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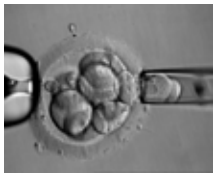
WINTER 2012

Pre-implantation Genetic Diagnosis

Pre-implantation Genetic Diagnosis, or PGD, has received extensive media attention in recent weeks for its use in later onset conditions including hereditary breast and ovarian cancer.

By Lynley Donoghue



While some reports have described the technology as a step towards "Designer Babies", the reality is far from this.

PGD uses IVF technology to test embryos prior to implantation for genetic conditions. The technology has been available for over 20 years, and has been utilised for a variety of conditions including Huntington's disease and Cystic Fibrosis.

In the case of breast and ovarian cancer caused by changes to the BRCA1 and BRCA2 genes, there is a fifty per cent chance of having a baby with the same changed gene. Changes in these genes give an individual between a 60 – 80% chance of developing breast cancer, and a 5 – 20% chance of developing ovarian cancer over their lifetime.

For some people, that risk for their child is too great, and PGD gives them the option to avoid this. The benefit of the technology is that only embryos without a specific genetic trait are implanted, which can prevent couples from going down the pathway of testing during a pregnancy at around 12 weeks, and then having to make the difficult decision whether to continue or not.

PGD is not an easy road to take however. Not every condition can be tested for, and setting up for the test can take as long as six months. The test then costs several thousand dollars on top of the usual IVF procedure. In addition, it can often take several cycles to produce a successful pregnancy, which can leave couples $20-30,000 out of pocket, or sometimes more.

While PGD can be an option for families with a genetic condition, it comes with its' own associated issues, which don't just include the financial costs but the emotional and physical stresses of an otherwise fertile couple undergoing IVF treatment.

For some, the stresses are worth it, as the technology allows them to have a child free from a condition that has negatively affected their own life. This is a far cry from the issue of designer babies in which a child is selected based on "desirable" traits. This is about survival and quality of life.

Dr David Amor, Clinical Geneticist and Director of the Victorian Clinical Genetics Service says "it is unfortunate that many couples who would otherwise choose PGD are denied access for financial reasons" and disappointingly successive governments have failed to deliver funding support for the technology.

"Whilst the cost of PGD for single gene disorders is relatively high, it is a much needed service that will only be utilised by a relatively small number of families. Moreover, the cost of PGD is minimal in comparison to the cost of treating a child with a serious genetic condition, a point that is frequently made to me by couples who are prevented from using PGD by the financial cost." ■

Contents

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Committee of Management 2012

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

General Committee Members
Christine Williams
Yvonne Waite
Maria Triantafillou
Amy Herlihy

Committee Meeting Dates 2012

Teleconferencing will be available at all meetings.

Thursday August 16
Thursday September 20
Thursday October 18 AGM Meeting
Thursday November 15
December (to be confirmed)

The GSNV is looking for Committee Members, email info@gsnv.org.au or phone (03) 8341 6315 for more information.

Message from the President

Since our last newsletter, the GSNV has undergone significant changes in relation to staffing. The Murdoch Childrens Research Institute has assumed the line-management of the GSNV staff since mid March; and under a spill and fill, restructured the team.

Louisa Di Pietro was appointed as the 'Group Leader' and heads a new team whose responsibility is to implement the plans of GSNV Inc., under line management of David Amor, Director, Victorian Clinical Genetics Services ("VCGS").

As before, GSNV Inc. through its management committee and office holders, remains responsible for the organisation's mission, strategic direction and compliance with all the legal obligations of an independently incorporated association.

GSNV Inc. and Professor David Amor, as Director of VCGS are developing a long-sought memorandum of understanding about how the two entities will work together to achieve GSNV Inc's purposes. This means developing some new management and improved communication processes.

The message from our Group Leader on page 3 explains some changes in the office but I take this opportunity to also cover some of the important issues discussed by the GSNV Committee at this time.

The recent Associations Incorporation Reform Act came into effect on 1st July this year. It changes many of the assumptions under which incorporated groups have operated in Victoria, and has encouraged GSNV Inc.'s management Committee to reconsider the constitution and articles of association.

This is both to comply with the Act and reflect the changed relationship it now has with the Murdoch Childrens Research Institute and the VCGS, while meeting our current and some anticipated future needs.

A review of the GSNV constitution will focus on the capacity of the organisation to assume its full capacity as a community-based organisation to raise funds, consult with its members and report independently on the provision of genetic services to people who are affected by genetic conditions.

The Committee's role and responsibilities, particularly in facilitating consumer participation and feedback on services, are important at this time.

In moving forward, the executive and management committee of the GSNV places on record the importance of people with genetic conditions that affect their lives participating in the design and evaluation of the many services that they need.

GSNV Inc. has always tried to be both an umbrella organisation of self-help groups and advocacy organisations as well as a support to individual members directly or indirectly affected by genetic conditions, working in genetic health or who have a particular interest in genetic health.

Over the coming months, the GSNV will be inviting individuals and families to participate in forums and planning activities designed to provide direct consumer feedback on genetic services and how the GSNV's services, delivered through VCGS and with its support, could best inform genetic health practice.

We need to grow the management committee, which can have up to 16 members, the majority of whom must be people who are affected by genetic conditions.

We can co-opt members pending the AGM later this year, and we hope that a lot of you want to be involved.

Help support your group to have a voice or share the experience of having a specific genetic condition: think about getting involved with the GSNV Committee.

We are continually looking to extend our Committee membership and really, truly want group leaders and people affected by genetic conditions to consider joining the GSNV Committee and have your voice go further.

The GSNV's Annual General Meeting, held around October of every year, is when official Committee nominations are accepted.

From now until October of this year, I invite any interested parties to consider having a trial experience, by being willing to be co-opted onto the GSNV Committee and formally seek nomination at the AGM if you enjoyed your experience – or thought you could make it better.

If you are interested in joining our Committee, please do not hesitate me (0407 613 777) or Louisa Di Pietro (03 8341 6315) to talk about it.

Moira Rayner
President

Message from the new team

Welcome to the winter edition of the GSNV Newsletter. Some of you may be aware that the GSNV has undergone some changes, including staff.

As the Group Leader, I will be managing Lynley Donoghue (Genetic Support Coordinator) and Christine Chan (Administrative Assistant) and working with the GSNV Committee. With a fantastic mix of skills and experience between us, we have genetic support development and administration covered.

