



Genetic Support Council of WA Inc

Gene Advocate

Issue 65

Personalised medicine and genetics

Have you ever wondered why some people develop cancer while others don't?

Or why a medicine may work well for you, but may be less effective or cause serious side effects in someone else?

Although the differences are due to a number of important factors such as age, weight and lifestyle, our genetic makeup also plays a part.



What is meant by the term 'personalised medicine'?

The way genes interact with each other can sometimes make us more likely to develop particular diseases. Research scientists have found a number of specific links between some genes and the development of certain diseases, as well as between some genes and the effectiveness of certain medicines.

'Personalised medicine' uses this knowledge of genetics to predict disease development, to influence decisions about lifestyle choices and/or to tailor medical treatment to an individual.

The result of personalised medicine is better disease prevention, more accurate diagnosis of disease and more effective treatment of disease by understanding the way specific genes work with medicines.

What impact does 'personalised medicine' have on disease diagnosis?

In the past, disease diagnosis was based on symptoms. However, several conditions can share similar symptoms, making diagnosis difficult. Nowadays, diagnosis of some diseases has become more accurate because we are able to test for genes that are known to be associated with the disease. Genetic testing can not only identify the presence of particular diseases, it can also determine which subtype of a disease a person has.

Knowing the subtype of a disease can help determine an accurate outcome for the patient. Identification of the disease subtype also allows doctors to prescribe a drug therapy that will specifically target the subtype through the use of pharmacogenetics.

What is pharmacogenetics and how can it improve the effectiveness of drug treatment?

At present, doctors use a 'one size fits all' approach to prescribe medicines for most

"We are more than the sum of our genes!"

diseases. Patients are first given a medicine at an average dose. The doctor then makes adjustments to the dose of medicine according to the patient's response.

Pharmacogenetics (or pharmacogenomics) aims to match the best available drug or dose to a person, based on his or her genetic makeup. A process known as gene expression profiling identifies the active genes in a patient. Profiling can tell the doctor how a medicine may be broken down by the patient's body and whether or not it will be effective.

Applying pharmacogenetics ensures patients are prescribed the most effective drug right from the beginning. Pharmacogenetics also minimises the number of adverse reactions to medicines as it can show whether a patient is likely to have a toxic response.

As scientists gain a better understanding of genetic variations, new subtypes of diseases could be identified and more specific medicines that target the new subtypes may be manufactured.

How can genetic information be used in disease prevention?

Individual variations (mutations) that may be present in our genes can determine how our bodies are likely to be affected by a disease, or affect our risk of developing a particular disease. Some variations can be protective and decrease risks. Other variations can increase the likelihood of developing a particular disease. For example, a person's genetic makeup could indicate an increased risk of developing diabetes or heart disease.

Early identification of these variations and calculation of the risks associated with them are important in disease prevention. This knowledge could be used to influence lifestyle choices and decisions about possible interventions that may prevent the disease from developing, to delay disease onset or to reduce the impact of the disease.

There are currently a limited number of genetic tests available for this purpose.

However, the field of personalised medicine is certain to expand rapidly in the near future.

When will personalised medicine be a reality?

Although the science is still in the early stages, pharmacogenetics is already being used to treat some diseases such as breast cancer. Before being prescribed the drug Herceptin®, breast cancer patients are tested to examine whether a particular gene, human epidermal growth factor receptor 2 (HER2), is present in the cancer cells.

Herceptin® is only effective when cancer cells have extra copies of the human epidermal growth factor receptor 2 (HER2) gene, or too many HER2 receptors. Testing to find this out allows doctors to predict the patient's response to Herceptin®, enabling a more appropriate medicine to be prescribed immediately.

Source: NHMRC
<http://www.nhmrc.gov.au/your-health/egenetics/personalised-medicine>

Individual funding approaches for disability support

Western Australia (WA) applies the full range of individual funding approaches, with portable funds held by providers, facilitators and direct funding to individuals and families. Direct funding is facilitated through the Local Area Coordination (LAC) program, which has been running for approximately 20 years, the longest in Australia. Approximately 1,500 people receive direct payments (approximately \$10 million per year).

When LAC began 20 years ago, the funding was spent locally, consisted of small amounts and focused on flexibility. Over time, administrative, accountability and reporting requirements increased and the funding amount and complexity has grown. A review of the LAC in 2003 identified that some consumers and families managed high levels of funding in complex packages. This presented risks to the person, families and LAC from the administrative burden and responsibilities of managing the direct payments.

To resolve this risk an additional shared management model is now available, where the funding is held in partnership with the service provider and person, allowing the person varying degrees of direction and management of their support. This shared system offers a continuum of administrative responsibilities. Consumers can fully manage their funds; or have a service provider manage the administration and finances, but still allow personal control over how the funding is spent, including employment of support workers.

All funding in WA is now individualised, except some respite and therapy. For example, all accommodation support providers such as group home operators now receive funding for individuals, not block funding. WA refers to this as a 'notional allocation against each individual', not an individual package.

The rationale behind individualising the funding through a notional allocation is to facilitate portability for the consumer. A result of the funding changes is that some consumers have moved from more traditional service types, such as group homes and day centres, to more personalised support types.

In some support types, block funding is still provided to the service provider to fund capacity and therapy services, for a number of places rather than people.

Examples

WA's system of direct payments is integrated with its LAC. Each LAC works with people and families to assist them to access and manage direct payments. WA has an accountability framework with guidelines for use of funds and responsibilities for managing public funds. They also have principles to guide the development and use of direct funds. Direct payments range from one-off grants of \$50, to small flexible funding packages of around \$5,000, through to larger packages up to approximately \$100,000.

Evaluations of LAC have identified that, generally, most people and their families highly value direct payments and consider them to be a key element of the LAC. Direct payment provides a level of control to individuals and families, allows them to 'call the shots' and decide who will provide the support and when. Support is often more informal and flexible than in the past, responding to the needs of consumers.

Future developments

WA is committed to continuing to offer the range of individual funding approaches. The state is refocusing the use of direct payments to build people's natural support networks. Families are also demanding better options, life experiences and futures for their family member.

WA will explore new options for people wanting to self-manage their funds, for example, through shared management models and new support arrangements such as microboards.

Officials identified streamlining financial and administrative systems as a priority, as well as a greater focus on outcomes and flexibility. They considered that individual funding should lead to more support options and personal choices and move away from shared care, group and congregate service models.

