‘swapped-out’ with the unaffected mitochondria from the donor egg. The resulting egg will have healthy donor mitochondria and the mother’s nuclear DNA. This egg could then be implanted and fertilised using IVF techniques.

These techniques therefore prevent any disease caused by faults in mtDNA from being passed on to the next generation.

**Conclusions**

Currently there are many uncertainties associated with mitochondrial replacement technologies. Further studies need to be performed to examine feasibility and safety. Public discussion of potential ethical concerns is also an important part of the approval processes. However, some of the fears and concerns raised can be tempered with further informed debate and the clarification of any misconceptions.


**ABC Broadcast**

David Thorburn, Head of the Mitochondrial Research Group at the Murdoch Childrens Research Institute, has discussed this issue on the ABC, and you can access the broadcast here:

John Stewart, 2013, ‘Three in one equals a healthy baby’ broadcast, ABC, 28 June

[www.abc.net.au/lateline/content/2013/s3792350.htm](http://www.abc.net.au/lateline/content/2013/s3792350.htm)

**Why has this caused so much controversy?**

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<td>Requires a third ‘parent’ (a woman who donates the unaffected mitochondria).</td>
<td>This technology is currently only experimental; it is currently not known if a mixture of mtDNA from two different origins is safe in humans, or if any unexpected problems may manifest in future generations.</td>
<td>Being born from genetic material from more than two parents has lead to concerns about issues of personal identity of the child.</td>
<td>A major area of concern is that this technology could lead humankind back into another era of eugenics.</td>
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<td>An intervention that modifies the human germline.</td>
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**On the other side of the debate:**

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<td>Current IVF techniques require a third ‘parent’ that donates the egg, sperm or embryo.</td>
<td>Future studies are required to address these concerns.</td>
<td>Advocates argue that identity is not in the genes but in the world in which we live in and our relationships with other people.</td>
<td>Just because a technology is available won’t necessarily mean it will be misused. Continued ethical debate and regulation can avoid misuse.</td>
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<td>This technique does not actually ‘modify’ DNA at the molecular level. Rather, it is a swapping-out of genetic material. In this sense it may not be ethically different to egg, sperm or embryo donation.</td>
<td></td>
<td>Family structures are currently varied: donor, adopted, surrogate, divorced/separated families, step-families, foster families.</td>
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**Mitochondrial replacement-assisted IVF**

- Involves the collection of eggs from both an unaffected egg donor and from a mother who has a genetic fault in her mtDNA.
- The mitochondria from the mother’s egg are then ‘swapped-out’ with the unaffected mitochondria from the donor egg.
- The resulting egg will have healthy donor mitochondria and the mother’s nuclear DNA.
- This egg could then be implanted and fertilised using IVF techniques.
- These techniques therefore prevent any disease caused by faults in mtDNA from being passed on to the next generation.
CMTAA funds major research

The Charcot-Marie-Tooth Association of Australia (CMTAA) has donated $75,000 to the University of Sydney’s Faculty of Health Sciences to fund research on Charcot-Marie-Tooth disease (CMT).

The research will establish the cost of CMT to the community and recommend strategies to reduce the health burden of this little known genetic disorder.

*“Some of the possibilities we are looking at to reduce the financial burden include the establishment of a ‘CMT Speciality Centre’, improvements in the way the education sector accommodates those with CMT, and Pre-Implantation Genetic Diagnosis (PGD) supported by Genetic Counselling,” says Associate Professor Burns, Co-director of the research group at the University of Sydney.*

Reproduced with permission from Heather Jacobs, Division of Alumni and Development, University of Sydney and CMTAA.

Inequalities in early childhood outcomes for Australian children

Cont. from page 1

Such children are represented across the population, but rates tend more to be higher in boys, Aboriginal children and older children and those from lower socioeconomic status communities.4

Our research also shows that approximately one in four children with special health care needs are developmentally vulnerable across the AEDI domains, compared to approximately one in 20 children without special health care needs.

How can communities respond to these data?