GSNV extends goodbyes and sincere thankyous to GSNV staff Stephanie Shepley, Penny Dodds and Tarli Bogtstra for their tireless contribution and work with the GSNV. We wish Stephanie and Penny all the very best in their careers and futures and wish Tarli all the very best with her new born baby and family life.

We also thank Alice Weeks who is retiring as joint secretary to pursue travel to Europe. Thank you to all for your excellent work.

Lynley, Christine and I are available to all support groups, members and our networks at any time to answer questions and/or assist with any support group needs.

Our website is a great place to start if you are looking for something from the GSNV and we also encourage you to phone the GSNV office on (03) 83416315 or send us an email to **info@gsnv.org.au**.

In the coming months, the GSNV team and committee will continue to work towards efficient processes and effective governance but above all, seek to improve on how we deliver support to individuals, families and support groups.

Once again, we encourage you to send us any important information you wish

Christine Chan, Louisa Di Pietro and Lynley Donoghue.

us to pass on your behalf and to use the GSNV as a portal to link support groups, advocacy and the community.

We're continuing to update our website and support group links and have been contacting groups individually in order to update and confirm contact information.

If you have been contacted by our office in recent weeks and have not spoken to one of our staff or volunteers, please do contact us if your support group or contact information has changed.

We are also using this opportunity to remind support groups and our members that the GSNV is happy to list important events and promote activities via our newsletter, website and e-bulletin.

We look forward to working with all our support groups and families in the coming months.

Louisa Di Pietro
Group Leader

SERVICES

VCGS Leaders in Genetic Health

NEW LOOK!

Victorian Clinical Genetic Services (VCGS)

The VCGS has recently consolidated its branding in order to simplify brand identification and represent its services.

A new look logo and website have been launched and refreshed the VCGS image.

The homepage of the new VCGS website separates navigation into two main parts, Clinical Services and Pathology Services.

For a pathway to the GSNV on this site please visit **http://www.vcgs.com.au** and click on the following tabs:

- Clinical Services
- Patients and Families
- Counselling and Support
- Information on the GSNV is found under Support Groups

Electronic health records legislation passes parliament

Patients will no longer have to repeatedly re-tell their medical histories to doctors after legislation passed parliament to set up an electronic health record system.

The federal government says the system will bring the management of health records into the 21st century and provide life saving information in emergencies.

The legislation passed the Senate on Tuesday evening with the support of the coalition despite the concerns about privacy from some opposition senators. The system aims to reduce the number of hospital admissions because of medication errors which equate to 190,000 a year as well as cutting down on medical errors due to inadequate patient information.

Australians' health records will be available online and protected by encrypted passwords. The electronic health system will be rolled out over time beginning on July 1, 2012.

Liberal senator Concetta Fierravanti-Wells raised concerns about the roll out of the system and sought a launch date from the government, particularly if a planned launch in Sydney on July 2 was still proceeding.

Labor senator Jan McLucas said she had no information about the date but said the system was undergoing a final round of vigorous testing.

"We know this is a complex area of national reform," she said. "I think Australians ... will be far more interested in getting a quality personally controlled electronic health record rather than when there will be a launch."

People can register for the system by phone or by going into Medicare.

Australian Greens senator Richard Di Natale said people would be shocked how "technologically primitive our health system is."

The Personally Controlled Electronic Health Records Bill 2011 and the related Personally Controlled Electronic Health Records (Consequential Amendments) Bill 2011 passed the Senate with amendments. The bills now go to the Governor-General for royal assent.

Source: AAP 20/06/2012

The Australian Childhood Vision Impairment Register

The Australian Childhood Vision Impairment Register is an initiative designed to capture uniquely Australian data on the number of children formally diagnosed with vision impairment.

The register captures data from every state and informs on the needs, and experiences of vision impaired children in order to plan and develop better treatments and services. The register is yet another example of how specific registries can not only provide accurate data and figures on the prevalence of conditions, but also support funding submissions, service planning, therapeutic and cure intent research and support individual and family needs.

For more information please go to: **http://www.ridbc.org.au/renwick/research/projects/acvir/**

Auslan in school cirriculum

Peter Garrett, Minister for School Education recently announced that Auslan, the first language of many Deaf Australians, is to be included as a subject in the national curriculum.

The Australian Curriculum Assessment and Reporting Authority are to start work soon on developing the content for the subject. The inclusion of Auslan in the curriculum will not only benefit Deaf students but will provide the opportunity for all students to study Auslan if they choose to do so.

More information can be obtained by contacting the Department of Education, Employment and Workplace Relations (DEEWR) at **www.deewr.org.au** or by phone on 1300 363 079.

Source: Listen Up! Magazine, Children with Disability Australia, April 2012.

SERVICES

Planning a trip?
Travel Insurance

Travel insurance is highly recommended for anyone travelling overseas.

By Rebecca Freedman, GSNV Volunteer

Travel insurance falls into two categories;

a) non-medical cover, covering unforeseen events such as cancellation of trips or flights, theft or damage of luggage, and

b) medical and disability cover. This covers overseas hospital and medical expenses and loss of income due to injury or illness.

Individuals with pre-existing medical conditions may have had the experience of being denied medical and disability cover when purchasing travel insurance.

This can be a frustrating process and whilst some companies have a total exclusion policy for anyone with a known pre-existing condition, this is not the case with every company. Some insurers will provide coverage to people with pre-existing conditions.

Policies will differ between and possibly within companies depending on the insurance company, the ages of those being insured, destination of travel and nature of the medical condition.

It is important to do your research when considering purchasing travel insurance to find a policy that best suits your individual and family's requirements.

In general, keep the follow tips in mind when purchasing travel insurance

- Read the Product Disclosure Statement carefully, ensure that you understand exactly what the policy covers, as travel insurance is not unlimited and exclusions apply (even for those without pre-existing conditions)
- Be aware of any excesses, how and where these apply
- Ask the insurer to explain and clarify any points that cause confusion
- Ensure the cover is adequate for potential medical expenses that might occur whilst in the country you are visiting
- Declare any pre-existing medical conditions you may have; this is very important and non-disclosure can result in non payment
- You are able to appeal if you are denied cover
- Consult an insurance broker, they may be able to assist you further
- Even with a pre-existing medical condition you should be able to purchase non-medical cover
- Provide a letter from your doctor which can outline how you are an acceptable risk, and the details of your condition

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 provided by other agencies or organisations whether it covers your condition or ask if they know of any companies that do.