Summary

- Australian disability agencies demonstrate a growing policy interest in individual funding and self-directed approaches. They are developing policies and guidelines, extending pilots, introducing and refining models and conducting evaluations to better understand consumer outcomes.
- The dominant approach to individual funding in Australia is a portable individual funding package held by a service provider, except in Western Australia, which has a longer history of individual funding, including a system of direct payments.
- Jurisdictions are aiming to offer a continuum of individual funding approaches, with varying levels of control and choice for the person. Jurisdictions confirmed that flexibility of service provision was a key motivator for the person with a disability and their family in seeking individual funding.
- One of the strengths of individual funding identified across jurisdictions is its potential to provide consumers with the opportunity to access a wider range of support, including non-traditional support.
- States and territories support a diversified disability support sector with a mix of funding approaches to achieve a range of viable disability support options including informal, formal, specialised, general and professional support.

Consumer preferences

The literature review suggests that good approaches to individual funding adopt principles about consumer preferences including consumer driven, person-centred support; facilitating independent living in the community; and flexibility in the way support is organised, each discussed below.

Leadbeater, Bartlett and Gallagher (2008, p. 9) argue for a transformational approach to public services, through the use of self-directed services that allocate budgets to people so they can shape the support and services they need.

Self-directed services and personal budgets can be responsive to user demand. They can shift away from inflexible, centralised, building-based services, such as day care centres and group homes, towards more flexible, informal and decentralised provision, often organised around a person's home in their community.

Consumer driven, person-centred support

According to the literature review, individual funding is intended to be driven by the person needing support in a way that focuses on empowerment, self-determination, consumer control and access to valued resources and supports. For example, the Seattle 2000 Declaration on Self Determination and Individual Funding sees self-determination as the rationale and guiding principle for individual funding.

Individual funding is underpinned by the principles and practice of person-centred support. Glynn and Beresford (2008, p. 2) describe person-centred support as:

... a new umbrella term to cover a wide range of new developments and approaches, from 'person-centred planning' to direct payments, concerned with putting service users at the centre of policy and practice to ensure that the support they receive is shaped primarily by their individual and collective rights and needs, hopes and goals.

The focus of individual funding approaches is on person-centred support, rather than on technical, administrative, financial and accountability issues (d'Aboville 2006).

Person-centred support underpins individualised planning and support as tools to enhance quality of life.

In the study, individual funding appeared to be successful overall in achieving its aim of person-centred disability support. People across all disability, funding and support types reported a high level of control over their support and a general feeling that their services were arranged around their needs. One person said:

I feel very empowered. I feel like I have power over who comes into my house, when they come into my house and what they do while they are there.

Over 90 per cent of people using individual funding or their family indicated that they were able to choose what they want to do regarding disability support type and who provides the support (See Table 9. http://www.fahcsia.gov.au/about/publicationsarticles/research/occasional/Documents/op29/part5_2.htm).

Source:

Department of Families, Housing, Community Services and Indigenous Affairs (FaHCSIA). Occasional Paper no. 29: Effectiveness of individual funding approaches for disability support. [Accessed 9/05/2011]. <http://www.fahcsia.gov.au/about/publicationsarticles/research/occasional/Documents/op29/default.htm>

Victoria to Trial Disability Insurance Scheme

The Victorian Government is set to trial the national disability insurance scheme, the Minister for Community Services Mary Wooldridge has announced - but not until 2014.

The Victorian Government has offered to trial the National Disability Insurance Scheme in 2014, to test out the concept ahead of the proposed national roll-out in 2015.

Speaking at the National Disability and Carer Congress in Melbourne recently, the Minister for Community Services, Mary Wooldridge said the new model would cut waiting times for those eligible for disability support payments, according to an ABC News report.

She said this would change the welfare-based disability support system into an entitlement-based support system.

Victorian Premier, Ted Baillieu announced earlier this year the establishment of a taskforce to advise the Government on the establishment of a new National Disability Insurance Scheme, and a National Disability Insurance Scheme (NDIS) Secretariat in the Department of Premier and Cabinet (DPC) has been set up.

Baillieu said the Victorian Government was committed to a new approach to disability funding, such as an NDIS, which would introduce a no-fault universal safety net - similar to those operated by the Transport Accident Commission and WorkSafe - for people with permanent or severe disabilities.

However the funding of the national scheme, which is expected to cost more than \$12 billion per annum, and how much will be paid by the federal and state governments, is still unclear.

Full Article Available: Pro Bono Australia 5/05/2011 <http://www.probonoaustralia.com.au/news/2011/05/victoria-trial-disability-insurance-scheme>

New Orphanet service - rare diseases & orphan drugs

Orphanet is the portal for rare diseases and orphan drugs. The new Orphanet service provides the scientific community with a free access dataset for rare diseases and orphan drugs.

Inspired by the growing number of requests for data from its database, Orphanet, the pan-European rare disease and orphan drug information portal, decided to create a comprehensive, high-quality and freely-accessible dataset related to rare diseases and orphan drugs that would be available in a reusable format. Launched last week, Orphadata was developed within the context of the RD Portal and the new Orphanet Europe joint action contracts funded by DG Sanco. Additional support is also provided by GSK.

The dataset is a partial extraction of the data stored in Orphanet and will be updated monthly. For user convenience, the date of the last data release will be provided. Freely accessible in six languages (English, French, German, Italian, Portuguese and Spanish) the Orphadata dataset encompasses:

- An inventory of rare diseases, cross-referenced with OMIM, ICD-10 and with genes in HGNC, OMIM, UniProtKB and Genatlas
- A classification of rare diseases established by Orphanet, based on published expert classifications
- Epidemiology data related to rare diseases in Europe (class of prevalence, average age of onset, average age at death) extracted from the literature
- A list of signs and symptoms associated with each disease, with their frequency class within the disease
- It is also possible, on request, to access other types of Orphanet data, including:

- An inventory of Orphan Drugs at all stages of development, from EMA (European Medicines Agency) orphan designation to European market authorisation, cross-linked with diseases

Summary information on each rare disease in six languages (English, French, German, Italian, Spanish, Portuguese)

URLs of other websites providing information on specific rare diseases

A directory of specialised services, providing information on centres of expertise, medical laboratories, diagnostic tests, research projects, clinical trials, patient registries, mutation registries, and patient organisations in the field of rare diseases, in each of the countries in Orphanet's network.