In communities where AEDI results are poor we know that doing nothing is not an option, but how do community sector organisations turn AEDI data into action?

Typically, the process of service redevelopment includes forming a local network of agencies that can make a difference to children’s outcomes. Vital to this process is to involve the relevant families and children. This act, in itself, aims to redress inequities and is likely to put the child and their family ‘front of mind’ in the planning process.

References


The Children’s Tumour Foundation

The Children’s Tumour Foundation of Australia is a national group supporting those with neurofibromatosis (NF), their families, carers and friends, as well as promoting awareness of NF and raising funding for research.

The Foundation has generated around $300,000 in fundraising in the 2013 financial year.

Professor Kathryn North, medical advisor to the Children’s Tumour Foundation of Australia and director of the Murdoch Childrens Research Institute, has been instrumental in forming a leading clinic in Sydney for individuals with NF and hopes to initiate a similar clinic in Melbourne.

Sources:

• 2013 Annual Report, NF Australia Ltd
• Guest presentation, Prof Kathryn North, Children’s Tumour Foundation of Australia Gala Dinner, Melbourne, Oct 25 2013
Disorders of Sex Development (DSD) and other variations of sexual development occur when a person’s development doesn’t follow the typical male or female pathway.

Variations of sexual development are estimated to affect 1 in 4500 live births in cases of genital ambiguity, while misplacement of the urethral opening affects as many as 1 in 250 boys.

A team of researchers from The University of Queensland (UQ), Prince Henry’s Institute and the Murdoch Childrens Research Institute has established a website to overcome misconceptions about sexual development by improving awareness and providing up-to-date scientific information.

"The resource we have developed provides accurate information to help people understand the molecular basis of typical sex development and how changes to this pathway can result in various types of DSDs," Professor Sinclair said.

Professor Vincent Harley at Prince Henry’s Institute in Melbourne said the website offered a much-needed scientific perspective to complement the valuable patient support and advocacy offered by community organisations.

"We hope that improving understanding and awareness will assist the wider community to better understand DSDs and the needs of those affected," Professor Harley said.

“The DSD web resource is a welcome introduction to further discuss and process the challenging issues of disorders of sexual development, both for individuals directly affected, their families and the wider community.

It is straightforward and informative approach assists in providing a language to think and talk about these issues.

The intent is very clear as an adjunct to medical advice and to social support groups but it speaks of the genetic and biological foundation of these conditions that has previously been less clear.

As a mother of a young person with a genetic condition and as a medical professional, I highly value resources and connections which acknowledge the issues that impact on the quality of life for the whole person and contribute to community understanding.”

Marie Karamesinis
Parent and medical professional

The website can be accessed at www.dsdgenetics.org

This website has been developed as part of the Research Program in Disorders of Sex Development funded by the National Health & Medical Research Council.

Exploring the psychosocial support services for women with disorders of sex development

by Chloe Hanna, Master of Genetic Counselling student

Chloe Hanna, Sonia Grover, Louisa Di Pietro (GSNV), Melody Menzies

Women with differences of sex development (DSD) usually have significant reproductive health needs presenting with complex genetic changes and gynaecological features. These differences can have an impact on them physically and affect their fertility, body image, and self-esteem. This qualitative study was designed to explore the psychosocial support needs of women who have a DSD. Ten interviews were conducted with women aged between 18 to 40 years with a range of DSD diagnoses.

Participants were happy with the psychosocial supports they had received chiefly from their families. Nonetheless they indicated that the development of more sophisticated and comprehensive psychosocial support resources were needed. Participants identified areas in life that could be improved with psychosocial support, such as: social acceptance, forming intimate relationships, coping with the physical implications, understanding reproductive options and maintaining better health outcomes. The majority of participants highlighted the value of appropriate peer relationships and wanted more of these connections.