The following list of website provides some useful information on purchasing travel insurance and insurance in general. Information in this article has been collected from these websites.

Chronic illness alliance: "work welfare wills"
http://www.chronicillness.org.au/workwelfarewills/travel_index.htm
Step by step information on how to apply for medical and disability travel insurance and what to expect from the process

Australian Securities and investment commission
https://www.moneysmart.gov.au/managing-my-money/insurance/travel-insurance#exclusions

Insurance council of Australia
http://www.insurancecouncil.com.au/for-consumers/types-of-insurance/travel-insurance

Smart traveller
http://www.smartraveller.gov.au/tips/insurance.html
Government website with information on safe travelling

Medicare
http://www.humanservices.gov.au/customer/services/medicare/reciprocal-health-care-agreements
contains information about Australia's reciprocal health agreements for treatment and services received in New Zealand, the UK and some European countries

Genetic discrimination and insurance
http://www.respondgenetic discrimination.com/discrimination-in-insurance.html

RESEARCH

Genetic Technology – Microarray

As humans we are made up of billions of cells. Each of these cells contains our genetic material, or DNA. The complete set of all of that material is called our genome. A microarray is a genetic test that looks for extra or missing segments of DNA in a person's genome.

By Lynley Donoghue

What is a microarray?

A microarray itself is a small chip, about the size of a thumbnail, onto which thousands of different gene sequences are attached in an orderly way. This allows for thousands of genetic tests to be performed all at the same time.

How does it work?

Our genome is organised into packages of genes called chromosomes. There are 23 pairs of chromosomes in each cell. Because our chromosomes come in pairs, we should have two copies of every gene in each cell. A microarray determines how many copies of each gene are present in our DNA.

Having more than two, or less than two, copies of each of our genes can cause problems in the functioning and development of an individual. This is called copy number variation, and this is what a microarray tests for.

Why are microarrays so popular?

Microarrays are a very cost-effective and relatively fast way of looking at almost the whole genome at the same time. Up until a few years ago chromosome analysis, or looking at chromosomes under a microscope, was the only way in which the whole genome could be analysed at relatively low cost.

However, chromosome analysis can only detect very large changes in the genome. Also, a health professional had to have a pretty good idea of what was causing a person's condition so that any other appropriate tests could be done.

With the advent of microarrays this is no longer strictly the case, as the test looks at thousands of different genomic regions at the same time and therefore has a higher chance of providing answers. In this sense, microarray technology has changed clinical testing.

Who can order a microarray test?

Many health professionals can order a microarray test, including geneticists, paediatricians and GPs. Microarrays are increasingly being utilised in paediatric investigations into developmental delay and other health concerns when a syndrome diagnosis is unclear.

They can also be used in the prenatal setting. This is often the case if an ultrasound shows that there may be something wrong with a baby, a microarray test can be used to try and find out what is causing it.

Why is a microarray test ordered?

A microarray test is commonly ordered if it is thought that extra or missing pieces of the genome are the cause of health or developmental problems, particularly if it is not known what is causing these problems. A microarray test can also be used to confirm a suspected diagnosis.

Does a microarray test for everything?

A microarray cannot test for everything. It only tests for changes in the number of copies of pieces of DNA. There are many other conditions in which there is a change, or a mutation, in a gene, but it does not change the number of copies of that gene. Copy number changes involving segments of DNA must also be of a minimum size before they can be detected by the microarray.

What results can you expect from a microarray?

You can expect two possible results from a microarray test:

1. No copy number variation (changes in the number of genes) is found. This is the most common result, and it means that the condition in question remains unexplained by this test. It does not mean that the cause of the condition is not genetic.

OR

2. A copy number variation is found.

What does it mean when a copy number variation is found?

This does not necessarily mean that it is the cause of the condition. There is a lot of variation in our genome that is responsible for making us all unique, and many of these differences do not cause a problem. After a copy number variation is found, it may have to be investigated further to see whether it is likely to be the cause of the condition.

There are several possible outcomes:

1. The variation is known to cause the condition. When this occurs, this variation has often been reported in other individuals with similar developmental and health concerns.

2. The variant is of *uncertain significance.* This means it is not fully understood how the variation has an impact on health and development. Although the variation may occur more frequently in individuals with health or developmental concerns it is also be found in individuals without these concerns.

RESEARCH

Sometimes a parent will carry the same variation as their child, and they may have a similar or milder health problem, or they may have no obvious health problems at all. It is not yet understood why this variability occurs.

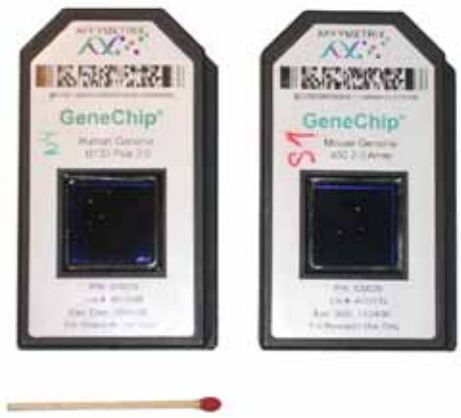
Other, unknown factors may play a role. When a copy number change of uncertain significance is found in a child it is usually necessary to study both parents to help determine the inheritance.

3. The variant is of *unknown significance.* When this occurs, further investigations are needed in order to try and clarify the result. Variants of unknown significance have generally not been reported previously, or may have been reported in only one or two individuals. Both parents of the child will need to be tested to see if either is a carrier of this variation.

    - If one parent has the variant and has similar features as the child, then the variant is likely to be responsible.
    - If one parent has the variant and doesn't have similar features as the child, then the variant is unlikely to be responsible.

Sometimes neither parent will have the variant carried by their child. This is called a *de novo*, or a *new* change and has probably occurred in the egg or sperm prior to conception.

A *de novo* finding increases the probability that the variation is the cause of the condition in the child, but this variation may still have *uncertain or unknown significance*.