Orphadata provides a guide for users that defines and describes the elements of the dataset. Orphanet hopes that this latest offer will contribute to furthering research toward better understanding and treating rare diseases.

The logo for Orphadata features the word "orphadata" in a lowercase, sans-serif font. The letters are colored: 'o' is blue, 'r' is grey, 'p' is grey, 'h' is grey, 'a' is orange, 'd' is yellow, 'a' is purple, and 't' is green. The 'i' is replaced by a small red dot above the 'a'.

Orphadata Available:

<http://www.orphadata.org/cgi-bin/index.php>

Erbix on new subsidised medicines list

Minister for Health and Ageing Nicola Roxon Media Release 21 June 2011.

More than 400,000 Australians will benefit from new subsidised medicines from 1 September 2011, including patients suffering multiple sclerosis, cystic fibrosis and various forms of cancer. Minister for Health and Ageing Nicola Roxon said the \$200 million announcement includes the listing of Cetuximab (Erbix®) to the PBS. Erbix, which can cost up to \$30,000 a year without a subsidy, will assist around 2700 patients who are suffering with late-stage bowel cancer.

Minister Roxon said that the listing of Erbix was complex given that it is the first time any Government has listed a medicine on the PBS while also attempting to list a co-dependent genetic test on Medicare. While the assessment of Medicare funding for the genetic test is still ongoing, the drug manufacturer has offered to pay for the genetic test in the meantime. This offer has allowed the Government to bring forward its consideration of Erbix®.

Other additions or extensions to the PBS and National Immunisation Program, subject to final listing arrangements being met, include:

- Fluconazole (Diflucan®) - for certain fungal infections in immunocompromised children and the elderly.
- Imatinib (Glivec®) - for patients with a high risk of recurrence of tumour following the surgical removal of a gastrointestinal tumour.
- Pegfilgrastim (Neulasta®) – to stimulate the production of white cells in immunocompromised patients with Hodgkin disease.
- Tobramycin (Tobi®) - for treatment of lung infection caused by a specific bacterium in patients with cystic fibrosis.

- Dalteparin Sodium (Fragmin®) - for the treatment of blood clots.
- Eltrombopag (Revolade®) - to treat adult patients with a potentially life threatening bone marrow disorder.
- Ferrous fumarate (Ferro Tab®) - for the treatment of anaemia.
- Flupenthixol (Fluanxol®) and zuclopenthixol (Clopixol®) - to treat patients with psychosis.
- Risperidone (Risperdal Consta®) – a long-acting injection for patients with bipolar disorder.
- Dutasteride with tamsulosin hydrochloride (Duodart®) - a treatment for enlarged prostate.

The Government has also agreed to increase the price of three medicines currently listed on the PBS - Amiloride Hydrochloride Tablet, 5mg (Kaluril®), Chlorthalidone (Hygroton 25®) and Thiamine Hydrochloride (Betamin®). In addition, the Government has agreed to list the new 60mg vial size of Herceptin through the Medicare-administered late-stage Herceptin program.

The Government will also list Fluarix®, a vaccine to protect against seasonal influenza, on the National Immunisation Program later this year.

Since 2007, we have listed over 500 new medicines and brands at a cost of more than \$4 billion.

For all media inquiries, please contact the Minister's Office on 02 6277 7220

Media Release

<http://www.health.gov.au/internet/ministers/publishing.nsf/Content/mr-yr11-nr-nr123.htm>

Endometrial cancer gene found

In a world first, researchers from the Queensland Institute of Medical Research (QIMR) and the University of Cambridge have identified an area of the genome that increases the risk of developing endometrial cancer.

Endometrial (uterine) cancer is the most common gynaecological cancers in the women of developed countries. It affects nearly 2,000 Australian women, and causes approximately 300 deaths per year.

Dr Amanda Spurdle from QIMR's Molecular Cancer Epidemiology Laboratory said this is a significant discovery. "We hope that studies like this will provide us with better understanding about how gynaecological cancers develop and what influences the risk of disease."

"This is the first endometrial cancer gene identified using the genome-wide association study approach, and involved comparing more than 1,200 endometrial cancer patients to more than 5,000 unaffected people for more than 500,000 genetic markers across the genome."

"The area we have identified is located close to the HNF1B gene, which has previously been associated with prostate cancer risk. It was surprising to learn that this area of the genome might also influence endometrial cancer risk, since these two cancers were not previously recognised to have similar underlying genetic causes."

"Our results show that research studies that screen genetic markers across the entire genome are important to identify previously

unknown cancer genes and pathways for future clinical studies."

This research was part of an international study including groups from Australia, the UK, Germany, Belgium, China, Norway, Sweden, Switzerland, and the USA and would not have been possible without funding from the National Health and Medical Research Centre.

The paper will be online published in the prestigious journal Nature Genetics on Monday 18 April.

Source:

http://www.qimr.edu.au/page/News__Events/Media_Centre/Media_Releases/Archive/2011/Endometrial_gene_found/

About Endometriosis

Endometriosis is a condition where the endometrial cells (cells that form the lining of the uterus) begin growing elsewhere in the body.

The endometrial tissue implants itself onto the surface of the tissue or organ where it has been deposited and begins to grow.

Endometriosis is very difficult to diagnose, because the symptoms are not well understood, and vary from one woman to the next. The condition is known to be associated with severe period pain and infertility. Some women may not have any symptoms at all.

Source:

http://www.qimr.edu.au/page/Our_Research/Research_Areas/Human_Genetics/Endometriosis/

Genetic Testing for Kids: Is It a Good Idea?

By Bonnie Rochman. TIME Magazine
April 18, 2011

With the advent of new genetic tests, it's increasingly easy to gauge whether you're predisposed to developing certain conditions — diabetes, say, or breast cancer. For adults, that knowledge can be simultaneously overwhelming and empowering.

For children, the ramifications of such predictions are especially controversial, which is why professional groups — the American Academy of Pediatrics (AAP - www.aap.org), in particular — have come out against genetic testing of children for adult-onset diseases.

Yet a new study published today in the AAP-affiliated journal *Pediatrics* finds that parents who were offered the option of genetic testing for themselves said they would also like to test their children.

The 219 parents surveyed indicated they believe that the risks of testing their children for eight adult-onset conditions — colon, skin and lung cancer; heart disease, high blood pressure, high cholesterol, Type 2 diabetes and osteoporosis — outweigh the benefits.