The general consensus among participants was that there were substantial gaps in psychosocial support services available for women with DSD. Participants suggested that the development of resources such as databases of specialised health professionals, support groups and access to up-to-date information about their condition would fundamentally improve their psychosocial support systems. This study may form the basis for further studies.
Your Say... Your Thoughts

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

Working towards equal opportunities for individuals impacted by cystic fibrosis

At Cystic Fibrosis Victoria we are all too aware of the equity issues faced by our community. Members Louisa Walsh and Monica Lay provide their perspective on equity in the cystic fibrosis community.

What is cystic fibrosis (CF)?

Cystic Fibrosis is a genetic condition, which mainly affects the lungs and digestive system. CF causes the build up of thick mucus in the lungs, which traps bacteria, causing infection which can lead to permanent lung damage over time. CF also decreases the ability of the body to digest fat and absorb nutrients.

People with CF have a chronic cough, can be short of breath, and often struggle to gain weight. People with CF need to take a lot of medication, and have to perform physiotherapy treatments and do exercise every day to keep their lungs clear and maintain their health. Many people with CF need to have regular hospital admissions to manage persistent chest infections.

CF is a recessive genetic condition. It is the most common life-shortening genetic condition in Caucasian people, with approximately 1 in 2500 Caucasian Australians being born with CF, and 1 in 25 people being carriers of a genetic mutation which causes CF.

Carrier testing for CF has been available for more than 15 years in Australia – despite this, the vast majority of parents of a child with CF have no idea of their carrier status before their child is born.

Carrier Testing and Equity

Equity issues can impact people who are seeking carrier testing – and these issues can begin before testing and continue throughout the whole journey of testing, finding out you are a CF carrier, and making decisions around having children.

Not all individuals have equal access to appropriate information. While some GPs, obstetricians and midwives will provide comprehensive information about the tests that are available, other clinicians don’t always provide comprehensive and up-to-date information. Similarly, some individuals will have better experience, knowledge and skills to help them find their own information to support decision making around their own reproductive choices.

Carrier testing isn’t cheap, and the subsidy for testing family members of people with CF is no longer in place. This means that individuals and couples wanting to be tested can spend up to $300 for their tests - even those people who are most likely to be carriers. This cost can be a substantial barrier for some people who would otherwise access testing. Should tests come back positive, people who are carriers then face reproductive decisions which can come with considerable financial (and emotional) burdens – such as accessing IVF treatment with pre-implantation genetic diagnosis (PGD) or antenatal testing.

Finally, people who are CF carriers can find themselves facing unequal support of their reproductive choices. Some people known to be carriers of genetic conditions who are deciding to start a family have described feeling an enormous amount of pressure to use reproductive technology to avoid passing on the condition. Some people who make the decision to not test, or to continue with pregnancies despite knowing that their child will have a genetic condition, have felt pressure to change their mind and discrimination for their decision. This situation is not unique to CF, and demonstrates inconsistencies in supporting and respecting the values and choices of individuals and couples.

Cystic Fibrosis Victoria is starting a carrier screening project next year, with the aim of raising awareness of carrier testing for CF, supporting informed reproductive decision making for people thinking about starting a family, and addressing some of the equity issues associated with carrier testing. If you are interested in finding out more about the carrier screening project, please contact the Cystic Fibrosis Victoria office on 03 9686 1811.

Equal inclusion in the community

Having CF can impact on an individual’s life in many different ways. Children and teenagers with CF can struggle to access full education, especially if they are frequently hospitalised or unwell and miss a lot of classes. This can mean either falling behind or losing interest in school, which can have flow-on effects to accessing higher education and work.

CF can have a financial impact, both directly and indirectly, on families and individuals. Having CF is not cheap – even with Health Care Cards and other government support, the costs of medication and equipment can run into thousands of dollars each year. Adults with CF, or carers of people with CF,
may also struggle to work, especially full time, or may have to take periods of leave without pay for hospital admissions – further increasing the financial burden.

Having CF can also affect opportunities for access to recreation. Sporting clubs can sometimes be reluctant to accept people with CF, due to worries about their care needs should something ‘go wrong’. Schools have sometimes disallowed students with CF to participate in camps because of concerns about the management needs of CF. Even holidays can be difficult, as people with CF can find it very difficult to access travel insurance which provides them adequate health cover.