4. The variation found involves genetic material that is unrelated to the condition being investigated, but is potentially associated with other health concerns. This finding is uncommon, but results in information being gained that was not expected. Known as an *incidental finding*, this can occur in the individual being tested or during testing of their parents. An incidental finding may require further follow up and counselling if it is likely to be significant for the individual and family's future health.

What are some of the issues associated with microarray?

As mentioned earlier, a microarray looks at almost the whole genome, which means a lot of data is generated from the one test. Most of this information is useful, but it can sometimes be unexpected, and sometimes unhelpful.

Data can be difficult to interpret, and can lead to findings that are of uncertain or unknown significance. This may be difficult for some families to deal with. Incidental findings pose another potential problem, particularly for late-onset conditions, where a person may not want to have that information.

Another issue is that microarrays can pick up very rare changes in a person's genome that cause a particular condition. Often, especially in children, a parent may want to find a family of an older child with that condition to get an idea of what to expect, however with these very rare changes in chromosomes this is not always possible.

For information about a particular unique chromosome condition, visit support organisation Unique **www.rarechromo.org**

References

Testing for missing or extra segments of DNA Fact Sheet, The Centre for Genetics Education, NSW Health.
www.genetics.edu.au

Acknowledgements

Thank you to Mark Pertile, Senior Medical Scientist and Dr Sue White, Clinical Geneticist for their help with this article.

In the Media

Noninvasive genetic test for Down syndrome and Edwards syndrome highly accurate

June 5, 2012 in Obstetrics & gynaecology
Current screening strategies for Down syndrome, caused by fetal trisomy 21 (T21), and Edwards syndrome, caused by fetal trisomy 18 (T18), have false positive rates of 2 to 3%, and false negative rates of 5% or higher. Positive screening results must be confirmed by amniocentesis or chorionic villus sampling, which carry a fetal loss rate of approximately 1 in 300 procedures.

Now an international, multicenter cohort study finds that a genetic test to screen for trisomy 21 or 18 from a maternal blood sample is almost 100% accurate. The results of the study are published online in the *American Journal of Obstetrics and Gynecology.*

For the complete story see **http://medicalxpress.com/news/2012-06-noninvasive-genetic-syndrome-edwards-highly.html**

Research Study

'Disability & Ability: How Young People with Impairments make the Transition to Adulthood'

A University of Sydney and Deakin University project, funded by the Australia Research Council are seeking to recruit young people aged 19-26yrs with impairments to be interviewed yearly for 3 years. Interviews will take a life history approach and explore a range of topics. The project has ethics approval from the University of Sydney Human Research Ethics Committee.

For further information about this study, email **transition.study@sydney.edu.au** or visit: **http://sydney.edu.au/health_sciences/afdsrc/young_adults/underway/transitions_adulthood.shtml**

RESEARCH

Boost for Leukemia Research

A new research centre to fast-track leukaemia and other blood cancer research projects from laboratory to hospital bedside was opened by the Minster for Health, David Davis.

The Australian Cancer Research Foundation Centre (ACRF) for Translational Research will speed cutting edge gene discoveries into better ways of detecting, treating and preventing leukaemia and other cancers of the blood.

Established with a $1million grant from the ACRF, the facility is located at The Royal Melbourne Hospital, which sees more than 120 new leukaemia cases and performs some 80 bone marrow transplants each year.

Executive Director of Research at The Royal Melbourne Hospital, Prof Ingrid Winship said such a centre was needed to improve outcomes for patients.

“Cancers of the blood are the third greatest cause of cancer death in Australia and place a major burden on our community. They are among the most expensive to treat, and for many patients current chemotherapy options have limitations,” Prof Winship said.

However, a number of world-leading advances have been made in gene discovery, and a Translational Centre such as this provides scientists and clinicians with a unique opportunity to rapidly progress these discoveries to clinical significance.

“This progression will have a positive impact on the diagnosis and treatment of hematological malignancy, leading to improved outcomes for patients, namely the ability to provide patient-specific therapy based on ‘translational’ discoveries. Thanks to the generosity of the ACRF, this Centre gives hope to thousands of people with blood cancers,” Prof Winship said.

The Translational Research Centre will provide a seamless link between the basic research facilities, including the Walter and Eliza Hall Institute of Medical Research, the University of Melbourne and The Royal Melbourne’s Diagnostic Haematology Laboratory and clinical services.

Australian Cancer Research Foundation Chief Executive David Brettell said "The ACRF is delighted to have provided seed-funding for the development of this world-class facility. The scientists involved in this undertaking truly are experts in the field of leukaemia research, and the best way to speed up the treatments and cures for this most devastating class of cancers is to allow our scientists to work with the most modern, cutting-edge technologies and facilities possible.

Victoria is widely acknowledged around Australia for its incredible research teams, and the ACRF is proud to have provided $24 million to support these scientists in developing better preventative, diagnostic and treatment measures for all types of cancer.”

The Centre will also serve as the template for the development of similar facilities investigating genetic links in solid tumour malignancies. While initially housed at The Royal Melbourne Hospital, it will eventually move to the Victorian Comprehensive Cancer Centre (VCCC), of which the hospital is a partner.

The $1billion VCCC will bring together research, education and treatment in what will be the largest cancer centre in Victoria, becoming a key referral service for the treatment of rare and complex cancers and providing support for regional cancer services to improve local access to specialists and multi-disciplinary care.

Source: Health Vic Vol 4, May 4 2012, p.16. ■

L to R: Prof Ingrid Winship, Executive Director Research, Melbourne Health, The Hon David Davis, MP, Minister for Health, Mr Carrillo Gantner AO, Trustee ACRF, Mr David Brettell, Chief Executive, ACRF

RESEARCH

Families' Experiences of ERT

by Rebecca Freedman, GSNV Volunteer

As part of the Master of Genetic Counselling course, each student submits a thesis to the university.

For many students, the projects provide a valuable opportunity to speak directly with individuals and families who have been touched by genetic conditions. This enables us to develop a greater understanding of the experiences of those living with genetic conditions and to some extent retell their stories.

My project was entitled "Receiving enzyme replacement therapy for a lysosomal storage disorder; exploring the experiences of young patients and their families". This was essentially a quality of life study, looking into influence of living with a chronic genetic condition and receiving an intense form of treatment on the health related quality of life (HRQoL) of young patients, their parents and siblings.