Full Article Available: <http://healthland.time.com/2011/04/18/genetic-testing-for-kids-is-it-a-good-idea/>

Modern Governance and Community Groups

Governance is about how your organisation is run – the structures and systems and understandings that enable you to make the right decisions and set the right course. It's not the same thing as management – what the Board does is governance; what the CEO does is management, although the two functions can overlap to some extent at the upper levels.

Governance isn't your actual policies, either, or your mission or your direction; it's how your organisation decides what that mission and that direction and those policies are.

There's no one perfect organisational solution, and a not-for-profit organisation can do very well under a number of possible structures of governance. What you're looking for is a structure that can combine:

Responsiveness Everybody who has an interest in a decision, and everybody who has information about the topic, should be able to be heard; if possible, everybody who has an opinion on the subject should be listened to.

Decisiveness After everyone has been heard a decision has to be made, and that decision must be implemented wholeheartedly across the organisation.

Ease of use As far as possible the system should be transparent, flexible, and straightforward. It should be possible to see immediately who's responsible for what, and how that responsibility relates to the whole mission and to other sectors of the organisation.

Complete Help Sheet Available:
Our Community Pty Ltd. http://www.ourcommunity.com.au/boards/boards_article.jsp?articleId=1298

World-first: Glaucoma genes found

Queensland Institute of Medical Research

MEDIA RELEASE.

03-May-2011

As part of a team of Australian researchers, scientists at the Queensland Institute of Medical Research (QIMR) have discovered two new genes linked to open angle glaucoma in the only study of severe glaucoma cases in the world, published online in the prestigious international science journal, Nature Genetics.

Dr Stuart MacGregor from QIMR's Queensland Statistical Genetics Laboratory said, "This discovery will help to identify patients at the highest risk of severe glaucoma. It opens the pathway to developing completely new ways of treating glaucoma patients that could delay disease progression and prevent blindness."

Open angle glaucoma, sometimes called chronic glaucoma, is the most common type of glaucoma, and tends to progress at a slow rate. Sufferers may not notice that they have lost vision until the disease has progressed significantly.

"We found that 18% of the population carry risk variants at these two genes, making them up to three times more likely to develop severe glaucoma than those that don't."

Glaucoma is the leading cause of irreversible blindness worldwide,

affecting an estimated 300,000 people in Australia, half of which are currently undiagnosed. It is the collective name for eye diseases causing irreversible loss of peripheral vision, often associated with too much pressure developing inside the eyeball.

"Although open angle glaucoma is the most common form of the disease, it is poorly understood and difficult to diagnose in its early stages. Many cases still remain undiagnosed until irreversible loss of vision has occurred."

Researchers from QIMR collaborated with groups from five other Australian universities on the ground-breaking study of 4,500 patients from Australia and New Zealand.

Dr MacGregor emphasized the important role of twins in the research. The genetic information provided by the twins was used in the study as a comparison to those affected by glaucoma.

"We would like to thank all the twins who have generously given their time to help medical research in Australia maintain an international standard, and help Australian research lead the way for better health worldwide."

The twins have volunteered to be part of a Queensland-based register, called QTwin.

The paper "Genome-wide association study identifies susceptibility loci for

open angle glaucoma at TMCO1 and CDKN2B-AS1" will be available at <http://dx.doi.org/10.1038/ng.824>.

QTwin is a Queensland-based registry of identical and non-identical twins who regularly provide information for medical research. By comparing identical twins to non-identical twins, QIMR researchers can measure the impact of genes versus environment to better understand the causes of disease: the nature versus nurture debate.

If you know any identical and non-identical twins, please encourage them to register at <http://ww.qtwin.org.au/> or by calling 1800 257 179.

Source: http://www.qimr.edu.au/page/News__Events/Media_Centre/Media_Releases/World-first_Glaucoma_genes_found/



Glaucoma Australia Support Group

Glaucoma Australia is the peak glaucoma awareness/education/support association in Australia. It is a registered, national, not-for-profit organisation incorporated in 1988.

Contact Details

Support line 1800 500 880

Web: <http://www.glaucoma.org.au>

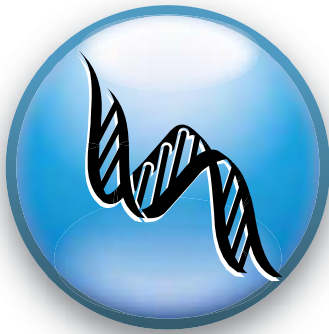
Students Living With a Genetic Condition: A Guide for Parents

Introduction

As parents of a child living with a genetic condition, you may feel apprehensive about sending your child to school. You know how to identify and manage symptoms at home, and it may be scary to have someone else manage your child's medical care in your absence. Before your child begins school, it may be important to explain your child's condition and how it may impact your student's school experience to certain members of the school staff. You may do this by sending a letter or by going to the school for a meeting.

This Guide provides information that may be helpful when writing a letter or when preparing to meet with your student's teacher, school nurse, and physical education teacher or coach. This may also be a useful resource for bus drivers, babysitters, church leaders, or any other potential caregiver. This packet includes general template letters that can be adapted to fit your personal needs. Once you develop your personal resource, it may be useful to laminate your resource and give it to your student to keep in his or her backpack. In addition, this resource includes ideas for how your child can talk to classmates or peers about his or her genetic condition.

Available: Genetic Alliance. How-to Guides. Accessed 2/06/2011. http://www.geneticalliance.org/sites/default/files/ksc_assets/pdfs/attending_with_genetic.pdf



Member Profile

The Chromosome 18 Registry & Research Society (Australasia) Inc

We are the Australasian arm of The Chromosome 18 Registry & Research Society which is based in San Antonio, Texas, USA. We cover all Australian states, New Zealand and Oceania.

We provide a contact point for individuals and families affected by a chromosome 18 abnormality e.g. 18q deletion syndrome, 18p deletion syndrome, tetrasomy 18p, ring 18, trisomy 18, and other unique rearrangements of chromosome 18

Mission:

To help individuals with chromosome 18 abnormalities overcome the challenges they face, so they might lead happy, healthy and productive lives by providing information, family support and supporting research

Services:

- Newsletter
- meetings
- parent network
- listserve (worldwide email chat room)
- information

The Registry

The Chromosome 18 Registry & Research Society is a lay advocacy organization composed primarily of the parents of individuals with one of the chromosome 18 abnormalities. We are also proud to count among our members affected individuals, extended family members and professionals. Membership is open to any interested person. We are a 501(c)(3) non-profit, tax-exempt public charity.