Having any chronic illness or genetic condition can have significant impacts on equity. Cystic Fibrosis Victoria works hard to advocate for people and families with CF to make sure they don’t miss out on opportunities that most people take for granted. If you have any questions about the work of CFV, or the programs and services we offer for people with CF, contact Monica Lay on 03 9686 1811.

We work on disorders of energy generation affecting the mitochondrial powerplants in our cells that convert food into energy. Severe mitochondrial problems cause impaired physical or cognitive development, neurodegenerative disease and other disabilities, with any age of onset.

Mitochondrial disorders are the most common group of inherited metabolic diseases, affecting at least 50 children born in Australia each year. Mitochondria require more than 1000 proteins for normal function and mutations in over 150 genes are known to cause mitochondrial disorders. A subset of 37 mitochondrial DNA genes are located in the mitochondria themselves and are inherited only from the mother.

For over two decades we have acted as the Australasian referral centre for children suspected of mitochondrial disorders and we have diagnosed over 500 affected children. Traditionally, diagnosis has required an invasive muscle or liver biopsy in which we measure the function of the mitochondrial enzyme complexes in order to guide further analyses of specific genes.

Much of our recent research has focused on using Next Generation DNA sequencing technologies to sequence over 1000 genes at once encoding all the known mitochondrial proteins. This has allowed us to identify 10 new disease genes in the last 4 years and markedly improve genetic diagnosis.

Key publication
Calvo et al., 2012 Molecular Diagnosis of Infantile Mitochondrial Disease with Targeted Next-Generation Sequencing. Science Translational Medicine 4, 118ra10 (featured as a Genome Advance of the Month by the American National Institutes of Health (www.genome.gov/27547295).
WHAT'S ON...

Van Wright Foundation Gala Dinner
Friday 21 February

MCRI Healthy Kids Seminar
Monday 24 February

Rare Disease Day 2014
Friday 28 February

Show of Hands Ball
Friday 28 February

MCRI Discovery Day
Sunday 2 March

Million Steps for SMA
Sunday 23 March

Vascular Birthmarks and Malformations Family Info Day
Saturday 3 May

SEEKING CONTACT

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

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

Disclaimer

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

ACKNOWLEDGEMENTS

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CALENDAR OF EVENTS

The Vascular Anomalies Clinic (Royal Children’s Hospital) and Genetic Support Network of Victoria partnership invites you to attend our

VASCULAR BIRTHMARKS AND MALFORMATIONS FAMILY INFORMATION DAY

Date: Sunday 3 May 2014, 1pm – 4pm
Venue: Ella Latham Auditorium, Ground Floor, Royal Children’s Hospital, 50 Flemington Road, Parkville 3052, VIC

For full details see our website www.gsnv.org.au/events.aspx

IN BRIEF

Access to parking at Melbourne’s public hospitals

The Chronic Illness Alliance (CIA) has commissioned a survey to engage the perspectives of individuals with chronic illnesses and their carers on the cost of parking at public hospitals. The report is available on the CIA website, www.chronicillness.org.au.

The report highlights that most of Melbourne’s public hospitals are in inner city suburbs such as Parkville, Prahran, East Melbourne and Fitzroy. Of the 322 survey respondents, 49% estimated they spent more than $150 per year on hospital parking. Participants adopted multiple strategies to meet the cost of parking such as using alternative transport, reducing spending on household items in order to afford the parking, changing doctors or hospitals, and some even reported missing appointments or going without medicines.

The CIA concluded that the cost of parking at public hospitals in Melbourne contributes to financial distress experienced by individuals with serious long-term illnesses.

“That people need to miss appointments or save in other areas such as medicines suggests that any further increases may compromise their health and contribute to hospital costs through emergency department visits and unplanned admissions.”


See this issue profiled in the Herald Sun: Mickelburgh, P., 2014, “Hospitals rake in $60m for parking”, Herald Sun, 13 January