The lysosomal storage disorders are a large group of inherited metabolic conditions. Each is caused by the absence of specific enzyme in the cells of the body. Enzyme replacement therapy (ERT) is now available to treat some of these conditions, including MPS I, MPS II, MPS VI, Pompe disease, Gaucher Disease and Fabry Disease.

The treatment works by providing the body with the enzymes it is missing. The aim is to prevent further progression and alleviate some of the symptoms of condition. Whilst this is an exciting advancement and so far we are seeing some great results, ERT is a treatment, not a cure.

The intervention itself is time consuming and demanding, patients are required to come into hospital once a week or once fortnight and receive the ERT through an intravenous infusion which dependent on the different condition and dose may last from two to six hours. In conducting this study I spoke to young people and to the parents and siblings of young people currently receiving ERT for MPS I, MPS II, Pompe disease or Gaucher disease. Participants took part in conversational style interviews with me in which they discussed the impact of the condition and the influence of receiving ERT on their lifestyle and that of their families.

The findings from this research, which are summarised below offer important insights for those who work with young people undergoing intensive treatment for a genetic condition and their family.

In reflecting on my research, I'd like to acknowledge the participants. Their courage and optimism was truly inspirational and I wish them all the best of luck with their ongoing treatment and future endeavours.

Research findings and conclusions

Receiving ERT results in several physical benefits for patients, in particular significant increases in energy levels were reported. The physical benefits have a subsequent positive impact on psychological and emotional well-being.

Education was deemed important by parents and young people, however parents expressed greater concerns about absenteeism, whereas young people felt they could manage to keep up with the demands of the curriculum and were less concerned with the amount of school missed.

Most participants described adaptive ways of coping with challenges, adjusting to treatment, life with the condition and managing stressors. Some participants also described experiences that had been stressful or traumatic. It is important for professionals working with this population to recognise psychological distress or sources of stressors and assist where possible. Access to ERT, attending additional appointments and the physical features of the conditions impact on activities such as travel, recreation and employment. Participants felt possessing a good working knowledge of the disorder and its treatment was important and linked this to fostering young people's independence.Young people (siblings and patients) commented on the importance of friendship and enjoyed pursuing recreation activities (such as sport).

Maintaining open communication with family members and health professionals alike was considered important. The manner in which information was provided was thought essential, particularly in the early stages of the journey with the condition, the diagnosis and prognosis.

Participants desired more flexibility with the way services were delivered, including greater choice about location and time; however they were prepared to access infusions at less convenient times because they felt the benefits outweighed the consequences of missing infusions or discontinuing the treatment.

Suggestions for future research arising from this study

Investigate opinions on home infusion in more detail, as several participants stated they would consider involvement in a clinical trial should one be conducted in Victoria.

Examine how best to educate health professionals in the wider community about LSDs or what to do when considering the possibility of rare genetic conditions and how to provide information to newly diagnosed families in a sensitive yet accurate way.

Other acknowledgements

Thank you to those who assisted me throughout this project – my research advisors, Heidi Peters, Margaret Sahhar and Lisette Curnow and to Jan Hodgson and Alison Archibald for their help. ■

Reform Underway in the not-for-profit sector

The Consumer Health Forum (CHF) has provided input to two recently-completed Treasury consultations on not-for-profit (NFP) sector reform, forming part of the government's commitment to strengthening and reform of the NFP sector.

In December, Assistant Treasurer, the Hon Bill Shorten MP, released exposure draft legislation for the establishment of a new independent statutory office, the Australian Charities and Not-for-profits Commission (ACNC).

The ACNC's role will be to work to improve public trust and confidence in the NFP sector, promoting governance, accountability and transparency of the sector.

The Treasury also released a Consultation Paper on not-for-profit governance arrangements, to result in the development of high-level principal-based mandatory requirements for the NFP bodies registered by the ACNC as well as guidance on good practice.

NATIONAL FOCUS

Frambu Resource Centre, Norway

By Dr Amy Herlihy, Public Health Genetics, Murdoch Childrens Research Institute

On Friday 1 June we had the pleasure of hosting an international colleague from the Frambu Resource Centre for Rare Genetic Disorders in Norway.

Dr Krister Fjermestad is a clinical psychologist with a PhD in child treatment research. Krister works at one of 16 Norwegian Government-funded specialist centres for rare disorders and current research at the Frambu Centre focuses on genetic conditions including Klinefelter syndrome, Fragile X syndrome and Turner syndrome. Krister presented to Genetic Health Services Victoria about the approach to rare genetic disorders in Norway.

To set the scene a little, Norway is a small Scandinavian country with a population of about 5 million, and around 30,000 people living with a rare hereditary or congenital disorder. Sixteen specialist centres for rare disorders are located across Norway, and the Frambu Centre is one of these.

'Frambu aims to collate, develop and impart knowledge about rare disorders and disabilities on behalf of individuals both with and without a rare disorder, their close/immediate family and the professionals who work with them, so that children, adolescents and adults with impaired abilities can live a life in harmony with their condition, aspirations and needs."

The goal of these centres is to develop individuals' strengths, encourage empowerment through information, build resilience and coping strategies and focus on ways individuals can reach their potential and have the best life possible, in spite of difficulties they may experience as part of having a genetic condition.

The Frambu Resource Centre is a truly exceptional service. As Krister explained, no medical treatment is carried out at the Centre, but rather it is a place for support, advice, exchange of ideas, information and assistance to those who are impacted by genetic conditions, across all ages.

The Centre has a permanent staff body of around 85 and offers a phone service and an outreach service. One of the main functions of the centre is to offer one-week residential courses, which are regularly run around a particular genetic condition. Frambu serves around 100 rare disorders. A course is offered for the most frequent of these around every second year. For example, every 24 months, there may be a week-long course held focussing on Klinefelter syndrome, where adults and children with KS and their families can attend.

During the week, a number of relevant information sessions are held, including medical updates, personal stories and invited speakers.

Usually, at least one of these days is also open to health professionals to attend to receive up-to-date information about the condition and current research in the area. Occasionally, trans-diagnostic courses are held, for instance for grandparents or youth.

During the summer months, Frambu arranges youth camps where children and adolescents spend 10 days at Frambu without their parents/guardians.