Our work is supported by memberships and donations from individuals and charitable organizations. We have met the strict criteria for fiscal responsibility set by the Combined Federal Campaign.

Conditions Involving Chromosome 18

There are five major conditions involving large changes of chromosome 18. Each of these conditions has a wide variety of characteristics. Additionally, each of the conditions can vary in severity.

The website provides information about each of the conditions and

discuss the genetic basis as well as the features of each of these conditions.

Although every child with a chromosome change is different, the online information provide a general idea of the medical and developmental concerns that are associated with the conditions.

Some individuals have chromosome changes involving chromosome 18 that do not fit neatly into one of these five conditions. For example, individuals with an unbalanced translocation may have a change in another chromosome in addition to the change on chromosome 18. Therefore, information may not be completely specific to all chromosome changes involving chromosome 18.

However, the information provided in this section can act as a starting point for information about the effects of the chromosome change.

What is chromosome 18?

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 18, one copy inherited from each parent, form one of the pairs. Chromosome 18 spans about 76 million DNA building blocks (base pairs) and represents approximately 2.7 percent of the total DNA in cells.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the

number of genes on each chromosome, the estimated number of genes varies. Chromosome 18 likely contains between 300 and 500 genes. These genes perform a variety of different roles in the body.

Genes on chromosome 18 are among the estimated 20,000 to 25,000 total genes in the human genome (Genetics Home Reference. <http://ghr.nlm.nih.gov/chromosome/18>)



Address and Contact Details:

61 Kuroki Street
PENSHURST NSW 2222

Phone : (02) 9580 5707

Website: www.chromosome18.org

E: chromosome18@optusnet.com.au

E: office@chromosome18.org

Operating Hours:

Contact as necessary

Information Source:
www.chromosome18.org



Coming Events

National Diabetes Week

Diabetes WA

Date: 10 - 16 July



A week to raise awareness about diabetes and its prevention.

Contact: 9325 7699

Website: www.diabeteswa.com.au

National Science Week

Department of Education

Date: 13-21st Aug 2011

Get ready for Science Week... There's tonnes of ways to get involved - for more information. Visit: www.scienceweek.info.au

Contact

Email: scienceweek@dest.gov.au

Website: www.scienceweek.gov.au

WA Self Directed Services Conference Planned for August 2011

The Department of Premier and Cabinet is aiming to progress the implementation of self-directed services in WA by holding a Self Directed Services conference in August. The Department recently met with peak community agencies and the Disability Services Commission

to discuss the timing and agenda for the conference. Among the issues discussed were the purpose of the forum, the target audiences as well as the date, possible venues and possible speakers and presenters.

The idea of a DVD which would feature the stories of service users who have experience with self-direction was also raised. The DVD would accompany the release of the concept paper in the lead up to the conference. A firm conference agenda will be fleshed out in coming months.

The move toward self-directed service design and delivery was recommended by the Economic Audit Committee (EAC) as a way of 'putting the public first'. The EAC envisaged the progressive implementation of the pilots of self-directed service design and delivery for individuals and communities in areas such as mental health, aged care, and homelessness as well as regional and remote communities and social housing.

- More information about self-directed services and the Economic Audit Committee agenda can be found on the Department of Premier and Cabinet website: <http://www.dpc.wa.gov.au/Publications/EconomicAuditReport/Pages/SelfDirectedServiceDesign.aspx>
- More about individualised support and funding in mental health is available on the Mental Health Commission website: www.mentalhealth.wa.gov.au

Patient Information session with Dr Carla Hollak

Fabry Support Group Australia



Date: 23 July 2011
Observatory Hotel
89-113 Kent St Sydney NSW

Fabry Support Group Australia (FSGA) in conjunction with Genzyme would like to invite you to a Patient Information session with Dr Carla Hollak. Dr Carla Hollak is an International Fabry Expert from The Netherlands.

RSVP BY FRIDAY 1ST JULY this will ensure the session will proceed and will assist with catering.

If you have any questions regarding this event please do not hesitate to contact:
Megan Fookes: 02 9967 4395 Lea Chant: 02 9772 1346 or Email: president@fabry.com.au

Travel Grants are available to FSGA Members, please contact us for details
<http://www.fabry.com.au>

Familial Hypercholesterolemia Patient Information Night

Date: 27th July 2011
Time: 6.15pm - 8pm



Location: Oasis Lotteries
House
37 Hampden Rd NEDLANDS WA 6009

- Free Entry
- Food & Refreshments Provided
- Question & Answer Time

Want to learn more about your inherited high cholesterol? Do you have questions? Do you want to hear about the latest treatments, living with a chronic condition, diet...?

If you are living with Familial Hypercholesterolemia (FH) this event is for you.

Speakers

Professor Gerald Watts
DSc MD PhD FRCP FRACP

“Latest Treatments”

Annette Wong
Accredited Practising Dietician

**“Eating with a cardiovascular disease:
Ways to reduce saturated fats, cholesterol rich
foods and improve your lipid profile”**

Rosemary French
Clinical Psychologist

“Living well with a chronic medical condition”

RSVP 15th July
Contact: 9485 8999
Web: www.fhfamilysupportgroup.websyte.com.au

ADHD Seminar

Communicating with & Teaching 'Tough Kids'

Date: Friday 16th September 2011

This event is primarily catering for professionals

Managing Tricky Behaviours at Home

Date: Saturday 17th September 2011

An event for parents: The presenter is an engaging speaker with practical experience in the area of ADHD & associated disorders, having recently won an international award for a published book on the topic.



Contact: 9346 7544
(M, W, F mornings)
Website: <http://www.ladswa.com.au>



Promote your event here and on our website at no cost! Contact us on

(08) 9389 6722 or e-mail
info@geneticsupportcouncil.org.au



Resources

NEW Young Carers WA Website!

Carers WA recently launched our Young Carers WA website! Visit the website at <http://www.youngcarerswa.asn.au>. The site provides information and advice for young people who care for a family member with a disability, mental or chronic illness or is frail aged.

Training group leaders how to include people with chronic disease in community activities

Abbott S, Vassallo A et al. (2010). Centre for Clinical Governance Research in Health, University of New South Wales and the Centre for Health Stewardship, Australian National University, College of Medicine Biology and Environment.

The purpose of this package is to support improvement of the inclusion of people with chronic disease in community activities in a local area. It contains information and resources to help plan, deliver and evaluate educational activities within a local community.

The first section details the aim, rationale and background for the development of this package. The overall aim of this package is to educate community group leaders about chronic disease issues. Community leaders equipped with such knowledge will be better able to support people with chronic

disease to manage their conditions while encouraging their participation in community group activities.

See: <http://eview.anu.edu.au/chronic/toolkit/index.php>

Stem cell legislation and research in Australia

An overview and briefing from the Australian Science Media Centre.

Visit: http://www.stemcellcentre.edu.au/NewsEvents/News/SC_legislation_research_in_Australia.aspx

Are you a GP, practice manager, specialist or allied health professional?

You can now register to receive Forum online. Register now to get your quarterly update of news and information from Medicare Australia delivered directly to your inbox.

Visit: <http://www.medicareaustralia.gov.au/provider/pubs/news/forum/subscribe.jsp>

Stay Smart Online

The Australian Government's cyber security website provides information for Australian internet users on the simple steps they can take to protect their personal and financial information online.

Visit: <http://www.staysmartonline.gov.au>

Telephone Allowance

Telephone Allowance helps with the costs of maintaining a telephone and home internet service.

There are two rates of Telephone Allowance, depending on your circumstances. Your eligibility for Telephone Allowance is assessed when you make a claim for a Centrelink payment.

Visit: http://www.centrelink.gov.au/internet/internet.nsf/payments/telephone_allow.htm

The Genomic Resource Centre

The Genomic Resource Centre (GRC) has been developed by World Health Organization (WHO) Human Genetics to provide information and to raise awareness on human genetics and more recently human genomics, a new and rapidly developing science.

This site provides an overview of the issues and concerns around human genetics and public health and, in particular, addresses the ethical, legal and social implications (ELSI) of genomics.

Visit: <http://www.who.int/genomics/aboutgrc/en/index.html>

Understanding genetic tests for breast and ovarian cancer that runs in the family - Information and decision aid

This booklet contains information for women who are considering genetic testing for breast and ovarian cancer that runs in their family. This information is not a replacement for discussing genetic testing with your

doctor or family cancer services. Choose the sections that are relevant to you, coupled with information from your health professionals, to make an informed decision that is right for you.

Visit: Centre for Genetics Education, NSW.
<http://www.genetics.edu.au/pdf/brovca-genetic-testing.pdf>

Orphanet is Creating Fact Sheets for Emergency Situations

Orphanet (The portal for rare diseases and orphan drugs) is creating a collection of fact sheets and emergency guidelines for healthcare professionals. These may, for example, be carried by patients to the ER in emergency situations. Ultimately, the collection will cover numerous diseases and will be available in several languages.

Topics available to date may be viewed on the Orphanet website: http://www.orpha.net/consor/cgi-bin/Disease_Emergency.php?lng=EN

National Health and Hospitals Network Bill 2011

The Parliamentary Library released a Bills Digest examining the legislation to establish the Australian Commission on Safety and Quality in Health Care as a Commonwealth statutory body.

Visit: <http://www.aph.gov.au/library/pubs/bd/2010-11/11bd046.pdf>



Information accessed through the World Wide Web is of varying levels of quality and accuracy. The material supplied is for information purposes only & is not to be used for diagnosis or treatment.



Grants

Do I need an authority to fundraise?

Yes, if you are any organisation that collects money or goods from the public for:

- the relief of the diseased, sick, infirm, incurable, poor, destitute, helpless or unemployed persons or their dependants;
- the relief of distress occasioned by war and the support of persons who have been members of the armed forces;
- the support of hospitals, infant health centres, schools, kindergartens and other activities of a social or welfare character; or
- any benevolent philanthropic or patriotic purpose.

You do not need one if you are the following:

- An organisation that collects and receives money from the public for non charitable purposes, such as for sporting clubs, conservation, environmental, animal welfare and schools or kindergartens that operate their own fund raising events.
- Any organisation that raises money for charitable purposes but does not collect from the public.

Note that if you are raising funds through government and Lotteries Commission grants, membership subscriptions, gaming machines,

raffles etc, these activities do not constitute fundraising for which a licence is required. There is no fee required for a license.

How do I apply for an authority to fundraise?

To obtain a licence you will need to lodge with the Administrative Officer, Charitable Collections Advisory Committee.

For further information:

Administrative Officer, Charitable Collections Advisory Committee, Department of Commerce, 219 St Georges Tce, Perth WA

Telephone: +61 8 9282 0832

Facsimile: +61 8 9282 0856

Email: consumer@commerce.wa.gov.au

Webpage: www.commerce.wa.gov.au/ConsumerProtection/Content/Business/Charities/index.htm

Different licences are required for other fundraising activities.

It is a requirement for all charitable organisations that collect monies and goods from the public to be licensed with the Charitable Collections Advisory Committee under the Charitable Collections Act 1946.

How to become a Licensed charity

To become licensed you need to complete an application to licence a charitable organisation. (It is a requirement for all charitable organisations that collect monies and goods from the public to be licensed

with the Charitable Collections Advisory Committee under the Charitable Collections Act 1946.

http://www.commerce.wa.gov.au/ConsumerProtection/Content/Business/Charities/How_to_become_a_charity/How_to_become_a_charity.htm

Source: Our Community Pty Ltd. [Accessed 13/05/2011] http://www.ourcommunity.com.au/funding/funding_article.jsp?articleid=48

Our Community Fundraising Formula

A good fundraising plan rests on six pillars. These are: Donations; Grants; Community-Business Partnerships; Membership/Alumni/Friends; Special Events; and Earned Income.

The workings of these six pillars are described in more detail in the help sheets below, or for a more comprehensive account of how to apply each pillar in your own group, refer to the Our Community publication, The Complete Community Fundraising Handbook available:

http://www.ourcommunity.com.au/marketplace/marketplace_article.jsp?articleId=2896

Google Grants

The Google Grants program empowers non-profit organisations to achieve their goals by helping them promote their websites via advertising on Google. Google AdWords ads appear when users search on Google. When you click one of the ads, you are brought to the website being advertised.