An important component to these residential courses is the social activities - which include sleighing in the snow, ice skating and sailing, as the Centre is located amongst beautiful scenery and next to a lake!!

User evaluations at the centre show those taking part in these residential courses receive immense value in meeting others who may have similar experiences, and this highlights the important social component of the stays.

We were all highly envious at Krister's presentation to hear about this amazing service. We hope that we can continue to develop the relationship between our counties, and learn from each other's experiences so that we can continue moving towards providing improved services and information for those diagnosed with genetic conditions here in Victoria and elsewhere.

More information about the Frambu Centre can be found here: **http://www.frambu.no/index.asp?lang=en**

FRAMBU Resource Centre for Rare Disorders

CMTAA Victoria Seminar

May 27, 2012

Previously* the GSNV published information on the management of Charcot Marie Tooth (CMT) in the classroom and the importance of physiotherapy, occupational therapy and educators working together to achieve the best outcomes for children and adults living with the condition.

The recent Charcot Marie Tooth Association of Australia Victorian seminar entitled *'Get up and Go'* further explored this theme and provided an excellent annual update to all who attended.

The presentation by Dr Catherine Green who is a practicing chiropractor explored the benefits of chiropractors in the treatment of CMT and how this allied health profession is progressing with knowledge and understanding of CMT.

Associate Professor Joshua Burns discussed the best practice services offered for CMT through the Children's Hospital at Westmead Neurosciences centre and the fantastic advances in diagnostics treatment and therapy in Australia.

A recurring theme during question time and the open discussion was that of the 'coordination' and management of CMT and who ultimately takes responsibility for the overall health care of the patient. Is it the GP, the physiotherapist, geneticist or the patient?

The GSNV looks forward to supporting the activities of CMTAA Victoria in the future and participating in important information sessions that empower and educate individuals and families affected by CMT.

*GSNV newsletter (Autumn 2012, Members Stories)

Counting on BloodTrack

Handheld scanners and information rich barcodes are making a difference at the bedside by helping to ensure patients are matched with the right blood product.

The Alfred became the first hospital in Australia to pilot *BloodTrack* last year, and its success was presented at the national haematology conference in October 2011.

Transfusion nurse, Christine Akers said *BloodTrack* is about enhancing safety and minimising error in the delivery of blood products.

"When a patient is admitted they are issued with a wristband, which includes their personal details in both eye-readable and 2D barcode format," Christine said.

"If they then need a blood transfusion, the blood bank cross-matches blood for the patient and prints a compatibility label for that unit of blood. The label contains all the details necessary to safely match that unit of blood to the patient.

"When the unit arrives on the ward, the nurse scans the 2D barcode on the patient's wristband and compatibility label using a handheld PDA to prompt them through the necessary checks," Christine explained.

"This is a far more secure way to check that the correct blood product is being given to the correct patient. If there's a mismatch, the scanner sounds an alert.

"The new system significantly increases the safety of the bedside check, and both patients and nursing staff have been satisfied and feel confident when it is used."

The *BloodTrack* pilot project was supported by Whole Timers Medical Specialist funding.

Reprinted from *Alfred HealthLink*, April/May Edition, Alfred Health. ■

GSNV in 2012

February

2 Early Intervention
7 DH and Melbourne Uni
16 GSNV Committee Meeting
29 Rare Disease Day

March

2 Seminar: Top 5 Issues for People with Rare Disease
7 Early Intervention Education Session
15 GSNV Committee Meeting

April

4 Grand Rounds Seminar: So you're saying it's all in my head
18 RCH Grand Rounds : The ethics of disagreement between treating team and parents: The Zone of Parental Discretion
19 GSNV Committee Meeting
20 Victorian Clinical Genetics Service new look brand and architecture

May

15 Child Neuropsychology Group Workshop: Shifting the Focus Of Rehabilitation: Therapists, Children and Families Working Together.
17 GSNV Committee Meeting
27 CMTAA Victoria Seminar

June

1 Genetic Health Services Victoria (GHSV) Seminar: An international model for rare genetic disorders, Krister Fjermestad
7 Clinical & Public Health Seminar: Reproductive Genetic Carrier Screening and Offering Fragile X Syndrome Carrier Screening

FUNDRAISING & EVENTS

Appearance Matters Symposium

Earlier this year, OzCleft and CleftPALS Victoria jointly convened the Appearance Matters Symposium which brought together health professionals, researchers, parents and young people.

Presenters included Professor Nicky Rumsey, Dr Di Harcourt, Dr Heidi Williamson and Dr Phillippa Diedriches from the Centre for Appearance Research (CAR) in Bristol, UK. The audience were challenged on their pre-conceptions relating to the psychology of appearance along and distilling beauty myths, alongside research into body image and disfigurement.

Whilst clinicians and researchers discussed best practice in working with families, parents had the opportunity to discuss how best to prepare both their child and school for transition into prep, secondary school or a new educational facility.

In a concurrent session, the Cleftstars and other young people present were asked to review the 'YP Face It' online modules to work through some of the competency exercises which focussed on issues such as overcoming fear, responding to peer remarks, making new friendships and developing social confidence and skills. While these modules were greeted with much enthusiasm, it may be some time before resources such as this could become available.

This inspired discussion amongst the groups, reaffirming the value and importance of forums like these that bring young people together. While the symposium focussed on cleft lip and palate, the issues discussed were relevant to many other genetic conditions, as well as those without given the emphasis society puts on appearance.

This symposium received lots of positive feedback as it provided useful tips and information for anyone concerned, or interested, in appearance.

For more information on the research conducted by the CAR visit: **http://www1.uwe.ac.uk/hls/research/appearanceresearch**

Thank you to MotorOne

The GSNV is pleased to announce that we have received an injection of funding of over $8,000 from the MotorOne group.

The MotorOne Group held a very successful fundraiser called 'Stop Drop and Bowl' as a team building exercise and as part of a management project conducted by a number of staff to bring the all the MotorOne business units together. 'Stop Drop and Bowl' was dedicated to the GSNV and Louisa Di Pietro attended, representing the GSNV Committee, staff and community. We had anticipated around $3,000 to $5,000 dollars and were ecstatic to receive a very large cheque for over $8,000!

Sales of raffle tickets, donations of prize packs from motor vehicle dealerships and service providers and generous donations from business owners exceeded our expectations. It was wonderful to see how an industry group rallied together, embraced the GSNV vision and message and raised money for us! The evening was spectacular and as a charity event and team building exercise, it could not have gone better.