In-kind advertising for non-profit organisations

Google Grants is a unique in-kind donation program awarding free AdWords advertising to selected charitable organisations. We support organisations sharing our philosophy of community service to help the world in areas such as science and technology, education, global public health, the environment, youth advocacy and the arts.

Further Information

Website: <http://www.google.com.au/grants/index.html>

Program Guidelines Eligibility and Details: <http://www.google.com.au/grants/details.html>

DonorTec

DonorTec provides donated and discounted technology products and services from companies such as Microsoft, Symantec, Sophos, Kaspersky and Cisco to eligible income tax exempt (ITE) Australian nonprofit groups.

Via this program nonprofits can access the latest technology via donations or generous discounts.

<http://www.donortec.org>



GSCWA can assist with grant applications and resources for your group!



Link Line

Genetic support groups are an important resource for families or people in a similar situation. The Link Line provides a supportive and confidential means of connecting individuals and families for whom no known genetic support group exists.

If any individual is seeking contact with others in these circumstances, The Link Line is available to you for this purpose.

To date, there appears to be no specific support group for the following conditions/syndromes'.

Monomelic Amyotrophy

A person from Western Australia who is living with monomelic amyotrophy would like to make contact with others living with this condition.

Synonyms: of Monomelic Amyotrophy: Benign Focal Amyotrophy, Hirayama syndrome, O'Sullivan-McLeod syndrome, Sobue disease and Single limb atrophy.

If you would like to contact with one of these families or individuals, please call Kristina at the Genetic Support Council for further details on 08 9485 8999 or email: info@geneticsupportcouncil.org.au

New Perth Cancer Support Group

Programme Overview:

The topics to be covered in the group include:

- Coping with cancer diagnosis and treatment.
- Dealing with your emotions and the emotions reactions of your family and friends.
- The principles of mediations and imagery.
- The role of diet and nutrition to prevent and help manage illness.
- Topics suggested by group members.

Format:

The group will be ongoing and meet weekly

When & Where

The group commences Friday 6th May.
335-337 Pier St PERTH (opposite Members Equity Stadium)

Please Contact:

Marie Smith SJG

Counsellor

Telephone (08) 9383 9930



Genetic Support Council WA

The Genetic Support Council WA Inc (GSCWA) is a not for profit organisation that acts as a peak body for genetic support groups in Western Australia. The Council provides information to individuals and families about genetic conditions including information on the services provided by Genetic Support Groups.

Genetic support groups can be a useful resource for individuals and families affected by a genetic condition. Support Groups can provide information about the condition and community resources, as well as an understanding and empathetic ear.

In Australasia, there are hundreds of such groups each focussing on a specific genetic condition. We can help put you in contact with these groups or provide the support and guidance to help you establish a new support group.

The Council may also be able to provide information and support for conditions so rare that there is no specific local support group.

The GSCWA maintains a database of Genetic Support Groups throughout Australia and with international links can provide up to date information on genetic conditions – including the latest information on research developments.

Information is also readily available on a range of associated topics - online resources, living with a family member with a genetic condition, parenting, and education in association with a genetic condition are some of these.

The Council conducts forums for members on issues of interest or concern to enable member's views to be represented to the wider community and State and Federal Governments. We also provide written and verbal submissions to Government and others in the field of genetics and participate with like bodies nationally to further represent Genetic Support Groups and their members.

We conduct forums and awareness events to enable participants to increase their skills and knowledge and access up to date information about resources for people affected by genetic conditions and their families. The Council maintains an up-to-date website and publishes a bi-monthly newsletter and information sheets for consumers.

The GSCWA actively supports the ongoing development of Genetic Support Groups. We are able to assist with the development of resources such as information packages, pamphlets and the printing of promotional material. Groups can also publish articles and advertise and promote awareness of their Support Group's activities on the GSC website and in our bi-monthly newsletter the "Gene Advocate".

Staff can assist groups and individuals in researching information regarding particular genetic conditions or predispositions and help keep members aware of matters of interest such as changes of legislation etc.

All GSCWA services are free to member groups and individuals. Please call us for further information or to access any of these services.

What causes genetic conditions?

There are three types of genetic conditions:

- Heritable conditions: Due to a mutation in a single gene
- Chromosomal conditions: Occurs when an individual is affected by a change in the number, size or structure of his or her chromosomes
- Multifactorial conditions: Due to the interaction of the genetic information and environmental factors such as diet, chemical exposure and lifestyle.

What can be done about Genetic Conditions?

Prevention

Some people are more at risk than others for developing a condition that is due to the interaction of environmental factors with their inherited genetic information. They are "genetically predisposed" to develop these conditions. However, the presence of an environmental "trigger" is necessary for the person to be affected with the condition. In some cases, prevention of the condition can be achieved by the person avoiding being exposed to the particular environmental factor that will trigger the condition.

For example, it is possible to prevent about 70% of the cases of spina bifida (a neural tube defect) in babies if women who are more likely to have a baby affected with spina bifida, because they are genetically "predisposed", take the vitamin folic acid before, and continue it during early pregnancy.

Early Diagnosis and Treatment

In some genetic conditions, early diagnosis, sometimes even before the symptoms appear, can lead to specific treatment. For example, all newborn babies in Australasia are screened for phenylketonuria (PKU) by a simple blood test.

Diagnosis and treatment within the first month of life are crucial to avoid intellectual disability.

Also, some cancers which have a genetic component, e.g. breast cancer, bowel cancer and melanoma, can be detected early enough to enable treatment to take place.

Checking the family health history may determine if a person or another blood relative are at risk for developing a genetic condition or for passing it on to their children.

Genetic Counselling

Genetic counselling is available to families and individuals that have concerns about a condition in their family which may have a genetic basis. A team of health professionals which may include clinical geneticists, genetic counsellors and social workers, work together to provide information and supportive counselling so that families may be better able to understand, and adjust to, the diagnosis of a genetic condition.

Genetic testing, if it is available and appropriate, can also be organised on the basis of informed consent. Genetics Services are available throughout Australasia and provide genetic counselling to assist in informed decision making regarding genetic testing.

Support Groups

Support groups provide affected individuals and families with information about the condition and community resources, as well as an understanding and empathic ear. There are over 200 genetic conditions for which there are over support groups/branches in Australasia.

Some points to remember when contacting a group

Many of the support groups contact numbers are home numbers, so please be considerate of the hours at which you call.