The GSNV specifically thanks John Weekley (WorldMark), Greg Lewis (MotorOne), Warren Koopmans (Tint A Car), David Lowrie (Sewells), George Mariotto (MEP), Tim Thorne and all who participated from High Performance Corporation, TCB Car Tinting, Top Edge Tinting, GWPM and VM Smiley.

We particularly congratulate and thank Sylvia Smiley, Marcia Yianni, Natasha Olive and Marcus Modoo from MotorOne, for their commitment, dedication and incredible negotiating skills in getting this project off the ground and making it such a success.

Thank you to all involved for providing an opportunity for the GSNV to give back to our support groups and community in helping them conduct their important work.

Proceeds from this event will be dedicated to the GSNV Small Grants Scheme 2012 and Special Grants 2012.

SUPPORT GROUP NEWS

Introfish – created by demand, powered by community spirit

Introfish is a specialised fishing service dedicated to people with special needs, or who are in rehabilitation, and was founded by keen fisherman Paul Bennett.

Introfish is a registered not for profit incorporation who work with individuals, their coordinators, carers and parents to provide a fun and active learning experience.

Boat fishing trips from October to April and land based trips throughout winter are conducted on Western Port and Port Phillip Bay, local beaches inland impoundments and various Lakes around Victoria.

The group rely on sponsors and generous donations to conduct these fishing trips with respite organisations. The number of bookings often exceeds the funds available, showing the great demand for this service.

The feedback received by the group so far has been outstanding, with praise being given to the professional way in which the organisation conduct their trips, as well as their patience and effective teaching and communication skills of those involved. The service has recently also been expanded to offer the opportunity to troubled youth in government care, again with outstanding results.

Paul Bennett would like to pass on his appreciation for the support received so far as "without it we couldn't show the community what we could achieve and how we can enhance the lives of people with special needs".

Introfish are currently applying for a $1000 grant through Leader Local Grants.
To vote for this organisation, visit **http://leaderlocalgrants.com.au**

For more information visit their website: **http://introfish.org**

Paul Bennett leading a successful fishing trip in November, 2011

Little Dreamers founders Madeleine and Rebecca with Adele and Erin who were the recipients of the very first Dream in 2010.

Little Dreamers

Little Dreamers is a charity that was set up in 2009 by two 16 year old girls who wanted to remind siblings of sick children and young carers that they are special.

Little Dreamers grants Dream Experiences for these special kids who sometimes believe that their happiness is not as important as other people in their house. They also aim to educate the public about the issues facing siblings and young carers.

Dreams granted so far have included an overnight stay at the Melbourne Zoo and the Hilton Hotel, a ride in a pink limousine, and tickets to Hairspray the Musical.

If you know someone who deserves a Dream Experience, or for more information about Little Dreamers, phone 0433 818 212 or send an email to **info@littledreamersonline.com**. Dream Application packs can be downloaded from **www.littledreamersonline.com** or completed online or print copies are available by post.

Source: NoticeBoard Magazine of the Association for Children with a Disability, Spring 2011.

PERSONAL STORIES

Notes from a Dragon Mom

By Emily Rapp, taken from New York Times Sunday Review 15 October, 2011

My son, Ronan, looks at me and raises one eyebrow. His eyes are bright and focused. Ronan means "little seal" in Irish and it suits him.

I want to stop here, before the dreadful hitch: my son is 18 months old and will likely die before his third birthday. Ronan was born with Tay-Sachs, a rare genetic disorder. He is slowly regressing into a vegetative state. He'll become paralysed, experience seizures, lose all of his senses before he dies. There is no treatment and no cure.

How do you parent without a net, without a future, knowing that you will lose your child, bit by torturous bit?

Depressing? Sure. But not without wisdom, not without a profound understanding of the human experience or without hard-won lessons, forged through grief and helplessness and deeply committed love about how to be not just a mother or a father but how to be human.

Parenting advice is, by its nature, future-directed. I know. I read all the parenting magazines. During my pregnancy, I devoured every parenting guide I could find.

My husband and I thought about a lot of questions they raised: will breast-feeding enhance his brain function? Will music class improve his cognitive skills? Will the right preschool help him get into the right college? I made lists. I planned and plotted and hoped. Future, future, future.

We never thought about how we might parent a child for whom there is no future. Our parenting plans, our lists, the advice I read before Ronan's birth make little sense now. No matter what we do for Ronan — choose organic or non-organic food; cloth diapers or disposable; attachment parenting or sleep training — he will die. All the decisions that once mattered so much, don't.

All parents want their children to prosper, to matter. We enrol our children in music class or take them to Mommy and Me swim class because we hope they will manifest some fabulous talent that will set them — and therefore us, the proud parents — apart.

Traditional parenting naturally presumes a future where the child outlives the parent and ideally becomes successful, perhaps even achieves something spectacular.

Amy Chua's "Battle Hymn of the Tiger Mother" is only the latest handbook for parents hoping to guide their children along this path. It's animated by the idea that good, careful investments in your children will pay off in the form of happy endings, rich futures.

But I have abandoned the future, and with it any visions of Ronan's scoring a perfect SAT or sprinting across a stage with a Harvard diploma in his hand. We're not waiting for Ronan to make us proud. We don't expect future returns on our investment.

We've chucked the graphs of developmental milestones and we avoid parenting magazines at the paediatrician's office. Ronan has given us a terrible freedom from expectations, a magical world where there are no goals, no prizes to win, no outcomes to monitor, discuss, compare. But the day-to-day is often peaceful, even blissful.

This was my day with my son: cuddling, feedings, naps. He can watch television if he wants to; he can have pudding and cheesecake for every meal.

We are a very permissive household. We do our best for our kid, feed him fresh food, brush his teeth, make sure he's clean and warm and well rested and ... healthy? Well, no. The only task here is to love, and we tell him we love him, not caring that he doesn't understand the words. We encourage him to do what he can, though unlike us he is without ego or ambition.

Ronan won't prosper or succeed in the way we have come to understand this term in our culture; he will never walk or say "Mama," and I will never be a tiger mom. The mothers and fathers of terminally ill children are something else entirely.

Our goals are simple and terrible: to help our children live with minimal discomfort and maximum dignity. We will not launch our children into a bright and promising future, but see them into early graves.

We will prepare to lose them and then, impossibly, to live on after that gutting loss. This requires a new ferocity, a new way of thinking, a new animal.

We are dragon parents: fierce and loyal and loving as hell. Our experiences have taught us how to parent for the here and now, for the sake of parenting, for the humanity implicit in the act itself, though this runs counter to traditional wisdom and advice.

Nobody asks dragon parents for advice; we're too scary. Our grief is primal and unwieldy and embarrassing. The

PERSONAL STORIES

certainties that most parents face are irrelevant to us, and frankly, kind of silly.

Our narratives are grisly, the stakes impossibly high. Conversations about which seizure medication is most effective or how to feed children who have trouble swallowing are tantamount to breathing fire at a dinner party or on the playground. Like Dr. Spock suddenly possessed by Al Gore, we offer inconvenient truths and foretell disaster.

And there's this: parents who, particularly in this country, are expected to be superhuman, to raise children who outpace all their peers, don't want to see what we see. The long truth about their children, about themselves: that none of it is forever.

I would walk through a tunnel of fire if it would save my son. I would take my chances on a stripped battlefield with a sling and a rock à la David and Goliath if it would make a difference. But it won't.

I can roar all I want about the unfairness of this ridiculous disease, but the facts remain.

What I can do is protect my son from as much pain as possible, and then finally do the hardest thing of all, a thing most parents will thankfully never have to do: I will love him to the end of his life, and then I will let him go.

But today Ronan is alive and his breath smells like sweet rice. I can see my reflection in his greenish-gold eyes.

I am a reflection of him and not the other way around, and this is, I believe, as it should be.

This is a love story, and like all great love stories, it is a story of loss. Parenting, I've come to understand, is about loving my child today. Now. In fact, for any parent, anywhere, that's all there is.

Source: http://www.nytimes.com/2011/10/16/opinion/sunday/notes-from-a-dragon-mom.html

Emily Rapp is the author of "Poster Child: A Memoir," and a professor of creative writing at the Santa Fe University of Art and Design. ■

Emily Rapp and her son, Ronan, who has Tay-Sachs disease.

Tay–Sachs disease

Tay-Sachs disease is a degenerative condition of the nervous system, and is caused by changes in both copies of the HEXA gene.

Symptoms often appear at around six months of age. Babies with Tay Sachs Disease don't often live past their fifth birthday.

While the condition is particularly common in Jewish people from central and eastern Europe (Ashkenazi Jews) as well as French-Canadians, people of other nationalities can also carry the faulty gene. Worldwide, about one in every 30 Ashkenazi Jews and one in 40 French-Canadians are genetic carriers of TSD.

In Australia, however, about one in every 25 Ashkenazi Jews is a genetic carrier for TSD. On the other hand, about one in 280 Jewish people from the Middle East (Sephardic Jews) and non-Jewish people will be genetic carriers of TSD.

If a man and a woman each carrying a faulty HexA gene conceive a child, there is a one in four chance that the child will have TSD. If only one parent has the faulty gene, the child won't develop the condition but has a 50 per cent chance of being an unaffected genetic carrier, just like their parents.

Source: http://www.betterhealth.vic.gov.au/bhcv2/bhcarticles.nsf/pages/Tay-Sachs_disease

IN BRIEF

NEW eHEALTH LEARNING CENTRE GOES LIVE

An online "learning centre" which details how the Australian Government's new electronic health record system will work for patients and practitioners has been launched.

From July 2012, people seeking healthcare in Australia will be able to register for their own personally controlled electronic health record – an eHealth record. Your eHealth record will be a secure online summary of your key healthcare information. You will control what goes into your eHealth record, who is allowed to access it, and who can see which information.

To find out more and to register, visit: **http://www.ehealth.gov.au/internet/ehealth/publishing.nsf/content/home#.T7mJlVKkLTp**

Source: DARU Update 21 May 2012

GSNV SMALL GRANTS SCHEME 2012

Every year the GSNV offers Small Grants to members for projects that support the work of a support group.

In 2012, the GSNV is reviewing its membership policy and has not issued membership renewals as yet.

However, in September/October of this year, the GSNV will allocate Small Grants and continue to support important projects.

All grants are between $50 and $500. We encourage you to start considering small projects you may wish to be funded and be ready for our applications process which will begin in August 2012.

Stay tuned for further communications on Small Grants.

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

- An 11 year old girl with **Osteogenesis Imperfecta** would like to meet another young person who also has this condition. Contact **info@gsnv.org.au** or phone (03) 8341 6315.
- A young boy with **Ring Chromosome 8** and his family would like to meet another child with the same condition. Contact **info@gsnv.org.au** or phone (03) 8341 6315.
- A young adult with **Langer-Giedion syndrome** is looking to meet with a similarly aged person who also has the condition. Contact **info@gsnv.org.au** or phone (03) 8341 6315.
- Do you have a family member affected by **Pallister-Killean syndrome**? If you are interested in talking to Victorian families also affected by this condition contact **info@gsnv.org.au** or phone (03) 8341 6315.
- Do you have a family member with an **undiagnosed genetic condition**? If you would like to meet with a Melbourne family whose child also has no diagnosis, contact Heather on 0400 447 801. ■

CALENDAR OF EVENTS

Haemochromatosis Awareness Week
13 – 19 August, 2012

ALDS 20th Anniversary Conference
18 – 19 August, 2012
Novotel St Kilda
For more information visit
http://alds.org.au

VCFS 22q11 Foundation Conference
19 August 2012
For more information visit
www.vcfsfa.org.au

VCFS National Awareness Week
20 – 26 August, 2012

SMA Annual Gala Dinner
25 August 2012
Laila Receptions, Brunswick
For more information visit
www.smaaustralia.org.au

CMTAA Youth Camp
7 – 9 September
For More information visit **www.cmt.org.au**

CAHSGA Family Conference Day
9 September 2012
Sydney Children's Hospital
For more information visit **www.cah.org.au**

COSHG Help and Support Group Awareness Day
13 September 2012
Ross House, Melbourne
For more information visit
www.cohsg.org.au

NSW CMT Awareness Day Seminar
23 September 2012
Concord Sydney
For more information visit
www.cmt.org.au ■