Many support groups do not receive funding and rely on their group members for income. Offering to pay for postage, photocopying and/or materials provided will be appreciated by many groups.



GSC Members

Full Members

Acoustic Neuroma Association of Australia, WA Branch
Alzheimer's Association of WA
Angelman Syndrome Association
Arthritis Foundation of WA
Arthrogyrosis Support Group
Australian Cystinosis Support group
Australian Dyspraxia Association
Australian Leukodystrophy Support Group
Australian Mitochondrial Disorder Foundation
Australian Pituitary Foundation WA Branch
Australian Tuberos Sclerosis Society Inc.
Cardio Facio-Cutaneous Syndrome Support
Charcot-Marie-Tooth Association Australia
CHARGE Syndrome Association of Australasia
Chiari & Syringomyelia Australia
CleftPALS WA
Coeliac Society of WA, Inc
Cornelia De Lange Syndrome Support Group
Cushing's Disease Support Group
Cystic Fibrosis WA
Diabetes Western Australia
Down Syndrome Association of WA Inc
Dyslexia SPELD Foundation WA Inc
Epilepsy Association of WA
Even-Keel Bi-Polar Support Association (Inc)
Fabry Support Group of Australia
Familial Hypercholesterolaemia Support Group WA
Fragile X Support Group WA Inc
Haemophilia Foundation WA Inc.
Heart Kids WA
Huntington's WA Inc
Klinefelters Support Group
Learning and Attention Disorders Society of WA (LADS)
LQTS Support Group WA
Lupus Group of WA
Lymphoedema Association of WA
Menieres Support Group WA
Mental Illness Fellowship WA
Mitochondrial Disease Foundation
Motor Neurone Disease Association of WA
Mucopolysaccharide & Related Diseases Society (MPS)
Muscular Dystrophy Association of WA
Neurofibromatosis Association of WA.
Parents of Children with Disabilities
Periodic Paralysis Society of Australia
PXE Support Group of WA
Retina Australia (WA)

Rett Syndrome Association of WA
Senses Foundation Inc.
Short Statured People's Association WA Branch
SIDS and Kids Western Australia
Spina Bifida Association of WA
Thalassaemia Association of WA
Tourette Syndrome Support Group, Perth
Turner Syndrome Association of Australia (WA Branch)
Support Organisation for Trisomy and Related Disorders of WA (SOFTWA)
Usher Syndrome Support Group

Corporate Associate Members

ARAFMI Western Australia
Androgen Insensitivity Syndrome Support Group Australia
Association of Genetic Support of Australasia
Australian Kidney Foundation
Carers Association of WA Inc
ConnectGroups
Ectodermal Dysplasia Support Group - OzED
Genetic Support Network of Victoria
Health Consumers Council WA
Kidney Health Australia
Learning Centre Link—Linkwest
Myasthenia Gravis WA Friends and Support Group
NZ Organisation for Rare Disorders
Office of Population Health Genomics
People with Disabilities WA
Self Help Organisations United Together (SHOUT)
Speak Easy Association Western Australia
The Chromosome18 Registry & Research Society
The Kalparrin Centre
The Neurological Council of WA Inc
Unique in Australia Chromosome Disorder Support Group
Western Australian Deaf Society Inc.

Individual Associate Members

Linda Bovill	Robyn Hendriks
Anja Hermann	Terry Keating
Sindhu Kurup	Amanda Samanek
Kristina Sengotta	Abdulla Sheikhi
Darren Webb	
Professor Charles Watson	

Membership Forms are available on the Web.
<http://geneticsupportcouncil.org.au>

Attach mailing
address label here

Contact Us



Genetic Support Council WA Inc. (GSCWA)

Address: Oasis Lotteries House
Suite 7, 37 Hampden Road
Nedlands WA 6009

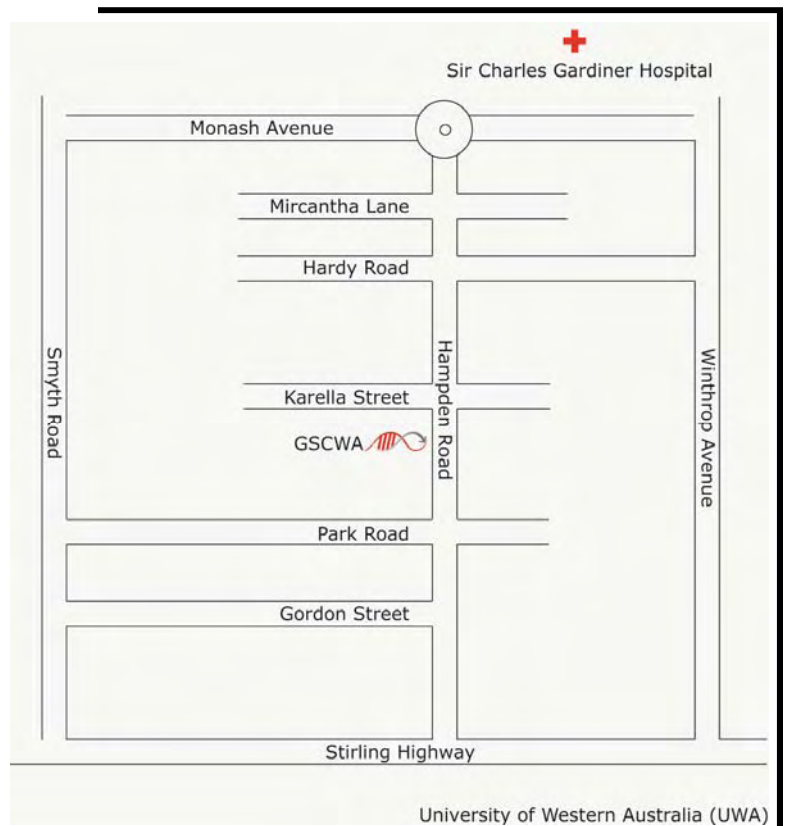
Phone: (08) 9485 8999

Email: info@geneticsupportcouncil.org.au

Web: www.geneticsupportcouncil.org.au

Office hours: 9am to 4pm Monday to Friday.

Staff: Sharon Van der Laan, Executive Director
Kristina Sengotta, Resource Officer
Joan Ryder, Book Keeper



▶▶ The views and opinions expressed in this newsletter are those of the individual authors and not necessarily those of the GSCWA. The material supplied is for information purposes only, and is not to be used for diagnosis or treatment.